Tasmanian Rare Disease Diagnostic Pathways

Improving Diagnostic Pathways for Rare Diseases in Regional Australia

Final project report

June 2024

Contents

List of Abbreviations

Abbreviation	
CPW	Clinical pathways
CRG	Community reference group
DoHAC	Department of Health and Aged Care (Cwth)
ERN	European Reference Networks
EU	European Union
GP	General practitioner
GSNV	Genetic Support Network Victoria
HETI	Health Education and Training Institute
HREC	Human Research Ethics Committee
MBS	Medicare Benefits Schedule
MDT	Multidisciplinary team
MoC	Model(s) of care
NDIS	National Disability Insurance Scheme
NHMRC	National Health and Medical Research Council
NBS	Newborn screening
OCP	Optimal care pathways
PHT	Primary Health Tasmania
PTAS	Patient Travel Assistance Scheme
RD	Rare disease(s)
RVA	Rare Voices Australia
SES	Socioeconomic status
TCGS	The Tasmanian Clinical Genetics Service
TRUDN	Tasmanian Rare and Undiagnosed Disease Network
UDN	Undiagnosed Diseases Network
UDP	Undiagnosed disease program(s)
UDN-Aus	Undiagnosed Diseases Network Australia
UDP-Tas	Undiagnosed Disease Program - Tasmania
UDP-WA	Undiagnosed Diseases Program - Western Australia
UN	The United Nations
VCGS	Victorian Clinical Genetics Services
WA	Western Australia

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This project was conducted by the Tasmanian Clinical Genetics Service (TCGS), within the Tasmanian Department of Health, with a $250,000 grant under the Australian Genomics 'Genomic Implementation Projects 2022 Expression of Interest' process for projects that address areas of strategic importance or identified areas of unmet need in the Australian genomic system, as part of the National Health and Medical Research Council grant to Australian Genomics for 2021-23 (GNT2000001).

This report was prepared in collaboration with Abt Associates.

Executive Summary

Project background

In Australia, a disease is considered rare if it affects fewer than 5 in 10,000 people.[1-3] Rare diseases are a significant public health concern. While individually rare, these diseases are collectively common, and can often be life-threatening or chronically debilitating. Rare diseases affect approximately 8% of Australians, including an estimated 35,000 to 45,000 Tasmanians.[1] An undiagnosed disease refers to a medical condition for which the health system has been unable to provide a diagnosis; a diagnosis includes an understanding of disease pathogenesis, linking genetic and clinical findings, and informs prognosis and therapy. Rare diseases can be difficult to diagnose, and while there is a spectrum of rare disease types (including undiagnosed cancers, infections, and autoimmune disorders), most (80%) have a genetic cause.[1, 4] The diagnostic journey may therefore require genetic testing and the expertise of clinical geneticists and genetic counsellors.

Rare diseases cannot generally be prevented or cured, but a diagnosis can provide an individual with access to the most appropriate care. This can be achieved through rare disease diagnostic pathways that set out a clear process for clinicians to follow to obtain a diagnosis and guidance for post-diagnostic care.

The Tasmanian Clinical Genetics Service (TCGS) received funding to investigate how to improve diagnostic pathways for rare diseases in regional Australia. Focusing on Tasmania, the project included a community needs assessment, followed by the co-development of innovative regional service models.

Methodology

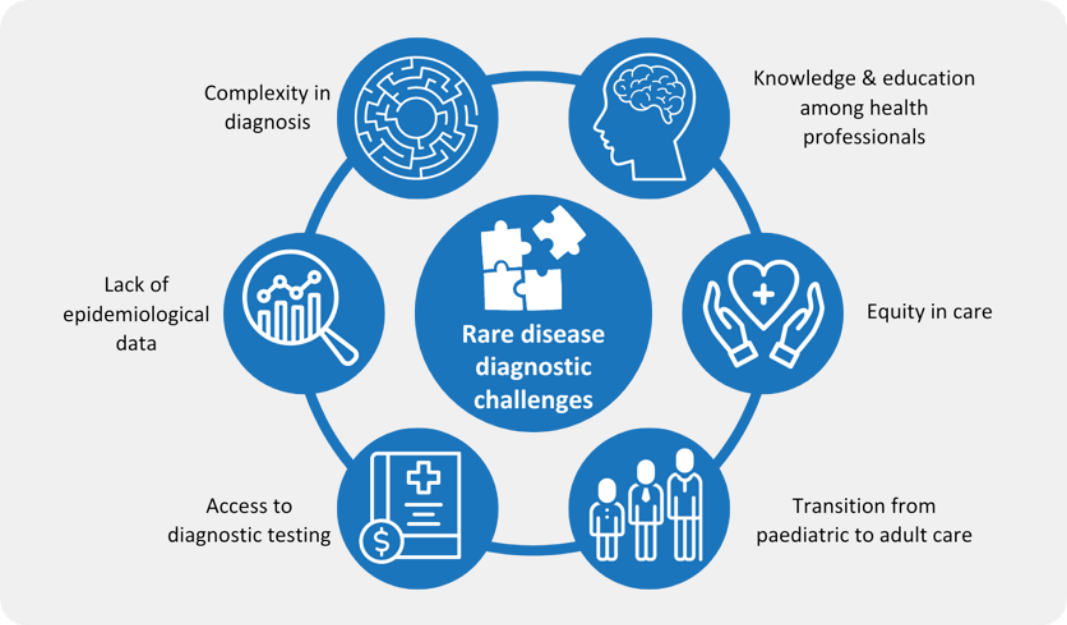
Mixed methods were used to gain a detailed understanding of the barriers to rare disease diagnosis in Tasmania and to identify targeted solutions and strategies to overcome these barriers. The 4 main stages of the project were: (1) Project Planning and Design; (2) Situation Analysis (literature review and key informant interviews); (3) Community Needs Assessment; and (4) Developing Models of Care and Recommendations.

The research design was collaborative, developed with guidance from the TCGS Community Reference Group and clincians and researchers with knowledge of the Tasmanian health system and rare disease. The research process was iterative, with the findings from each stage helping to determine the conduct of the following stage.

Situation analysis

Six key challenges in rare disease diagnosis were identified through a review of the literature (Box 1 below) and these were reaffirmed by key informant interviews with clinicians, researchers and administrators (n=6). There were also disease-specific issues expressed by some of the specialists interviewed, which could inform the development of disease-specific care pathways.

Box 1: Key challenges in rare disease diagnosis



A rare disease impact report illustrated that it takes on average 5.6 years to obtain a diagnosis for a rare disease in the UK, and 7.6 years in the USA.[56] This 'diagnostic odyssey' has been recorded around the globe, including in Australia, with a lack of diagnosis being a major barrier to accessing care.[5]

With reference to Tasmania specifically, the table below provides a summary of the main challenges as found in the literature and reported during key informant interviews during situation analysis.

Summary of identified barriers to rare disease diagnosis in Tasmania

Patient level	Primary care level	System level	Related to genetic testing
Inequity of access due to geographical, educational or another socio-economic disadvantage	Lack of suspicion/ consideration of a rare disease	Limited ability to establish multi-disciplinary teams due to the limited availability of specialists	Funding of genetic testing
Unclear diagnostic process and fear/ confusion about genetic testing	GPs feeling overwhelmed due to limited knowledge and understanding	Disproportionate workforce distribution impacting regional and remote areas	Lack of genetics and genomics skills across the workforce
Isolation and stress felt by families due to the rare disease diagnostic odyssey	Lack of knowledge of easily available and accessible information for GPs	Issues with recruitment and retention of health professionals critical in caring for rare disease	Difficulty in accessing diagnostic testing due to costs, limited availability, lack of awareness amongst healthcare professionals and/or need to take time away from employment for carers
Navigation of a complex health system	Unclear diagnostic process	Specialist shortages	
		Transition from paediatrics to adult health care system	
		Lack of epidemiological data to identify gaps in knowledge and guide resource allocation	

Community needs assessment

Surveys, interviews and focus groups were used to engage the various stakeholder groups to provide their experience of the rare disease diagnostic care pathway in Tasmania. A total of 1014 stakeholder consultations were conducted. A summary of the community needs assessment is provided in the tables below.

Method of Engagement	Participation
1. Surveys	• 880 patient/carer survey responses • 31 clinician/researcher/advocacy organisation survey responses
2. Interviews and focus groups post surveys	• 15 clinician/research interviews • 11 individual patient/carer interviews • 63 patient/carer focus group attendees • 5 individual Community Reference Group member interviews • 9 individual advocacy and patient group interviews.

What We Heard

What makes diagnosis challenging?	What helps diagnosis go well?
• GP shortages (especially North West, rural areas) & limited capacity • Limited knowledge/understanding of rare diseases among doctors (noting no expectation from patients/carers that doctors can know everything) • Lack of listening & empathy from many doctors • Limited access to local specialists & diagnostic tools • Long wait times for specialists (e.g., neurology, paediatrics & rheumatology) • Impact of interstate travel (financial, work, family/friends); some having to relocate • Moving from child-specific to adult health services • Lack of support to navigate & understand service system, including genetics service • Poor communication & co-ordination from/between care providers • Financial barriers (out-of-pocket expenses)	• Strong trust with a consistent GP • GP has experience with rare diseases • GP listens and is committed to finding a diagnosis • Multidisciplinary team approach • Caring allied health professionals • Emotional/mental health support from family/friends, others with rare diseases • Practical support (e.g., transport, PTAS) • Patient/carer health literacy and commitment to finding a diagnosis • Coordinated support to navigate and access health system and associated supports

What Patients and Carers Said They Need

- **Health professionals who**
  - listen
  - consider all symptoms, whole context, family history
  - research and explore the condition
  - investigate and refer for testing/specialist review as needed
  - communicate well and regularly
  - communicate and coordinate with other health professionals.
- **Less time** between testing and diagnosis
- **More information** about rare diseases
- **More support** for rare diseases through patient networks/groups
- **More assistance** to access health services and social/disability supports, including access to NDIS
  - e.g., advocacy, referrals, support networks within Tasmania.

Models of Care

Models of care outline how health services should be delivered. The 'National Strategic Action Plan for Rare Diseases' prioritises the development of models of care through priority action 2.1.1.1:[2]

> Establish standards for care and support that are integrated and incorporate clear pathways throughout all systems. Ensure these are informed by clinical and consumer rare disease experts and that such consultation informs policy development.

To address the contextual nuances in Tasmania, the key components of models of care for people with rare diseases in Tasmania have been developed, detailing the context, the current Tasmanian situation, and options for consideration. Seven components were identified that capture health system touchpoints, and reflect a continuum of care that echoes the patient journey:

1. Accessing information
2. Accessing the health system
3. Knowledge and attitudes of cinicians
4. Coordination and integration of care
5. Timely and appropriate investigations
6. Accessing research and clinical trials
7. Accessing support

The detailed models of care can be found in Section 6 of this report.

Recommendations

The options for consideration under each component of the models of care were summarised into a series of recommendations designed to achieve implementation of the models of care in the Tasmanian health system. The recommendations are based on the stakeholder consultations (survey, interviews and focus groups), research undertaken in relation to best practice diagnosis and management of rare diseases and a review of the current pathways in Tasmania. Draft recommendations were reviewed by the Tasmanian Clinical Genetics Service (TCGS) Community Reference Group as well as Tasmanian Rare and Undiagnosed Disease Network (TRUDN) members, and all feedback was incorporated into the final recommendations.

A recurring recommendation provided by all stakeholders, and supported by evidence from the literature, was the establishment of a Tasmanian Rare Care Centre. This centre would be a single point of care for people affected by a rare disease, ensuring comprehensive, integrated care delivered by personnel with expertise in the diagnosis, management and support of rare diseases.

A summary of the recommendations based on each component of the models of care is provided in the table below. Please note this is a research project report only and the Tasmanian Department of Health will consider these recommendations in the planning and delivery of associated services.

Recommendations to improve models of care for rare disease diagnosis in Tasmania

Components of Best Practice Diagnostic Care for Rare Disease						
Accessing Information	**Accessing the Health System**	**Knowledge & Attitudes of Clinicians**	**Coordination & Integration of Care**	**Timely & Appropriate Investigations**	**Accessing Research & Clinical Trials**	**Accessing Support**
What does it look like?						
Information is readily available on the underlying causes, mechanisms and clinical presentations of rare diseases for both health professionals and patients and their families and carers.	Patients have access to timely primary and specialist health care when needed and are supported in navigating the system.	Health professionals are well informed about rare diseases and available diagnostic and referral pathways. Health professionals listen to their patients and take proactive steps to seek a diagnosis.	Patients have access to holistic, coordinated, multidisciplinary care.	Patients have access to timely and affordable diagnostic testing and investigations recommended by appropriate clinical advice and available to all relevant treating clinicians.	Patients can access research relevant to their condition, supported by local clinicians.	Patients and their families and carers can access emotional and mental health support and practical logistical supports during their diagnostic journey and in ongoing care.

Components of Best Practice Diagnostic Care for Rare Disease						
Accessing Information	**Accessing the Health System**	**Knowledge & Attitudes of Clinicians**	**Coordination & Integration of Care**	**Timely & Appropriate Investigations**	**Accessing Research & Clinical Trials**	**Accessing Support**
How can we achieve this in Tasmania?						
Establish a Tasmanian Rare Disease Support Group. Promote existing information resources to both patients and clinicians. Establish a Tasmanian Rare Care Centre.	Recruit and retain more clinicians. Improve access to clinicians in rural and regional areas via telehealth. Establish care coordination for patients with rare diseases, including help accessing associated supports. Facilitate access to rare disease specialists in other jurisdictions.	Conduct training and awareness raising to GPs on rare disease and public clinical genetics services. Develop and promote resources to guide clinicians in identifying and seeking diagnosis of rare diseases. Develop rare disease referral pathways for inclusion in GP HealthPathways portal.	Establish a Tasmanian Rare Disease Multi-Disciplinary Diagnostic Clinic. Establish a Tasmanian Rare Care Centre. Promote use of electronic records systems.	Improve local workforce and laboratory capacity to undertake genetic and genomic analysis. Ensure electronic records systems are used to safely document and share patient records.	Establish a Tasmanian Rare Disease Registry. Provide ongoing support for translational research participation in relation to rare diseases in the Tasmanian Health Service.	Establish a Tasmanian Rare Disease Support Group.
What needs to happen on a national level?						
Establish a national platform or program to encourage information sharing.	Increase number of training places for GPs and specialists across Australia.	Improve education on rare diseases and genetics provided to medical undergraduates and in specialist training.	Work with primary care software vendors to improve rare disease patient identification and management.	Increase the range of genetic and other relevant diagnostic tests supported through the national public health system.	National funding for translational genomic research should specifically support the inclusion and participation of patients, researchers and clinicians in rural and regional Australia.	Existing support groups on mainland Australia could establish a presence in Tasmania, or establish expertise and services specifically for rural people.

1 Introduction

1.1 Project background

In 2022, the Tasmanian Clinical Genetics Service (TCGS) within the Tasmanian Department of Health received funding from Australian Genomics to review and develop new rare disease (RD) diagnostic pathways suitable for rural and regional areas, including a community needs assessment and co-developed innovative service models.

In Australia, a disease is considered rare if it affects fewer than 5 in 10,000 people.[1-3] RD are a significant public health concern. While individually rare, these diseases are collectively common, and can often be life-threatening or chronically debilitating. RD affect approximately 8% of Australians, including an estimated 30,000 to 40,000 Tasmanians.[1] An undiagnosed disease refers to a medical condition for which the health system has been unable to provide a diagnosis; a diagnosis includes an understanding of disease pathogenesis, linking genetic and clinical findings, and informs prognosis and therapy. RD are very often undiagnosed, and while there is a spectrum of RD types (including undiagnosed cancers, infections, and autoimmune disorders), most (80%) have a genetic cause.[1, 4] The diagnostic journey may therefore require genetic testing and the expertise of clinical geneticists and genetic counsellors.

The Australian Government Department of Health commissioned the development of the 'National Strategic Action Plan for Rare Diseases', published in 2020.[2] The 'Action Plan' aims to improve health and wellbeing outcomes for priority populations, particularly those living in regional, rural and remote areas, and people experiencing socioeconomic disadvantage. All of Tasmania is considered regional or remote under the Australian Standard Geographic Classification, making undiagnosed RD patients in this area particularly vulnerable. Implications for this priority population with RD, as outlined by the Action Plan, include:

- a reduced likelihood of a timely and accurate diagnosis
- poor continuity of care and access to appropriate treatments
- an increased exposure to modifiable risk factors, such as smoking, high alcohol consumption, poor exercise levels and obesity, which are associated with an increased incidence of rare congenital anomalies.

RD cannot generally be prevented or cured, but individuals with RD need a specific diagnosis to access the most appropriate care. As a patient, knowing the diagnosis and any genetic variation related to the medical condition can help a patient to participate in research, connect with others in similar situations and know what therapies are safe to take. Alternatively, any delays in diagnosis may not allow for patients to get the maximum benefit from available treatments or may cause patients to miss a treatment window entirely and can slow collective ability of a health system to drive support, progress and cures.[6] The many benefits of a timely diagnosis have been well-documented in other studies and undiagnosed disease programs (UDPs). Benefits of a specific diagnosis include: certainty, enabling access to timely and best practice medical care; reduction in unnecessary investigations; reduction in inappropriate management/admissions; clarification of recurrence risks; availability of additional reproductive options; reduced isolation and access to peer support; and access to social and educational services.

Consistent with studies in other countries, however, diagnostic delays and misdiagnoses are common and unacceptable for patients with RD in Australia. Complex multi-system disorders require a diagnostic process that involves multiple medical specialties and a systematic method of excluding common conditions. It has been said that "families affected by rare diseases represent a medically disenfranchised population that falls through the cracks of every healthcare system in the world".[5]

The complexity of rare disorders and the need for multidisciplinary care has a significant impact on health providers and health budgets. This is where care pathways, also known as clinical pathways, can be helpful to systematically plan and follow up focused patient or client care. Care pathways are defined as "a complex intervention for mutual decision-making and organisation of care processes for a well-defined group of patients during a well-defined period".[7] The aim of a care pathway is to enhance the quality of care across the continuum by improving risk-adjusted patient outcomes, promoting patient safety, increasing patient satisfaction, and optimizing the use of resources. Models of care (MoC) pathways are a way of setting out a process of best practice to be followed in the diagnosis and treatment of a patient. Where possible, evidence-based care pathways need to be specific to the condition.

Australian Genomics has investigated perceived unmet needs in clinical genomics, showing a dependence on existing pathways for delivering healthcare.[8] These pathways are more established in populated areas and often siloed within speciality services. This dependency exacerbates the gap between access in metropolitan versus rural areas. This study recommended further analysis of unmet needs in rural and remote populations.[8]

1.2 Objectives

This project aimed to gain a detailed understanding of the barriers to RD diagnosis in Tasmania and to identify targeted solutions and strategies to overcome these barriers using a mixed-methods approach. Success of the project is measured by: a) the development of a comprehensive and up-to-date map of current care pathways and community need; and b) the development, through broad stakeholder engagement, of innovative MoC pathways and recommendations that are acceptable to the community, clinicians, researchers, and policymakers.

1.3 Relevant government policy, legislation, and rules

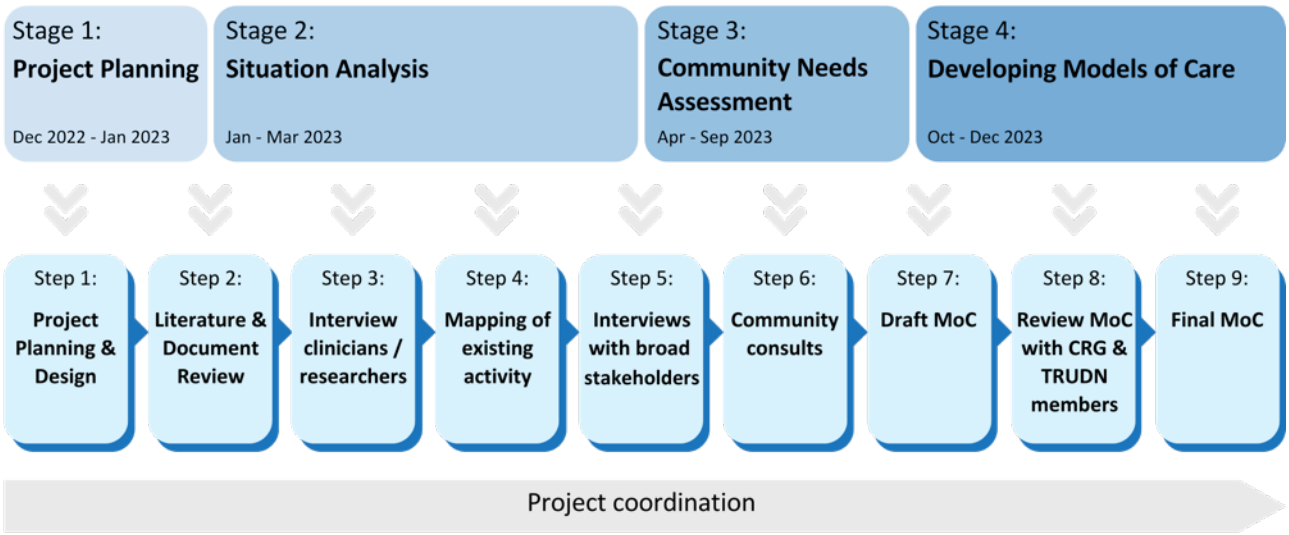
The project aligns with identified need in the Australian Government Department of Health's 'National Strategic Action Plan for Rare Diseases' [2], and broadly aligns with the Tasmanian Government's Strategic Priorities for 2021-23 'Reforming the delivery of care in our community', as a project that reviews subacute access to service delivery.

The project ensured alignment with the requirements of the NHMRC's policies and guidelines for human research, including requirements for working with Aboriginal and Torres Strait Islander peoples and vulnerable people. Ethics approval was granted by the University of Tasmania Human Research Ethics Committee (HREC) (HREC # H0027870).

2 Methodology

The project methodology included four main stages: (1) Project Planning and Design; (2) Situation Analysis; (3) Community Needs Assessment; and (4) Developing MoCs. Within these stages there were 8 key steps. Detailed methodology is provided below. Figure 1 gives an overview of the project.

Figure 1: Overview of Project Timeline



The project had a strong focus on i) community co-design and close stakeholder engagement, aiming to provide a greater understanding of community needs, ii) novel solutions to enable geographically equitable RD diagnostic and care services, and iii) greater trust and improved genomic literacy within the community.

2.1 Project planning and design

In the planning stage for this project key informant interviews and a preliminary document review were undertaken, and a project plan was developed and agreed upon.

Key informant interviews

Six key informant interviews with health system, clinical and research genomics expertise were conducted with two main purposes:

1. To further assist in ensuring the proposed project processes produced expected outcomes given the key informants' detailed knowledge of the project background, context and the key issues it is trying to address.
2. By engaging these key informants early in the project, to ensure the project processes were well designed and supported, for example, by other key colleagues/services, in identifying critical literature or documentation and in ensuring optimal participation in the consultations.
3. To inform the Situation Analysis under 2.2.

In addition, the Tasmanian Rare and Undiagnosed Disease Network (TRUDN) Clinical and Research Working Group, and its Community Reference Group (CRG), were consulted on the design of the research project and plans for stakeholder engagement. Through these consultations, a range of issues, challenges and opportunities for improvement were identified relating to timeliness of care, knowledge regarding RD diagnosis and access and availability of the right care at the right place (see Appendix 2 for a summary of the interviews).

2.2 Situation analysis

The aim of the situation analysis was to understand the RD diagnostic care pathways currently in place, nationally and internationally. It also sought to document current research findings on best practice diagnostic care and what clinicians and researchers think are the changes needed to achieve better diagnostic care for patients with RD. The situation analysis included two key processes: (i) a review of existing program documentation and relevant literature on RD diagnostic care pathways; and (ii) interviews with key informants.

The key question used for the literature review was 'What model of diagnostic care pathways needs to be implemented to enable earlier and improved diagnosis and management of rare and undiagnosed conditions?' The literature review also aimed to gather required knowledge and context for (i) issues and challenges in diagnosing these conditions; (ii) knowledge and awareness amongst affected patients and carers, (iii) issues in providing effective health care and (iv) key components of diagnostic care pathways in rare and undiagnosed conditions. The detailed literature review methodology is included in Appendix 1.

2.3 Community needs assessment

A range of methods were used to engage the various stakeholder groups to provide their experience of the RD diagnostic care pathway for Tasmania.

2.3.1 Community Reference Group interviews

The CRG established by the TCGS is a group of six individuals who have lived experience of an RD (either as a patient or a family member). Five of these members participated in one-on-one interviews and provided insights into the diagnostic journey.

2.3.2 Patient/carer engagement

Patient/carers were invited to participate in an online survey and if they chose, follow this up by participating in a focus group or individual interview.

Survey

An online survey (Appendix 3) was developed and circulated via the Tasmanian Department of Health's Facebook page and through RD advocacy organisations. This anonymous survey was the first step in obtaining information from patients with RD and their family/carers. There were 880 valid responses to the survey (significantly more than expected), with the complete analysis included in Appendix 4, and key findings integrated into relevant sections of Chapter 4 'Community Needs Assessment'.

Interviews

At the end of the survey, participants could nominate (and provide their contact details) if they were willing to contribute further to the research through a process of direct consultation via interview. Participants agreeing to be contacted (n=248; 28%) were emailed with 16 online or face-to-face (Devonport, Launceston or Hobart) focus group options to select from (morning, afternoon and evenings over 5 days). The option of a one-to-one interview was also offered to those who could not attend and/or did not want to participate in a focus group. From 248 invitations, 63 people participated in a focus group and eight people had one-to-one interviews. An additional three people participated after becoming aware of the consultations through an advocacy organisation, resulting in a total of 74 individuals participating.

The areas of focus for the consultations were:

- Experience and journey of the health condition; diagnosed or undiagnosed
- Barriers and enablers in the diagnostic journey
- Costs associated with getting the diagnosis and other cost-related barriers
- Interactions with the health system and quality of care.

2.3.3 Clinician/researcher engagement

Clinicians and researchers were engaged through a survey and/or interviews.

Survey

A short survey provided an opportunity for clinicians and researchers across the state to participate (Appendix 4). There were 31 responses to the survey and the results from this analysis are included in the Survey Report in Appendix 3.

Interviews

Clinicians and researchers who play a critical role in the RD field were nominated by project managers and key informants. Fifteen of these clinicians/researchers provided input from various perspectives, including: a professional within the system (enablers and barriers); their observation and experience of the patient's perspective of the diagnostic journey: and as quality of life for a patient beyond diagnosis (interview questions in Appendix 5). This group of stakeholders provided input into:

- System enablers and barriers for clinicians/researcher in supporting early diagnosis
- Diagnostic care pathways that patients follow
- Enablers on the diagnostic pathway
- Barriers, gaps, issues on the diagnostic pathway
- Prioritising and addressing the barriers/issues of the diagnostic pathway.

Table 1: List of researchers and clinicians interviewed

No	Name	Affiliation
1	Prof John Christodoulou	The Undiagnosed Disease Networks of Australia; Director of the Murdoch Children's Research Institute's Genetics Research Theme
2	Tiffany Boughtwood	Managing Director, Australian Genomics, The Murdoch Children's Research Institute
3	Thulasee Sri Ganeshan	Senior Program Manager, Health Education and Training Institute, Centre for Genetics Education, NSW Health
4	Dr Mathew Wallis	Clinical Geneticist, Clinical Director of the Tasmanian Clinical Genetics Service, Royal Hobart Hospital
5	A/Prof Mimi Berman	Clinical Geneticist, Lecturer at University of Sydney, Vice President Human Genetics Society of Australasia
6	A/Prof Stephanie Best	Senior Research Lead, Implementation Science at Peter Mac in the Department of Health Services Research; Senior research fellow at the Australian Institute of Health Innovation (AIHI), Macquarie University, Sydney.
7	Prof Matthew Jose	Professor of Medicine at the University of Tasmania's College of Health and Medicine, and Chair of the Australian and New Zealand Dialysis and Transplant Registry (ANZDATA) Steering Committee
8	Jan Williamson	The Royal Hobart Hospital Medical in Charge, Molecular Medicine Scientist at Department of Health, Tasmania

No	Name	Affiliation
9	Dr Natasha Brown	Clinical Geneticist, Victorian Clinical Genetics Services
10	Rachel Pope-Couston	Senior Genetic Counsellor, Tasmanian Clinical Genetics Service
11	Dr Alison Turnock	Medical Director - GP & Primary Care, Department of Health, Tasmania
12	Dr Hannah Jackson	GP, Senior Lecturer at University of Tasmania, co-founder of Doctors with Disabilities Australia
13	Joy Mendel	Manager, Clinical, Technical and Ethical Advice, Department of Health, Tasmania
14	Christine Padgett	Associate Lecturer at University of Tasmania
15	Jenny Atkins	PhD student at University of Tasmania
16	Dr Nicholas Blackburn	Research Fellow in Computational Genomics at the Menzies Institute for Medical Research
17	Prof Kathryn Burdon	Menzies Institute for Medical Research, University of Tasmania
18	Prof Gareth Baynam	Clinical Geneticist, Clinical Genomics Policy Advisor at WA Health, Director of the Undiagnosed Diseases Program
19	Dr Georgia Hay	Research Fellow, Curtin University, Perth, WA
20	Lauren Dreyer	Nurse Coordinator, UDP-WA
21	Jodie Courtney	Program Coordinator, Primary Health Tasmania

2.3.4 Advocacy organisations interviews

From 15 advocacy organisations located in Tasmania that were invited to participate, eight provided insights into the patient journey and a patient perspective on receiving care (interview questions in Appendix 5). The questions focused on areas such as:

- Understanding patient journeys
- Barrier and enablers in patient journeys
- Cost of getting a diagnosis and treatment
- Supports required during the diagnostic journey.

2.4 Developing Models of Care

MoC components were developed based on information gathered from the range of stakeholder consultations (survey, interviews and focus groups) and research undertaken in relation to best practice diagnosis and management of RD. The draft MoC components and associated recommendations were reviewed by the CRG as well as TRUDN members, and all feedback was incorporated into the MoC components and recommendations provided in this report.

3 Situation analysis

3.1 Introduction

Although RD cannot generally be prevented or cured, individuals with RD need a diagnosis to access the most appropriate care. This can be achieved through RD diagnostic pathways that set out a clear process for clinicians to follow to obtain a diagnosis and guidance for post-diagnostic care. A rare disease specialist in Western Australia (WA) highlighted that "this is an area of such severe and large unmet need where rare and undiagnosed diseases are like a hidden, global epidemic. Firstly, just attaining a diagnosis is distressing and complex. On average it takes five years to get a diagnosis for a child with a rare disease".[9]

3.1.1 Defining rare disease

While there is no universal definition for RD, Rare Diseases International (RDI), working with the World Health Organization (WHO), developed an internationally endorsed description of RD as a medical condition with a specific pattern of clinical signs, symptoms, and findings that affects fewer than or equal to 1 in 2000 persons.[10]

Other definitions tend to be driven by the regulated designation of orphan medicinal products noting prevalence varies from country to country. For example, orphan medicinal product regulation in the European Union defines RD as "conditions affecting less than 50 per 100,000" and is limited to individuals in the European population. Similarly, orphan drug legislation in the United States (US) defines rare diseases as "conditions affecting less than 200,000 individuals in the United States"[3] (or 1 per 1,662 based on the US 2020 census population of 331,449,281 people).[11] Similarly, the Therapeutic Goods Administration (TGA), Australia indicates the orphan drug designation eligibility criteria for rare conditions that affects fewer than 5 in 10,000 individuals in Australia.[12]

The Australian Government Department of Health has defined a RD as having a prevalence of fewer than five cases for every 10,000 persons (or 1 per 2,000), mirroring both the TGA and RDI/WHO Operational Definition of RD.[1, 10, 12]

3.1.2 The hidden prevalence of rare diseases

There are more than 7,000 RD, many are life threatening or chronically debilitating. Around 8% of Australians (2 million people) live with an RD,[1] including an estimated 35,000 to 45,000 Tasmanians. Furthermore, the inadequate diagnostic coding systems for RD lead to inconsistent coding and a limited understanding of the disease burden for these conditions in the community, leading to subsequent challenges for planning and delivering appropriate health services to meet current and future needs.[13]

Data collection is critical for RD because the small number of individuals with each RD results in a lack of data, evidence and knowledge. The impact of RD remains largely hidden due to inadequate information systems within healthcare systems designed to respond to individual diseases with much larger patient numbers.[2, 14] Globally, there is an increasing recognition of RD in public health. The United Nations (UN) has highlighted the importance of recognizing RD in policies, and in 2021, it adopted the UN Resolution to promote greater integration of RD in the UN agenda.[3]

At present, most information about RD as a collective group, including incidence and prevalence, are estimates given the absence of systematic patient data collection (e.g., registries) in Australia.[15, 16] In

2018, Rare Voices Australia (RVA) established the National Alliance of Rare Disease Registries.[1] The Alliance aims to promote person-centred best practice, encourage uniformity around key principles and commit to further developing a growing understanding of the national RD picture. Subsequently, RVA was commissioned by the Australian Government to develop a National Strategic Action Plan for Rare Diseases.[2] Published in 2020, the Plan calls for a national approach to person-centred RD registries to support national standards, best practice and minimum data sets. Examples of registries or resources that track the epidemiology of RD in Australia have been identified [16] and include the Australian Paediatric Surveillance Unit,[2] Australian Inherited Retinal Disease Registry,[3] Australian Neuromuscular Disease Registry[4] and Australian Cancer Atlas.[5]

Similarly, RD are underrepresented in current health system coding system, and identifying RD from hospital activity data poses significant challenges. Accurate diagnostic coding, coder expertise, and the use of correct codes are essential components in this process. Ensuring unambiguous diagnostic codes is vital to facilitate RD recognition and ultimately, timely access to appropriate care for patients. While it is possible to approximate the prevalence of RD using hospital activity data,[17] RD are known to be underrepresented in the main coding system used in Australia, ICD-10.[14]

In response to the underrepresentation of RD in ICD-10, the WHO established a Topic Advisory Group for RD, managed by Orphanet. ICD-11 was subsequently adopted by the World Health Assembly in 2019 (effective from January 2022) and now encompasses nearly 5,500 RD.[18] Orphanet also developed, and maintains, an inventory of RD with a unique ORPHAcode cross-referenced to ICD-10 and ICD-11.[6] These ORPHAcodes identify clinically unique and distinct entities with prevalence equal to no more than 1 in 2,000 in the general European population. In Finland (Helsinki University Central Hospital in collaboration with Apotti systems), understanding the burden of RD has been facilitated by the implementation of ORPHAcode classification.[19] Recommendations from the Rare 2023 Foresight Study, released in 2021, include implementation across Europe of ORPHAcodes nomenclature into health information systems to improve data; and integration of key diagnostic information into ORPHAcodes to support patient access to appropriate care prior to diagnosis.[20] ORPHAcodes also represent a key future opportunity in Australia to further enhance the national approach to improve data for RD prevalence and health care. Despite this progress, the full impact on global statistics once health systems switch to ICD-11 or ORPHAcoding will take several years to become apparent.

In addition to limited data, it is evident that as a group, RD patients face similar challenges regarding diagnosis, treatment, and care.[12, 13] A study conducted in the United States study indicated that in 2019, there were an estimated 15.5 million children (n = 1,322,886) and adults (n = 14,222,299) with one of 379 RD, resulting in a total economic burden of $997 billion.[14] As this study demonstrates, the economic burden of RD is very high and therefore, coordinated efforts are being made internationally to identify the gaps and promote equity in providing access to rare disease diagnosis and treatments.[21, 22]

[1] https://rarevoices.org.au/research/#:~:text=National%20Alliance%20of%20Rare%20Disease%20Registries&text=The%20Alliance%20aims%20to%20promote,the%20national%20rare%20disease%20picture.
[2] https://www.apsu.org.au/
[3] https://www.scgh.health.wa.gov.au/Research/DNA-Bank
[4] https://www.australiannmdregistry.org.au/
[5] https://atlas.cancer.org.au/
[6] https://www.orpha.net/consor/cgi-bin/Disease.php

3.1.3 National actions towards improving rare disease diagnosis in Australia

The Australian Government has taken a multi-faceted approach to address RD diagnosis, including initiatives to improve access to genetic testing, increase awareness of RD, and enhance coordination between healthcare providers.

The 'National Strategic Action Plan for Rare Diseases'[2] comprises three core pillars, with each pillar outlining priorities, actions and implementation areas: (1) Awareness and Education; (2) Care and Support; and (3) Research and Data. The Action Plan sets out a framework for improving the diagnosis, treatment, and management of RD in Australia. It includes actions such as improving access to genetic testing, increasing awareness of RD, and enhancing coordination between healthcare providers.[2]

The Australian Government acknowledges that access to testing is critical for timely diagnosis and provides financial assistance through the Medicare Benefits Schedule (MBS) for eligible genetic testing services. Genetic testing services covered under the MBS are generally categorised under the pathology services section. The specific genetic tests covered, and the level of reimbursement, can vary depending on the type of test and the patient's circumstances. A healthcare professional or genetic counsellor can provide specific guidance based on individual circumstances and the specific genetic test in question.

The Australian Government also supports research programs, such as the Australian Genomics, through the Medical Research Future Fund.[23] Australian Genomics is an NHMRC funded, national collaboration of researchers, healthcare providers, and patient groups that aims to use genomic sequencing to improve the diagnosis and treatment of RD. The Alliance has established several projects to support the diagnosis and management of RD.

Other initiatives include:

i. *Increased funding for research*: The Australian Government has increased funding for rare disease research through initiatives such as the Medical Research Future Fund. This funding has supported research into RD and has helped improve the understanding of these conditions.[24] One example is the Australian Undiagnosed Diseases Network (UDN-Aus), which is a collaboration between healthcare providers and researchers. The program aims to help patients with undiagnosed conditions by using genomic sequencing and other advanced diagnostic tools.
ii. *Commissioning the National Rare Disease Framework*: Patient advocacy group RVA represents people with RD. In 2017, it documented a 'Call for a National Rare Disease Framework', which listed six strategic priorities: diagnosis, access to treatments, data collection, coordinated care, access to services, and coordinated research.[25] This culminated in RVA leading the development of the above-mentioned 'National Strategic Action Plan for Rare Diseases'.[2]
iii. *The National Health Genomics Policy Framework*: RD are included in the scope of this policy framework that aims to help "people live longer and better through appropriate access to genomic knowledge and technology to prevent, diagnose, treat and monitor disease" through a person-centred approach, a skilled workforce, sustainable and strategic investment, safe and high quality services, and data.[26] The Framework includes an implementation plan.
iv. *RARE Portal*:[7] RVA is leading the collaborative development of the Portal, which is a key deliverable of the National Strategic Action Plan for Rare Diseases. The Rare Awareness Rare Education (RARE) Portal contains current, reliable and straightforward information and resources for all RD stakeholders customised for the Australian context.

[7] https://rarevoices.org.au/rarest-project/

v. *RArEST Project[8] and Project ECHO:[9]* The RArEST Project was awarded $1.9 million in funding from the Australian Government and will develop and deliver RD awareness resources, education, support and training. The RArEST Project comprises three streams:

- Stream 1: Support for individuals, including mental health and wellbeing resources
- Stream 2: Health professional education, support and training: The RArEST Project team is using the Project ECHO® model to create a community of clinical learning practice to increase awareness of RD and provide health professionals with multidisciplinary peer learning and evidence-based, clinically informed expert support to deliver contemporary best practice health care nationally
- Stream 3: Adopting a co-design approach to awareness and education for systemic improvement in RD care and support.

3.1.4 Rare disease research and clinical support initiatives in Australia

Across Australia there are several programs, services, research networks and initiatives that are focused on genomics and RD. There are several programs that help people with RD receive a timely and accurate diagnosis. Detailed below are some of the initiatives focused on RD diagnosis (noting some services deliver a broader range of genetic and genomic support or projects).

i. *Undiagnosed Diseases Network Australia (UDN-Aus)[10]* is undertaking a national pilot project that will support patients with an undiagnosed disease to access further genomic analysis and facilitate linkages to relevant research teams interstate and internationally to enable diagnosis.

ii. *Undiagnosed Disease Program, Western Australia*: The Government of WA is taking a leading role in establishing a Rare Care Centre that is aimed at providing a holistic MoC for children with rare and undiagnosed diseases.[9] It also reinforces the challenges associated with i) timely and accurate diagnosis, ii) coordinated and integrated care, and iii) addressing mental health and Aboriginal health needs. It is believed that establishing the Centre will, in time, reduce the cost to the WA health system. The Centre will be based at Perth Children's Hospital and connect to medical experts from around the world. A population-based study in WA noted that while 2.0% of the WA population was registered as having an RD, this group accounted for 4.6% of people discharged from hospital and had a greater than average length of stay, for a total of 10.5% of overall hospital discharge costs.[14] According to the TCGS research group, this estimate is believed to be similar to the Tasmanian context, reinforcing the potential burden of RD for patients and the impact on the health system if not diagnosed early and managed effectively.

iii. *Queensland Genomics, Queensland*: Queensland Genomics implemented a project for early identification of RD in children. The project explored the use of rapid whole genome sequencing as a first line tool in the diagnostic pathway for babies and children in intensive care with suspected rare disease.[27] The project specifically aims to improve timeliness of diagnosis and access to key services for regional patients and their families through a new MoC which supports and upskills general paediatricians in rural and regional Queensland. It proposes a new MoC that involves i) providing genomic testing for children with rare neurodevelopmental disorders to improve diagnostic understanding of patients with complex disorders such as Fragile X Syndrome, Angelman's Syndrome and Rett Syndrome and ii) establishing a multidisciplinary team of experts to

[8] https://rarevoices.org.au/rarest-project/
[9] https://rarevoices.org.au/rare-disease-project-echo/
[10] https://www.udnaus.org/

support general paediatricians across Queensland caring for patients with rare neurodevelopmental disorders.[27]

iv. *Rare Disease Now:* Rare Disease Now (RDNow) is an initiative to deliver genomic diagnoses and precise, personalised care to children at The Royal Children's Hospital (RCH) Melbourne. Drawing on the research and clinical expertise at the Murdoch Children's Research Institute and Victorian Clinical Genetics Services, RDNow is establishing a pathway for children who remain undiagnosed after a genomic test such as exome sequencing. This will give them the best chance of receiving a diagnosis and access to the latest clinical trials and treatments.[11] The initiative provides opportunities for families to take part in studies that will increase the Institute's ability to provide families with care and support and enhance clinical and scientific knowledge of rare conditions.

v. *Diagnostic support services:* One third of RD have facial clues and identifying these can aid diagnosis. Below are such examples of the technologies used for identifying these clues for a diagnosis.

- CliniFace:[12] Cliniface assists with medical diagnosis, patient screening, treatment monitoring, clinical trials, and surgical planning through an objective, transparent, and unobtrusive analysis using 3D facial imaging. Diagnosing patients with syndromes/RD using facial phenotyping can be very challenging and requires a high level of expertise and experience. Because RD are individually uncommon and the patterns are often very subtle, they can go unrecognised. 3D facial analysis offers a precise and objective method of highlighting the clinically salient aspects of facial variation to help clinicians in making earlier and accurate diagnoses.
- FaceMatch:[13] The FaceMatch Project uses computer vision technology to match the faces of individuals from around the world to help parents searching for a diagnosis for their child. It aims to help people with a possible genetic condition find a diagnosis by matching their facial features with people who already have a diagnosis.

vi. *Clinical services across Australia:* There are many genetic services across Australia that offer a diagnostic service for people who have complex or undiagnosed medical conditions. These services generally use a team-based approach to assess and diagnose patients, including medical specialists such as geneticists, genetic counsellors, and laboratory technicians. Some of these comprehensive services offer a full range of multidisciplinary clinics for individuals and their family members who have concerns about their personal and family history of genetic illness. The services provided are risk assessment, genetic counselling, genetic testing and medical advice as well as psychological support to individuals and their family members. The full list of services by jurisdictions across Australia is available on the Human Genetics Society of Australia's website.[14] Two genetic services are listed within Tasmania:

- Public Genetic Service: Tasmanian Clinical Genetics Service located at the Royal Hobart Hospital
- Private Genetic Service: Icon Cancer Centre Genetic Counselling Service located in Hobart

vii. *Genetic Support Network Victoria (GSNV), Australia :* Genetic Support Network Victoria (GSNV) [28] is an incorporated association, which provides state-wide support services to all people with genetic, undiagnosed and rare conditions, and those who support them. It has a Patient Pathways Program [29] where a Telehealth Patient Pathways Nurse provides free information and support

[11] https://www.mcri.edu.au/research/strategic-collaborations/flagships/rare-disease
[12] https://cliniface.org/about
[13] https://facematch.org.au/home
[14] https://www.hgsa.org.au/Web/Web/HP-Resources/Clinical-genetics-services-by-state/Clinical-Genetic-Services.aspx?hkey=e11c39d6-37f4-4e68-8709-21eff5f6e006)

services and links patients with genetic, undiagnosed and rare disease communities within Australia.

The GSNV Directories is the first-ever initiative that addresses the multidisciplinary support that exists for rare genetic conditions on a national, international, and virtual level. The database of these Directories captures the most up to date information about genetic support groups, with the help of a dedicated and skilled group of GSNV volunteers. It also has a Directory of Rare Disease Health Professionals which provides a list of health professionals with specialised knowledge and experience in rare and genetic diseases. This register can encourage collaboration, a multidisciplinary approach and be a valuable resource for health professionals caring for people with rare and genetic diseases.

Besides the above-mentioned programs, projects such as "Investigating perceptions of unmet need in Australia" by the Australian Genomics[8] and "Improving access to Genetic Health Services for Aboriginal and Torres Strait Islander Queenslanders" by QIMR Berghofer, have indirectly assisted the RD field through engaging with and developing care models and referral pathways to improve access to genetic health services. The latter is implemented in collaboration with the Queensland Aboriginal and Islander Health Council and Genetic Health Queensland.[30] New undiagnosed disease programs, such as the Undiagnosed Diseases Network (UDN) of Australia and Undiagnosed Diseases Network International, offer opportunities for clinicians to enroll patients and families in cutting-edge clinical research.[31]

3.1.5 Context of rare disease care delivery in Tasmania

About 80% of all RD have a genetic cause,[2] and a genetic diagnosis can be critical to finding appropriate management or treatment. Conducting genomic testing in a timely manner can be challenging, particularly in remote and rural areas or if a patient has another complex condition. Initiatives that improve knowledge and awareness of rare disease will help improve the timeliness of rare disease diagnoses. An Australian publication by Elliot et al (2015)[5] indicates that these include, but are not limited to:

- Developing and disseminating information regarding RD that is relevant to the Australian context for patients, parents, carers and the general public, thereby improving health literacy
- Providing educational resources and networking opportunities for health professionals to allow them to better identify and manage RD
- Increasing knowledge of the epidemiology and impacts of RD in Australia to clarify opportunities for improved data and coding
- Raising awareness of the burden of RD on families, health professionals, and the community
- Improving healthcare for people with RD through better access to diagnostic tests, new treatments and specialised services.

To implement these actions in Tasmania, it is critical to understand the Tasmanian context in terms of the challenges to accessing care and diagnosis. Tasmania is home to a regionally dispersed population of over 570,000 people.[15] Information from Primary Health Tasmania's needs assessment[34] indicates that Tasmania's ageing population and relatively high levels of socioeconomic disadvantage are contributing to significant pressure on the entire health system. In 2015, the Tasmanian Government set a target to grow the population to 650,000 people by 2050 to create jobs and drive economic growth.[16] Some key concerns influencing health outcomes of Tasmanian people include income, education, employment opportunities and ability to participate in the community.

[15] https://www.abs.gov.au/statistics/people/population/national-state-and-territory-population/latest-release
[16] https://www.stategrowth.tas.gov.au/policies_and_strategies/populationstrategy

Barriers to accessing care and diagnosis in Tasmania include:

- ***Dispersed population:*** Tasmania has the third smallest population by jurisdiction in Australia, and it is significantly dispersed with 66% of the total population residing outside of Greater Hobart. In addition, only Greater Hobart and Greater Launceston regions are considered 'inner regional' under the Australian Statistical Geography Standard (ASGS) Remoteness Structure (Edition 3, accounting for accessibility to services using the Accessibility/Remoteness Index of Australia Plus (ARIA+)); whereas the remaining areas are considered 'outer regional' or 'remote' (e.g., on mainland Tasmania, Bruny Island) and 'very remote' on islands further offshore (e.g., King Island, Furneaux Group). Twenty-one of the total 29 local government areas are 'outer regional' or 'remote'[35, 36].
- ***Geographical inequity:*** Qualitative consultations identified concerns about uneven distribution of health care professionals across Tasmania. In particular, the North West region has a lower density of allied health professionals, medical professionals and nurses and midwives than elsewhere in Tasmania. While some additional health professionals would be expected in the South and North as a reflection of the State's clinical services profile, this disparity is larger than expected. In medicine, there are a number of specialty areas with a lower supply of specialists than the national average, with the number decreasing per head of population from the south to the north and to the north-west. In nursing and midwifery, the North West has a lower number of Registered Nurses and Midwives per capita compared to Tasmanian and national rates.[37]
- ***Transport disadvantage*****:** People living in regional Tasmania experience greater difficulty in accessing transport than people living closer to the main population centres. This includes limited access to public transport (e.g., limited and longer bus services compared to urban centres). The public transport system struggles to meet the needs of the State's dispersed population. In addition, the higher relative socio-economic disadvantage means that car ownership is not possible for some families.[38, 39] These factors limit many Tasmanians' access to health services. While the Tasmanian Government provides subsidies and funding for services for individuals who need to travel for healthcare, such as the Patient Transport Assistance Scheme (PTAS) and Community Transport Services Tasmania, these services do not always meet existing need.
- ***After-hours and emergency care*****:** As mentioned in the key informant interviews, Tasmanians have few options to access general practice in the after-hours period, especially in outer regional areas. Limited face-to-face options contributes to people using ambulance services and emergency departments for less urgent care.
- ***Socio-economic and educational disadvantage:*** There is a higher proportion of Tasmanians (14.8%) compared to Australians (9.8%) residing in areas (SA1 or statistical areas 1) with an Index of Relative Socio-economic Disadvantage (IRSD) in the lowest decile. This indicates that Tasmanian people and households overall, have relatively poorer economic and social conditions compared to Australia. Similarly, there is a higher proportion of Tasmanians (21.1%) compared to Australians (9.6%) residing in areas (SA1) with an Index of Education and Occupation (IEO) in the lowest decile. This indicates that Tasmania has lower education and occupation levels overall compared to Australia.[40]
- ***Health Literacy*****:** Low education levels in Tasmania exacerbate low levels of health literacy. Tasmanians with low levels of health literacy find it hard to access health information and services, understand health information, and use information to make informed choices about their health and health care.[38, 41] A 2022 study supported by the Tasmanian Government, 'Optimising Tasmania's Healthcare', reports that Tasmania has overall lower levels of education, particularly with reference to early school leavers (28.6% compared to 22.6% nationally) and those who had completed university (27.4% compared to 33.8% nationally).[42] The study also indicates relatively low health literacy levels among some population sub-groups and suggests that tailored focus on

improving health literacy is required in Tasmania to optimise equitable access to and engagement with health information and services and to support the health and wellbeing.

- ***Accessing primary care*:** People in regional and remote communities can experience barriers to accessing primary care services. General practice, allied health and community nursing services are less accessible locally for people living outside urban population centres in Tasmania. Communities may rely on visiting services, which present challenges in delivering continuity of local primary care.[38]
- ***Recruitment and Retention:*** Attracting and retaining healthcare professionals, including doctors, nurses, and allied health workers, has been a persistent challenge for Tasmania. The state has experienced difficulties in recruiting and retaining skilled healthcare workers, particularly in regional and remote areas. These professional areas include but are not limited to critical care, maternity services, mental health services and aged care services.[37]
- ***Specialty Shortages:*** Certain specialty areas may face shortages of healthcare professionals. Tasmania has identified shortages in specialties such as neurology, psychiatry, emergency medicine, general surgery, and some allied health professions. These shortages can impact the provision of specialized care and lead to longer than clinically recommended waiting times for patients.[37, 38] Stakeholder consultations identified that there is limited RD diagnosis system capacity to support the needs of Tasmanians including the clinical workforce, laboratory staff, geneticists, genetic counsellors, paediatricians, and genomics. This has led to long wait lists for diagnostic testing.
- ***Limited ability to establish multidisciplinary teams****:* Diagnosis, treatment, and care of patients with RD require multidisciplinary collaboration between medical and paramedical specialities and with patients and families,[13, 43] requiring more complex care pathways. Limited availability of the specialists across Tasmania results in a lack of multidisciplinary input in patient diagnosis and care. Currently multidisciplinary teams (MDTs) in Victoria support Tasmania in clinical decision making and planning the next steps to take in patients' diagnostic journeys.
- ***Consumer lack of awareness:*** Consumers are often unaware of the services available to them, the cost of services, and how services can be accessed in Tasmania. This results in consumers receiving care from services that do not best meet their needs, and challenges navigating the service system.[44]

These and other health system factors make the Tasmanian context challenging, and are common to rural and regional areas across Australia. Early diagnosis of disease is essential as it results in longer periods of higher quality of life, better patient outcomes and reduced expenditure on hospital admissions.[45, 46] While research shows that implementation of care pathways promotes timely diagnosis, person-centred care and better management of the condition,[47-49] the combined impact of all the above factors needs to be considered in designing improved diagnostic pathways for individuals with rare disease.

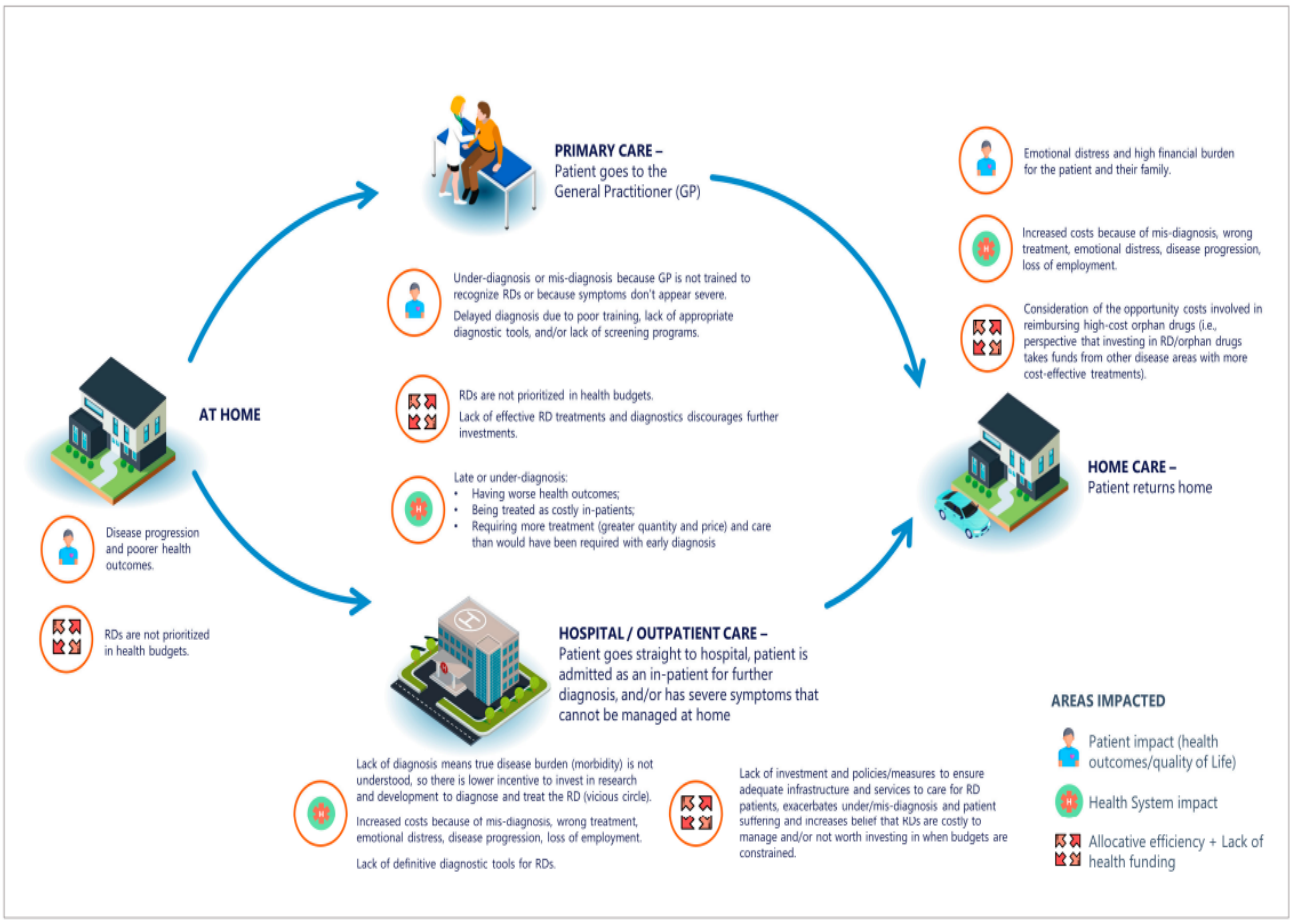
3.2 Challenges to rare disease diagnosis

The challenges associated with obtaining a timely and accurate diagnosis for individuals with RD are multifaceted and cover the entire diagnostic journey.[49] Diagnosis of RD has been revolutionised over the last decade, however, and these advances have led to significant improvements in diagnostic rates and a rapid increase in the number of genes underlying rare diseases being identified. In recent years, genetic testing has advanced from single gene testing offered in specialist services to increasingly accessible genomic testing involving massive parallel sequencing, such as exome or even whole genome sequencing. GPs can currently request a range of genetic/genomic tests and all Australian states and territories have specialist genetic risk assessment, testing and counselling services.[50] Challenges continue, however, even with whole genome sequencing, with around 50% of patients with RD not receiving a diagnosis.[51] 'Next

generation sequencing' also generates vast amounts of information that can be difficult to interpret and presents greater risk of misuse.

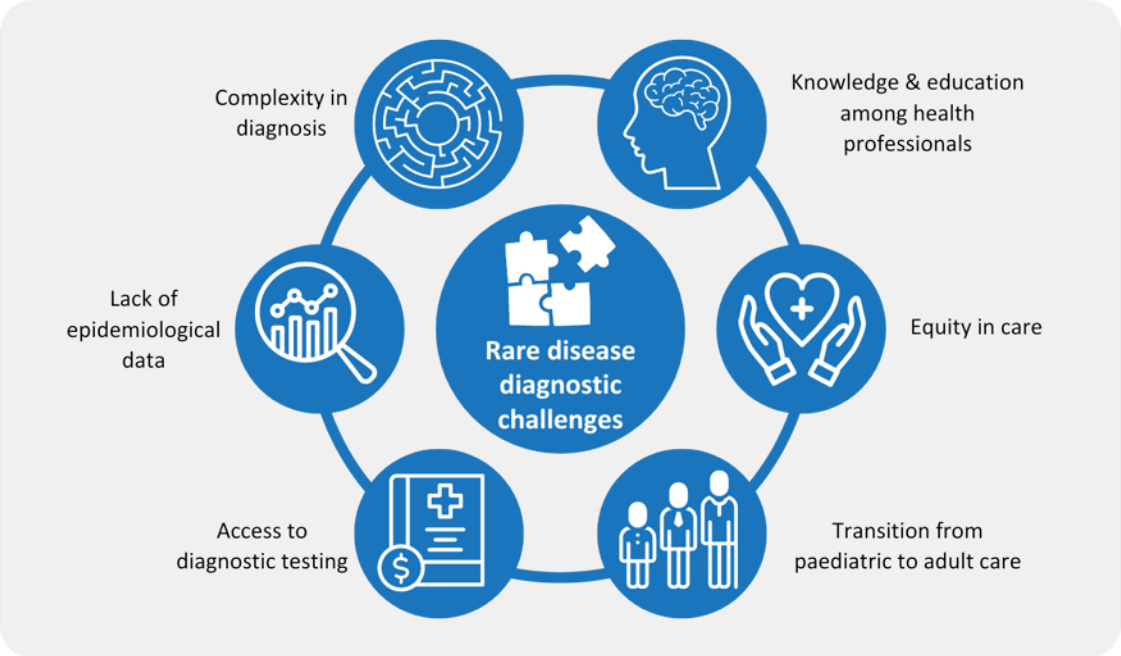
In a recently published international paper, key challenges experienced by RD patients along the journey from onset of symptoms to diagnosis and care were identified and are illustrated in Figure 2 below.[52] Diagnosis is often a lengthy journey, regardless of the country or region, with many rare disease patients having to endure multiple interactions with specialists and healthcare providers to be accurately diagnosed.[52, 53] Moreover, even after diagnosis, there are ongoing challenges for some, with a lack of approved medical treatments for some rare diseases.

Figure 2: Pictorial representation of the challenges to rare disease diagnosis and care, international evidence



The six themes in Figure 3 below were identified in a review of the literature, as well as by stakeholders interviewed for this project. There were also disease-specific issues expressed by various specialists through consultations which could inform the development of disease-specific care pathways. These themes are discussed in further detail below.

Figure 3: Key challenges in rare disease diagnosis



3.2.1 Complexity in diagnosis

An accurate diagnosis is vital in managing an RD appropriately and identifying specific resources and interventions for the best possible clinical outcome for patients. Specific benefits of timely diagnosis include enabling access to timely and best practice medical care, reduction in unnecessary investigations, reduction in inappropriate management or admissions, clarification of recurrence risks, potential availability of additional reproductive options, reduced isolation and peer support, and enhanced access to timely social and educational services.[9, 43, 47] In Spain, risk of diagnostic delay for RD was associated with the following determinants:[54]

- People who first sought medical advice from their primary health care provider had a higher risk of diagnostic delay compared to specialist advice
- People who had to travel to hospitals or specialists other than those usually consulted in their home province had a higher risk of diagnostic delay compared to those who did not need to travel as much
- People affected by diseases of the nervous system had a higher risk of diagnostic delay compared to other RD
- The higher the frequency of visits to specialists, the longer the diagnostic delay, especially in cases where patients consulted specialists more than 10 times indicating complexity in diagnosis

It is reported that 30 per cent of Australian adults living with an RD are impacted by a diagnostic delay of more than five years, while almost half have received at least one misdiagnosis. Both diagnostic delay and misdiagnosis can negatively impact the experience of care and support received by individuals.[2]

Other factors that contribute to delayed diagnosis include:

- **Not suspecting RD:** A study conducted in Japan identified one of the most important factors related to delayed diagnosis of RD is the lack of suspicion of RD or under-recognition by patients and their medical professionals.[55] Current policies tend to focus on the stage of the diagnostic journey between suspecting RD to the time of a clear diagnosis and post-diagnosis care, whereas it is similarly important to focus on the interventions that are needed to facilitate patients and primary care clinicians to suspect a rare condition (for example via a detailed family history or

noticing a clinical presentation consistent with genetic conditions) and its early symptoms along the patient journey.

Many RD are not well-known, even among healthcare professionals. The rarity of these conditions means that they are less likely to be encountered in routine medical practice. As a result, healthcare providers may not consider them initially, leading to delays in diagnosis. Unnecessary consultations cause substantial costs for the individual and for healthcare systems; before the correct diagnosis is made, patients see an average of 7.3 physicians.[56] Therefore, there is an urgent need to improve RD diagnosis.[57] The stakeholders consulted commented on this with a specific note that if there is no family history known to the family then it takes even longer to suspect the RD.

- **Not respecting patients' observations about their own or their child's health:** It was noted by multiple stakeholders that having a practitioner believing in the patient and taking patient symptoms and complaints seriously was the single most important factor in moving the patient along on their diagnostic journey in a timely manner. Finding a practitioner who will provide patient-centred care that is built on mutual respect and sharing of knowledge was mentioned as a critical factor.
- **Diagnostic Processes:** Consistent with studies conducted by the International Rare Disease Research Consortium, diagnostic delays and misdiagnoses are common for patients with RD in Australia.[46] This also reflects the complexity of diagnostic processes which differ for each patient based on symptoms, family history, presence of other conditions, and knowledge of the health professional. Complex multi-system disorders require a diagnostic process that involves multiple medical specialties and a systematic method of excluding common conditions. Some of the reasons for complexity in identifying the rare diseases are:
    - ***Non-specific symptoms****:* Rare diseases often have a wide range of symptoms that can be non-specific and overlap with more common conditions. This makes it challenging for doctors to recognise the pattern and associate the symptoms with a specific rare disease. Patients may present with symptoms that are vague, intermittent, or fluctuating, further complicating the diagnostic process.[5]
    - ***Limited research and knowledge****:* Due to their rarity, many RD have not been the subject of significant scientific research and therefore there is limited information available on the underlying causes, mechanisms, and clinical presentations of these conditions. This scarcity of information makes it difficult for healthcare professionals to accurately diagnose rare diseases.[58]
    - ***Diagnostic testing challenges:*** The diagnostic journey for RD often involves multiple specialists, extensive testing, and a process of ruling out more common conditions. Diagnostic tests specific to RD may not be readily available or may require specialised laboratories. Moreover, some RD have overlapping symptoms with other conditions, leading to misdiagnosis or delayed diagnosis.[22]
    - ***Fragmented healthcare systems***: Healthcare systems are often designed to address common conditions and may lack the infrastructure, expertise, or resources to effectively diagnose and manage RD. This fragmented nature of healthcare can contribute to delays and difficulties in obtaining an accurate diagnosis.[53] Key informants also noted that fragmented care leads to complexity in diagnosis.

3.2.2 Knowledge and education among health professionals

General practitioners (GPs) are critical in RD diagnosis, especially in the Australian healthcare system where GPs act as gatekeepers to specialists.[59] Many patients with RD will present to a GP with their symptoms as

a first step. They will also see their GP in between visits to a specialist, and for a range of other primary and preventive care services. This ongoing relationship that also has an accessible, relationship-based advocacy and support role can be a foundation for positive experiences of patients with RD.[60] However, many GPs have reported feeling overwhelmed when caring for RD patients.[61]

For a positive interaction and timely diagnosis, practitioners require sufficient knowledge and information on testing and referral options.[61] GPs face a difficult task when caring for people with diagnosed or undiagnosed RD and the need for training, development, improved communication, and better awareness is highlighted in the literature.[5, 61] GPs caring for children and adults with RD have a crucial role in making appropriate referrals, providing care coordination and linking families to psychosocial and other forms of support. It is important, therefore, that GPs are aware of information portals and educational resources that will assist them to help patients with a RD.[5]

For example:

- A clinician's knowledge regarding available screening programs is critical. Screening and diagnostic programs play an essential role in the diagnosis of RD. Prenatal and newborn screening (NBS) programs[17] are vital in enabling early detection of diseases and, in some cases, early intervention that may lead to better prognosis and outcomes.
- As reproductive genetic carrier screening becomes more widely accessible, ensuring uptake by primary healthcare professionals is essential to equitable and appropriate service provision.[62]

A key to early diagnosis is also specialists' knowledge of RD. Greater collaboration among GPs and specialists with expertise in RD may help to expedite the lengthy process to a correct diagnosis. Recognising that multiple symptoms may be linked rather than remaining focused on treating individual symptoms may help to reduce the time to diagnosis.[63]

Limited knowledge of available diagnostic services and processes was identified by the stakeholders as a limitation across primary care and referral service providers. It was discussed that this resulted in possibly fewer referrals for testing proportionate to need. Stakeholders identified challenges faced by laboratories, including test sample management, clarity of clinical notes and sometimes the risk of delayed notification of the results due to lack of appropriate coordination.

The key informants identified this lack of awareness and knowledge regarding RD among health professionals as one of the critical issues that leads to delays in diagnosis. Clinicians noted that barriers for GPs in diagnosis include limited GP continuity, pressure of time, including for consent process, lack of knowledge, skills and confidence to talk about RD, limited referral pathways, limited knowledge of appropriate tests and referrals, and limited awareness of/access to RD diagnostic resources.

Tasmania has diabetes and kidney transition care clinics which support a range of prevalent genetic diseases, but there is an identified lack of rare genetic disease clinics. RD specialists highlighted the importance of addressing this imbalance by supporting education on rare, chronic complex diseases in young people and adults both for medical students and through post graduate medical education. This research has identified a need to build the capacity of GPs and laboratory scientists in genomics. Availability and access to RD information might also help the GPs to play a role in empowering patients with RD to improve their self-management. Many sources of information are already available and listed on the Australian Paediatric Surveillance Unit at Kids Research website[18] and were highlighted by the stakeholders. Examples include the RArEST Project and Australian Genomics discussed in 3.1.4, as well as:

[17] https://www.health.gov.au/our-work/newborn-bloodspot-screening
[18] http://www.apsu.org.au/rare-diseases/links/

- Centre for Genetics Education, NSW Health web portal for health professionals and patients is focused on genetic conditions and provides fact sheets and guidelines. The Centre for Genetics Education focuses on empowering health professionals to continue delivering improved outcomes in health care and well-being for individuals, families and the community, by providing credible genetics and genomics resources and information, in collaboration with our clinical and research partner organisations.[19]
- The Centre for Genetics Education, Health Education and Training Institute (HETI) offers NSW healthcare professionals with an interest in clinical genomics scholarship opportunities to attend a short course in Clinical & Laboratory Diagnostic Genomics. Scholarships and grants offered by HETI aim to provide equal access to education in areas of need. This course in genomics has been designed for non-genetics trained medical officers and other healthcare professionals, who are currently employed in NSW Health across local health districts or specialty health networks, with emerging practical needs in delivering genomic healthcare.

Many such resources are available for health practitioners improve their skills in clinical and relational management of RD. The stakeholders indicated that the key is to make clinicians aware of these resources, including easy access to retrieve them when required.

European Centres of Expertise acknowledges that access to training is essential to enable health care providers to gain knowledge about RD or refer patients to expert centres, as it is possible not to have an identified national expert for some rare diseases.[13]

3.2.3 Equity in care

RD are often complex, serious, chronic and progressive conditions. Treatment of a rare disease often involves multidisciplinary teams, including a variety of clinical, nursing and allied health specialists, including a geneticist. Some of the system level challenges to service provision in the Tasmanian context are described in section 2.1.6. These challenges include overall low health literacy across Tasmania, rurality and socioeconomic status. For example, specialist services, such as genetic services, mostly operate within urban hospital settings. Patients living in remotes areas are frequently required to travel long distances to access these services, impacting the patient and family in multiple ways such as having to take a day off from work or travel long distance for a short appointment.[64]

Other challenges are specific to cultural and language barriers. Despite the advances in genomic technology use in rare disease diagnosis, “the benefits of these advances are disproportionately experienced within and between populations, with Indigenous populations frequently experiencing diagnostic and therapeutic inequities” more often than the general population.[65] The stakeholders consulted also identified the inequity in diagnostic care for vulnerable communities including Aboriginal and Torres Strait Islander peoples, people with a disability, and culturally and linguistically diverse communities. These factors need to be considered in designing a comprehensive and equitable diagnostic care model.

3.2.4 Transition from paediatric to adult care

The majority of RD are initially diagnosed and treated in childhood, therefore much of the expertise resides with paediatricians, and children’s hospitals. Due to advances in medical technology more children with RD survive into adulthood. The transition period to adult healthcare presents many challenges for paediatric RD. The transition is uniquely difficult to counter because it is driven by a variety of factors, including the

[19] https://www.genetics.edu.au/SitePages/About-us.aspx

complexity of their care, differing specialist recommendations, and the adult service providers' lack of experience with rare paediatric diseases, among other factors.[66]

The providers also struggle to perform post-transitional care when a patient or family's beliefs and expectations of adult care do not align with that of the provider, causing distrust and resistance to change.[67] Many times a lack of communication with the patient's paediatrician limits a clinician's ability to treat the patient, especially when they do not have access to medical records and histories. Stakeholder consultations identified that some cohorts including young people in transition from paediatric to adult services, and older people are 'falling through the gaps' as RD supports have tended to focus on hospital paediatrics services.

> *"Children's hospitals do not see the transition as their problem then those children come to access adult services around at the age of 18-19 which is too late. Issues will be better managed if transition visits were organised earlier. Currently, both patients and clinicians are not prepared for that."*
> (Clinical geneticist)

3.2.5 Access to diagnostic testing

Access to diagnostic testing in RD can be challenging and addressing these issues requires a multi-faceted approach. These barriers to accessing care include:[68]

- **Lack of awareness amongst healthcare professionals**: Many healthcare professionals, including primary care physicians, may have limited knowledge or experience with RD. This can result in delayed or misdiagnosis, leading to difficulties in accessing appropriate diagnostic tests.
- **Limited availability**: Diagnostic tests for RD may be limited in availability, especially in regions with fewer resources or expertise in RD. This can result in longer wait times.
- **Out-of-pocket costs:** If the tests are not Medicare funded then diagnostic tests for RD can be expensive, particularly when they involve specialised techniques or technologies.
- **Time away from employment for carers:** Genetic testing might not be available easily or require travel. This also has impacts for the carers including needing to take time off from work.

A report on unmet needs in genomic testing indicated that almost all clinics had a gatekeeping process for ordering genomic tests and ordering was subject to MDT discretion.[69] Unmet need was defined as patient groups for whom a clinical geneticist thinks a genomic test is clinically indicated and who want genomic testing but cannot access it. The report included the following categories of unmet need:

- Populations who are unable to access genomic testing
- Populations not presenting to genetics services
- Budgetary constraints resulting in challenges in meeting needs
- Workforce implications to ensure clinicians know when to refer, how to refer, and what genetics can offer.

It was suggested that future research should be directed at:

- how to increase equity of genomic testing.
- finding ways to direct outreach services to less served populations.
- how to address funding challenges within genomic testing.

Clinical diagnosis of RD is supported by molecular/genetic testing. The key informant interviews supported the findings listed above indicating that lack of specific tests across Australia, lack of knowledge about the tests and testing facilities, cost of the tests and fear/misunderstanding associated with genetic testing are key issues for fair and equitable access to diagnosis. Access to and funding of genetic testing continues to

be a challenge and can lead to delays in diagnosis, with stakeholders identifying cost and funding structures and responsibilities remaining unclear.

3.2.6 Lack of epidemiological data and supportive information

Currently, high quality national data on congenital anomalies, including RD, is unavailable, despite recognition that they are a significant public health problem in Australia.[2] This lack of data further exacerbates the challenges in providing a consistent approach to diagnosis. Commonwealth, state and territory approaches to RD remain fragmented.[70]

The impact of a lack of epidemiological data on early diagnosis is particularly pronounced in the context of RD. Epidemiological data can:

- provide insights into the prevalence of RD, the characteristic signs and symptoms, and the age of onset, helping healthcare providers consider these conditions earlier in the diagnostic process. The lack of epidemiological data can further prolong the diagnostic process by delaying referrals to specialists and delayed access to diagnostic tools and expertise.
- be instrumental in promoting research and innovation in the field of RD. It helps identify gaps in knowledge and allocate resources for developing diagnostic techniques and treatments. The stakeholder consultation supported these concerns.

  The National Rare Disease Plan for Ireland 2014-2018 included key options in improving access to research and innovation including:[13]

  - Enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of RD, informing care management, research and health system planning
  - Develop a national research strategy for RD to foster, support and drive all types of research for RD, contributing to agreed priorities and systematically addressing gaps
  - Ensure research into RD is collaborative and person-centred
  - Translate research and innovation into clinical care; clinical care informs research and innovation.
- help support advocacy organisations to estimate the number of affected individuals and advocate for increased awareness, funding, and research.

Given the challenges associated with the lack of epidemiological data related to RD, efforts are underway to improve data collection and establish dedicated registries and networks to facilitate early diagnosis, research, and support for individuals affected by RD.

Key informants indicated that there are ad-hoc data, information and resources relating to various RD, and that having resources or information links across multiple platforms is a challenge for navigation through to diagnosis. Across Australia there are many initiatives and resources available to gather information, but how to gather information effectively so that it can be used for health service planning and how to promote the information to ensure doctors and consumers are accessing it are two key concerns.

Current sources of information include:

- Disease specific organisations (guidelines for management)
- Consumer websites (various disease-specific information)
- Primary Health Networks (referral resources for GPs including HealthPathways)
- Existing provider networks (colleagues across practice, hospital)
- Clinical genetics network
- Agency for Clinical Innovation (NSW Govt)

- Centre for Genetic Education (NSW Govt, resources for GPs)
- The Rare Awareness Rare Education (RARE) portal, RVA (prevalence data)
- Australian Genomics (including community advisory group, PanelApp Australia).

3.2.7 Impacts of delayed diagnosis

Impacts of delayed diagnosis vary based on factors such as disease symptoms, socio-economic status, geographical location of the patient as well as patient's literacy levels. The impacts of delayed diagnosis on individuals and families are listed below.

- An accurate diagnosis is essential for genetic counselling and family planning decisions, which can be challenging when the specific genetic cause of the rare disease is unknown. The stakeholder consultations with the advocacy organisations indicated that there are parents who have two children with similar RD diagnoses. It occurs due to a delayed diagnosis or incorrect information provided to the parents before deciding on having the second child. This results in parents having the second child in a hope that the child will not have the condition. Prompt, correct diagnosis is very important for families, as it enables them to explain their child's disease to others, to stop blaming themselves for their child's condition, it may restore reproductive confidence and alleviate some of the stress of not knowing what is wrong and what to expect in the future.[71]
- Children who have undiagnosed RD may face educational challenges or difficulties in school due to unexplained symptoms, frequent medical appointments, and a lack of understanding from educators and peers. As they transition to adulthood, these educational challenges can continue and may impact future career opportunities. Even after diagnosis these consequences continue. [72]
- Accessing support and resources in a timely manner is possible with a diagnosis. Not having a definitive diagnosis can equate to missed opportunities such as access to support groups, advocacy organisations, and resources tailored to the specific rare disease. The patients also indicted that having certain specific symptoms encouraged them to do their own research and find the support organisations. The support organisations and their websites were very useful to patients in advocating their own possible diagnosis with the health professionals.
- Living with an undiagnosed RD can create social challenges as the individual may struggle to participate in typical activities due to their symptoms. This may affect friendships, relationships, and overall social interactions. This includes high risks of experiencing poor quality of life, mental illness, social isolation (patients and caregivers), and negative effects on work-life balance. [22]
- The financial costs of the diagnostic odyssey may be high and unaffordable. Pursuing an accurate diagnosis can involve numerous medical tests and consultations. Additionally, without a diagnosis, health insurance might not cover specific treatments or therapies, leading to increased financial strain. The stakeholder interviews indicated that sometimes having to pay for the tests could be a deterrent to getting a test done.

In addition to the above-mentioned impacts of delayed diagnosis on individuals and families, there is strong evidence regarding these impacts extending to the health system. Some of the key health system-level consequences of delayed diagnosis include:

- Misallocation of resources, as patients may receive treatments and interventions for conditions they do not actually have.
- Increased healthcare costs due to a longer and complicated diagnostic process due to the need to undergo multiple tests and specialist consultations to receive an accurate diagnosis. A study published in September 2023, included seven RD and estimated the avoidable per patient medical

costs and productivity losses attributable to delayed diagnosis at between $86,000 and $517,000 per patient.[20]

- With limited availability of specialist care in Tasmania, a delayed diagnosis mean that patients do not receive appropriate care from these specialists in a timely manner.
- Strain on healthcare resources, including laboratory facilities and specialised equipment.
- Collectively RD affect a significant proportion of the population, and delayed diagnosis can lead to a higher overall burden on the healthcare system. This includes longer hospital stays, increased emergency department visits, and extended outpatient care.[73]

The journey to identify a rare disease can be emotionally and psychologically taxing for patients and their families. It often involves multiple doctor visits, inconclusive tests, and uncertainty. The rare nature of the condition may lead to feelings of isolation and frustration. Complexity of RD makes service navigation by families difficult. The research identified that families affected by RD represent a medically disenfranchised population that falls through the cracks of every healthcare system in the world.[5] The Australian Paediatric Surveillance Unit indicates that people living with RD face significant challenges, including diagnostic delays, lack of available treatment and difficulty in finding the right health service. Families feel isolated, under-supported, and often face economic hardship.[74]

This was reinforced through the consultations where stakeholders cited that families often feel isolated, forgotten and fearful in their diagnostic journey. The TCGS Community Reference Group (CRG) emphasised the importance of individuals knowing what to expect from a consultation with a practitioner. Communicating with individuals experiencing the same journey and having support groups was identified as one of the important elements for patients and carers. The research conducted by European researchers on RD care pathways in the European Union reinforced that patient organisations and resource centres for RD have a special role in patient empowerment and self-management, and this needs to be further promoted.[49]

The next section discusses existing care pathways and includes some best practice diagnostic pathways for consideration.

3.3 Overview of current and best practice diagnostic pathways

Many people living with RD report a difficult diagnostic process from the symptom onset until they obtain the definitive diagnosis. Section 2.3 discussed the determinants related to the RD diagnosis and factors contributing to the diagnostic delay. The Australian health system has established diagnostic pathways. The purpose of this project is to identify how diagnostic pathways for patients with RD in Tasmania, and Australia more broadly, can be improved. It is important to note that although this project focuses on diagnostic pathways and its challenges, there is also a need to improve post-diagnostic pathways, in particular, access to post-diagnostic support. Research by Castro et al[75] highlighted "..the complexity of rare diseases, their strong relation to disability and the current unmet social and daily life needs of people living with a rare disease must not be underestimated and require urgent attention from all stakeholders involved in care provision, from healthcare to social and community services".

Care pathways are defined as "a complex intervention for mutual decision-making and organisation of care processes for a well-defined group of patients during a well-defined period". [7] The aim of a care pathway is to enhance the quality of care across the continuum by improving risk-adjusted patient outcomes, promoting patient safety, increasing patient satisfaction, and optimising the use of resources. Defining characteristics of care pathways include:[7]

[20] https://everylifefoundation.org/new-study-measures-economic-impact-of-delayed-diagnosis-of-rare-diseases/

- an explicit statement of the goals and key elements of care based on evidence, best practice, and patients' expectations and their characteristics
- facilitation of communication among the team members and with patients and families
- coordination of the roles and activities of the multidisciplinary care team, the patients and their relatives
- documentation, monitoring, and evaluation of variances and outcomes
- identification of the appropriate resources.

MoC pathways are a way of setting out the steps that should be followed during the diagnosis and treatment of a patient, specific to a condition where possible. The complexity in rare disease care necessitates the need for care pathways that can facilitate referrals between general medical services and expert resources and testing, and for the sharing of expertise and information between health specialties as well as between health and social services. Care pathways can also be used to inform patients and families about the stages of their individual patient journey, to update health care providers involved in various parts of the treatment and care and to train novices in the field.[13, 43]

While the care pathway encompasses the entire journey of a patient through the healthcare system, including the diagnostic, treatment, and follow-up stages, the diagnostic pathway is a crucial component of the overall care pathway for a patient. It represents the series of steps involved in diagnosing a patient's condition, starting from the initial presentation of symptoms to reaching a definitive diagnosis. The diagnostic pathway is an essential part of the care pathway because an accurate and timely diagnosis is fundamental to providing appropriate care and treatment. The diagnostic pathway typically involves a range of activities, such as medical history assessment, physical examinations, laboratory tests, imaging studies, and consultations with specialists. It aims to identify the underlying cause of the patient's symptoms or condition with an aim to guide healthcare providers in delivering timely and effective interventions based on a confirmed diagnosis. Diagnostic pathways are a critical area of focus RD. These pathways are better established in other fields of medicine, such as cancer diagnosis, when compared to RD.[76]

The sections below discuss some current diagnostic pathways internationally and nationally and their critical components. The first introduces diagnostic pathways in cancer care, as this field has been well established over decades. The following section discusses the currently available diagnostic care pathways in RD nationally and internationally.

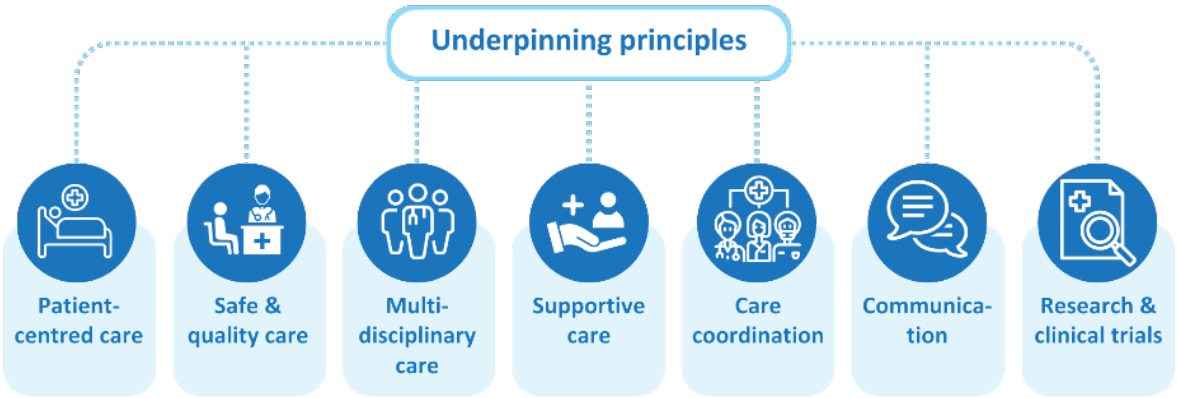
3.3.1 Diagnostic pathways in rare cancer care

Similar to rare cancer diagnosis, RD requires integrated care pathways due to their complexity. Integrated care pathways define the expected course of events in the care of a patient with a particular condition, within a set timescale.[77] They specify the best local standards of care, together with the patient's expected progress. They also provide guidance on the timing and correct sequencing of appropriate investigations and treatment. Integrated care pathways can be effective in improving documentation of progress made in treatment goals and communication with patients, carers and health professionals.[78] The integrated pathway represents optimal care for patients from diagnosis to treatment.

Optimal care pathways (OCPs) are well established in cancer care, promote quality cancer care, define the principles of cancer care and the critical steps in a cancer patient's journey – from prevention and early detection through to end-of-life-care. Currently, OCPs map the care journey for more than 25 tumour types.[79] Following established OCPs is expected to reduce unwarranted variation in cancer treatment as well as improve efficiency, equity and patients' experience of care. Linked routine data sets inform this work. Cancer Council Victoria identified seven principles of the OCP for cancer patients: patient-centred care; safe and quality care; multidisciplinary care; supportive care; care coordination; communication; and

research and clinical trials, which are all applicable to RD diagnostic pathways (see Figure 4 below).[80] Evidence shows that effective implementation of OCPs encourages consistent optimal treatment and supportive care at each stage of a patient's journey.

Figure 4: The seven principles underpinning the optimal care pathways



These principles can be applied to the implementation of the OCPs used for various conditions, especially those requiring complex care.

3.3.2 Diagnostic pathways in rare and undiagnosed disease

As detailed in section 2.3, it is evident that timely and correct diagnosis of RD is required to improve patients' quality of life and satisfaction with care received. A streamlined diagnostic pathway can ensure that:[22, 43, 54]

i. Healthcare providers follow a systematic approach, reducing the time taken to reach an accurate diagnosis.

ii. Individuals receive the appropriate diagnostic tests leading to targeted and effective treatment plans.

iii. Patients are connected with support networks, patient communities, and resources specifically tailored to their condition.

iv. Individuals are enabled to participate in clinical trials and research studies aimed at developing new therapies or improving existing ones.

v. Patients can make informed decisions about family planning, potential risks, and available options for prenatal or preimplantation genetic testing.

These benefits of having streamlined diagnostic pathways were also discussed by the stakeholders. They mentioned that even the process of creating such pathways will help to enhance the collaboration among different medical specialties, research institutions, and patient advocacy groups, fostering a more coordinated and comprehensive approach to RD. Some examples of such pathways are presented below.

Development of national rare disease care pathways, Ireland

A pilot study conducted in Ireland[81] explored the best approach for developing national RD care pathways in the Irish healthcare system. Irish clinical specialists and patient/lived experience experts were involved in mapping existing practice against evidence-based clinical practice guidelines and best practice recommendations from the European Reference Networks (ERNs) to develop OCPs. The study focused on more prevalent, multisystem rare conditions that require multidisciplinary care, services, supports and therapeutic interventions. Overall, 29 RD were selected across 18 ERNs and genetic counselling was highlighted as a core discipline in 27 pathways, demonstrating the importance of access to cinical genetics

services for patients with RD. It suggested care pathways for RD with three main components (see Figure 5 below):

- *Diagnosis:* Diagnostic criteria, common signs and symptoms, red flags for GPs, testing including genetic testing.
- *Care:* coordination of multidisciplinary care, symptoms treatment, prophylactic treatment, health and social care professionals' roles.
- *Core information/resources:* various international and local resources, for example Orphanet, links for each condition, staff resources, clinical lead contacts and centres of expertise.

The study highlighted the importance of collaboration with patient groups besides co-working with clinical leads, peak bodies and researchers in developing care pathways. Several stakeholders also indicated that a collaborative approach to establishing criteria for admission into a genetic service will be beneficial. Note that the consultations for this mapping project highlighted similar opportunities for improving diagnostic care pathways in Tasmania, that is, through education and awareness raising, integrated multidisciplinary care and curated resources.

Figure 5: Components of suggested care pathways for rare disease (adapted from Ward et al (2022)[81])

Timely diagnosis

- Diagnostic criteria
- RD- specific clinical signs and symptoms
- Red flags for GPs/other primary health care professionals
- Lab tests
- Genetic testing
- Other investigations such as ECG, scans
- Disciplines involved in diagnosis

Integrated care (community/clinic/hospital-based)

- Core medical disciplines required for routine management of the condition
- "As required" medical disciplines for complex clinical needs
- Co-ordinating clinical specialty
- Symptomatic screening
- Treatment for symptoms
- Follow-up for at-risk family members
- GP role
- Health and social care professionals' roles
- Psychosocial supports e.g., medical social work, psychology and social care information links

Resource (core information)

- Orphacode(s)
- Orphanet definition of the condition
- Orphanet link for each condition
- Clinical practice guidelines and medical references used to inform the care pathway
- Links to relevant ERN, Australian websites
- Links to ERN or Australian endorsed disease-specific resources
- Staff resources (including linked services) required to deliver the care outlined in the care pathway
- Clinical lead names and Centres of Expertise

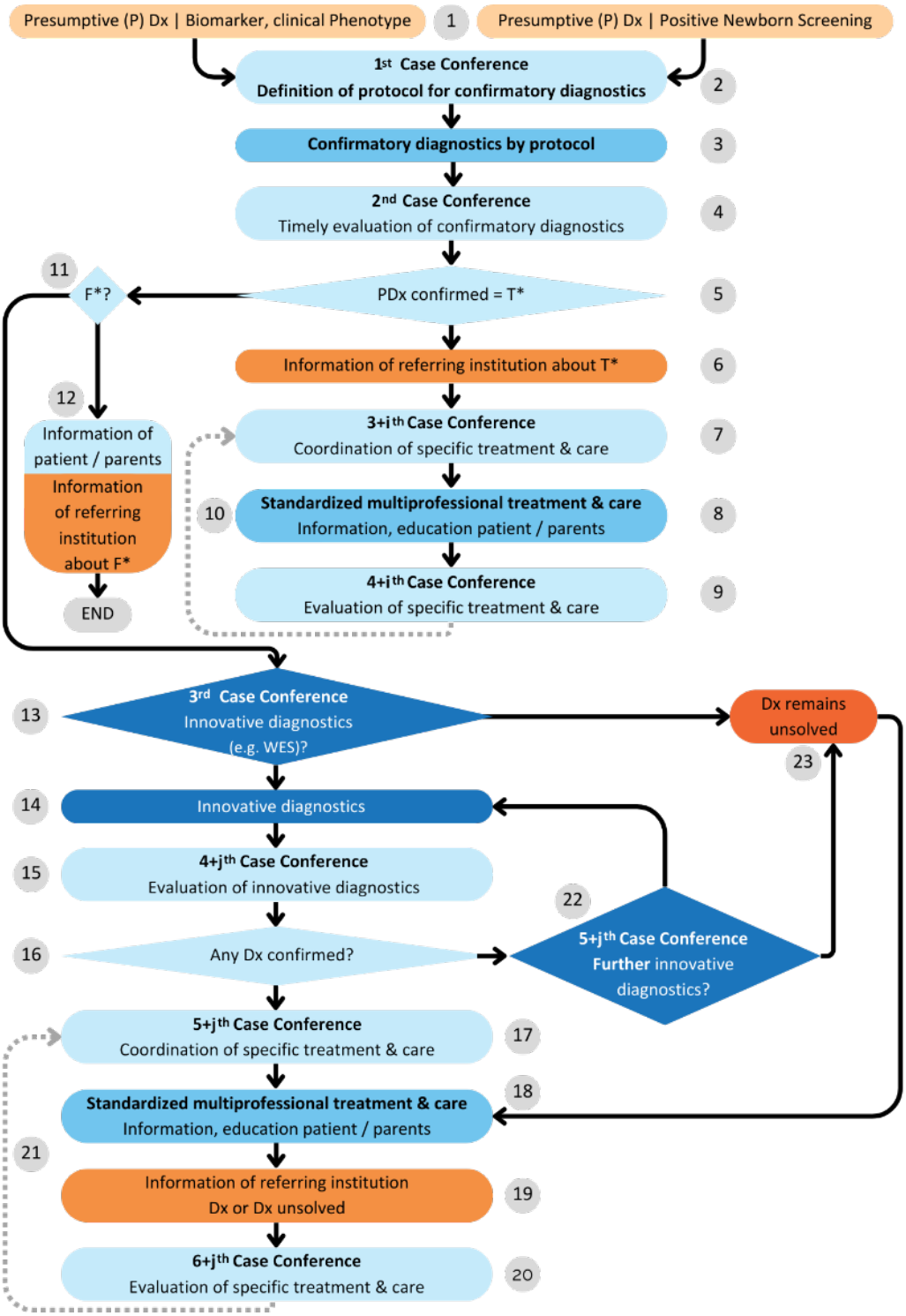
The timely diagnosis component of the care pathways in Figure 5 above identified a range of essential interactions within the diagnostic stage. These are listed below and will be explored for mapping in this project and include the following: [81]

- Newborn screening (routine newborn screening such as cystic fibrosis for neonates)
- Diagnostic criteria for suspected/identified disease
- RD-specific clinical signs and symptoms
- Red flags for GPs for further testing and referral to specialist
- Laboratory tests
- Genetic testing (important testing in RD in particular)
- Investigations (such as ECG)
- Scans (such as CT)
- Multi-disciplines involved in diagnosis (and the availability of experts)

Structured multi-disciplinary clinical pathways, Germany

A German study suggests structured multidisciplinary clinical pathways (CPW) that can guide diagnosis, treatment, and care of patients with RD, linking scientific evidence from clinical practice to clinical outcomes whilst optimising clinical efficiency.[43] The backbone of the generic CPW is a set of multidisciplinary structured case conferences projecting and evaluating diagnostic and/or therapeutic steps, enforcing the integration of best scientific evidence with clinical experience. The generic CPW is documented as a flowchart and a checklist which can be used to record and document the structure, process and results of a patient's pathway, but also as a data model for research. The actual application of the CPW resulted in shortening the days required in establishing confirmatory diagnosis. Median process time from first contact until confirmation of diagnosis by genetic testing was 109 days, much shorter than diagnostic delays reported elsewhere in the literature. Similar generic clinical pathways can be proposed in developing RD diagnostic pathways in Tasmania. These pathways would help health services support patients to find the right diagnosis by applying the best possible diagnostic strategy that would lead to appropriate treatment and care. Existing disease-specific clinical guidelines can be integrated into generic pathways.

Figure 5: The clinical pathway as a flowchart (from Choukair et al, 2021[43])



Such CPWs are highlighted and discussed by the ERN that includes 24 networks with 1200 member organisations. It is acknowledged that as compared to usual CPWs developed for common diseases, the development of RD CPWs presents unique challenges that are the result of the features of RD and a common pan-European organisational culture for RD management is being established. [49]

Clinical Genomics Model of Care, New South Wales, Australia

NSW Agency for Clinical Innovation proposed a clinical genomics MoC to ensure equitable access by patients to NSW genomics services.[82] It identified the four clinical priority areas of Access, Timeliness, Optimising health and Ongoing care. These key areas and processes for facilitating best practices for genomics services can be applied or modified in developing a RD diagnostic model of care.

Figure 6: Clinical Genomics Model of Care[82]

Undiagnosed Disease Program, Western Australia

The Undiagnosed Disease Program in WA (UDP-WA) sits within the statewide Clinical Genetics Service (CGS).[83] The pathway consists of a comprehensive collection and analysis of a patient's entire history followed by a panel review of that history and consultation with the patient and family. The panel review is held monthly, with only one new patient being reviewed.

The selection criteria for the UDP-WA includes:

- 6 months to 25 years old
- Chronic/complex and typically multisystem diseases; multiple specialist assessment/admissions
- Clinical factors support obtaining a diagnosis with current approaches, yet remain undiagnosed
- Assessed by CGS WA already, and known to WA health services, specifically the Perth Children's Hospital, Princess Margaret Hospital for Children and Sir Charles Gairdner Hospital.

Figure 8 below shows the steps involved to get to the diagnosis stage or to refer a patient to the research organisation or other support organisations.

Figure 8 : UDP-WA – Pathway for patient management[83]

GP or Other	Identifies complex case and refers to specialist
Specialist	Numerous investigations; refers to UDP (can be presumed genetic or non-genetic)

UDP (within WA CGS)	
1. Director accepts or declines based on selection criteria	• Where declined, discussion is held with referring specialist around options including research.
2. Registrar initial review	• Comment/reply to referring clinician as required
3. Genetic Counsellor (GC)	• Phones patient/family to discuss UDP referral, UDP program, requirements and consent form • Send info including consent form
4. Clinical Nurse (CN)	• Comprehensive review of all medical records to document a medical history summary • Follow up with family to discuss gaps in history, source privately held records/results and obtain photos as required
5. Registrar subsequent review	• Reviews medical history summary (adds/subtracts as required) • Email medical history summary to Lead for review
6. UDP Panel meeting preparation	• Chair, Lead and registrar prep including slides of differential diagnosis and possible plans
7. UDP Panel meeting	• 45 min: Patient presentation to UDP Panel, feedback and discussion • 15 min: Planning and plan summary with UDP Core Team only
8. UDP Planning and Implementation	• CN contacts family 1-2 days post meeting to provide info on next steps proposed • CN/Registrar arranges all follow up request forms
9. UDP Genetic Counsellor	• Telehealth Consultation (+/- 1 hour duration, 1 week before UDP appointment) • Both parents to attend GC Telehealth Consultation (if possible) • Psychosocial assessment • Discuss the requirement for consent to Genetic Testing for all 3 (MAT, PAT + Proband)
10. UDP Appointment: Lead, Registrar in attendance **11. CN to attend unless not indicated clinically (case by case basis)**	• Registrar to facilitate UDP appointment under the supervision of Lead • Discuss patient medical history using UDP Medical History Summary as a reference • Perform a physical examination and physical measurement as indicated • consolidation/confirmation of patient phenotype as indicated in Medical History Summary • Consent family Genetic Testing - GC will have discussed this briefly with family prior
12. Result Coordination and additional appointments	Registrar • Provide feedback regarding abnormal results as required • Contact patient/family via Telehealth (as directed by Lead) to provide feedback regarding abnormal result if required. • In collaboration with the lead, request additional testing or facilitate research based testing as required/indicated
13. Discharge preparation	• CN prepared Visual Summary/VS draft using UDP Medical History Summary and UDP Panel (one for diagnosed and one for undiagnosed) • Registrar liaise with GPS/GSWA Geneticist regarding discharging the UDP patient back into their care and required follow up • Arrange follow up
14. UDP follow-up appointment at discharge	• Registrar facilitates telehealth appointment under supervision of Lead

The evidence on the key determinants of RD diagnosis, the examples provided above that identify various components of RD diagnostic pathways and the stakeholder consultations (Appendix 2) provide a comprehensive picture of the key considerations in developing MoC for RD. These are discussed in the section below.

3.4 Key considerations and challenges identified

A RD impact report illustrated that it takes on average 5.6 years to obtain a diagnosis for a RD in the UK, and 7.6 years in the USA.[5] This 'diagnostic odyssey' has been recorded around the globe, including in Australia, with a lack of diagnosis being a major barrier to accessing care.[5] Many barriers to timely RD diagnosis are discussed in sections 3.2 and 3.3. With respect to Tasmania, a summary of the main challenges as found in literature and reported during key informant consultations thus far are listed in Table 2. These are further detailed in the community needs assessment (Chapter 4).

Table 2: Evidence on challenges gathered from literature and key experts in the field

Patient level
Inequity of access due to geographical, educational or any other socio-economic disadvantage
Unclear diagnostic process and associated fear/confusion about genetic testing: patients
Isolation and stress felt by the families due to RD diagnostic odyssey
Navigation of complex health system
Primary care level
Lack of suspicion of a rare disease
GPs feeling overwhelmed due to limited knowledge and understanding
Lack of knowledge of easily available and accessible information for GPs
Unclear diagnostic process: clinicians
System level
Limited ability to establish MDT due to limited availability of specialists across Tasmania
Disproportionate workforce distribution across Tasmania
Issues with recruitment and retention of health professionals critical in caring for RD
Specialty shortages, including geneticists, in Tasmania
Transition of care including during diagnosis from paediatrics to adult health care system
Lack of epidemiological data that can help identify gaps in knowledge and allocate resources for developing diagnostic techniques and treatments.
Related to genetic testing
Funding of genetic testing
Lack of genetics and genomics skills across the workforce
Difficulty in accessing diagnostic testing due to out-of-pocket costs, limited availability of testing, lack of awareness amongst healthcare professionals and/or need to take time away from employment for carers

4 Community needs assessment

4.1 Introduction

The focus of the community needs assessment via consultation with stakeholders was to identify gaps in the rare disease diagnostic pathway in Tasmania, gain a detailed understanding of the barriers to RD diagnosis in Tasmania and identify targeted solutions and strategies to overcome these barriers. This chapter presents the findings of the stakeholder consultations. A total of 1020 stakeholder consultations were conducted across three stages.

Table 3. Summary of stakeholder consultations

Stage/Process	Level of engagement
1. Situation Analysis	– 6 key informant interviews
2. Surveys	– 880 patient/carer survey responses – 31 clinician/researcher/advocacy organisation survey responses
3. Interviews and focus groups post surveys	– 15 clinician/research interviews – 11 individual patient/carer interviews – 63 patient/carer focus group attendees – 5 individual Community Reference Group member interviews – 9 individual advocacy and patient group interviews.

Through the focus groups and interviews we were able to record the diagnosis for 74 participants. Whilst not suggesting this is representative of the prevalence of RD in Tasmania, the categories[21] were:

Table 4. Categories of rare disease of participants

Category of rare disease	Number
Rare neurological disease[22]	17
Rare systemic and rheumatological disease	9
Rare genetic disease	8
Rare inborn error of metabolism	7
Rare neoplastic disease	6
Rare haematologic disease	5
Rare respiratory disease	4
Rare skin disease	4
Rare bone disease	3
Rare circulatory system disease	3
Rare endocrine disease	2
Rare gastroenterologic disease	2
Undiagnosed	4
Total	74

[21] Approximately 80% of respondents have a genetic condition which is consistent with the reported frequency of genetic forms of rare disease. Rare genetic was chosen as the category for those with a complex multisystem genetic disorder.
[22] Rare neurological diseases are known to be overrepresented in rare disease diagnoses, therefore 17 out of 74 is not unexpected.

4.2 Needs assessment findings

Information and perspectives gathered during the stakeholder consultations have been grouped into three overarching categories: 1) health professionals, 2) advocacy, support, and information, and 3) health system. Within each category the findings are presented under themes, noting barriers, enablers, and suggestions for consideration.

Where relevant, findings from the surveys undertaken with patients/carers and clinicians, researchers and advocacy organisations are integrated into the following analysis.

4.2.1 Health professionals

The following section describes the findings that relate to access to, knowledge of, and understanding and attitudes of health professionals.

4.2.2 Access to multi-disciplinary team

Diagnosis, treatment, and care of patients with RD requires multidisciplinary collaboration between medical and paramedical specialties and with patients and families,[13, 43] requiring more complex care pathways. A limited availability of specialists within Tasmania results in few formal MDT approaches or independent multidisciplinary input in patient diagnosis and care. In some cases, MDTs in Victoria and other Australian jurisdictions (often based on the focal point of a RD) support Tasmanian health professionals in clinical decision making and planning the next steps in a patient's diagnostic journey.

All stakeholders consulted agreed that an MDT approach is needed in Tasmania and where patients have had this opportunity they have experienced better diagnostic and healthcare outcomes. A summary of key findings relating to MDT is provided below.

- Citing that the Australian health system is complicated and difficult to navigate, GPs highlighted the importance of having access to an MDT to refer to, especially for people with RD. This is all but non-existent in the Tasmanian context for someone with an RD. For the same reason it was suggested that a *'single point of care'* such as a Rare Care Centre is required for people with RD. This would ideally include a navigator role.

> *"Multidisciplinary team is critical in clinical genetics. Accurately interpreting a genetic test requires MDT."*
> (Researcher/Clinician)

> *"Patients end up with various specialists who don't see the whole picture – which is important in the role of a GP. For GPs, navigation of the system is the challenge, as the GP's best option is to refer to various specialists and write multiple referrals. GPs rely on specialists to be coordinated/communicate to work out what's the best option for their patient holistically and therefore a multi-disciplinary team approach is critical"*
> (General Practitioner)

- In line with the health professionals' view that having MDT care available is critical, patients and carers provided the following feedback:
    - A multidisciplinary specialist clinic for RD is needed to avoid siloed medical care, for example, following the neurology path, then the endocrinology path, then back to the neurology path.

- Where access to a MDT during the diagnostic journey or for treatment and management has been available, this has made a significant improvement to the patient's journey and outcomes (expediency and quality).

> *"With the new complex respiratory team commencing [in Tasmania], my experience with the system has never been better and my child is getting much better outcomes."*
> (Parent)

> *"What I have been able to organise is a monthly meeting of my specialist, GP and myself. This has made a big difference to my management."*
> (Person with a rare disease)

4.2.3 Access to general practitioners

GPs are critical in rare disease diagnosis, especially in the Australian healthcare system where GPs function as gatekeepers to specialists.[59] Many patients with RD will present to a GP with their symptoms as a first step. They will also see their GP in between visits to a specialist, and for a range of other primary and preventive care services. This ongoing relationship that also has an accessible, relationship-based advocacy and support role can be a foundation for positive experiences for patients with RD.[60]

Local access to general practice, allied health and community nursing services is worse for people living outside urban population centres in Tasmania. Communities often rely on visiting services, which present challenges in delivering continuity of local primary care.[38] Attracting and retaining healthcare professionals, including doctors, nurses, and allied health workers, has been a persistent challenge for Tasmania.

The issue of access to GPs was highlighted by stakeholders during the consultations and a summary of key findings provided below.

- A shortage of GPs in Tasmania was identified as impacting a patient's diagnostic journey, and this is amplified in rural areas, particularly in North West Tasmania. The impact of GP shortages identified include:
  - GPs are under pressure to see a large number of patients each day and do not have sufficient time to spend with patients
  - There can be long waiting times to get an appointment with a GP, for example, 6 weeks
  - Some GPs have 'closed their books' and are not accepting any new clients
  - Multi-GP practices means that some patients accept the next available appointment with a GP in their practice rather than their 'regular' doctor, impacting continuity of diagnostic journey or management
  - Many GPs are locums, also impacting continuity of care.
- Consistency in seeing the same GP in a practice was indicated to be necessary to build trust and for the GP to know and fully understand the patient. When a patient is well known to a GP before the RD onset, the diagnostic journey is easier as the GP is able to compare a patient's symptoms and condition over time. In cases where there was no or less continuity, some patients felt dismissed by their GP as they struggled to "put the story together". Not having a "family GP" anymore was noted as a possible reason why patients' diagnostic journeys are extended.
- Locum GPs were reported to be both a barrier and enabler by patient/carer participants.

- As above, with limited understanding of patient history and no continuity of care, patients reported having to repeatedly explain their situation and medical history every time they see a new locum. The opportunity to build a relationship and journey with the doctor was negated.
- Conversely, the arrival of a locum GP had positive outcomes for some patients. Some came with a fresh approach and were considered to be more open to greater exploration of the presenting symptoms (compared to the view that a long-standing GP had become dismissive and was viewing the patient as psychosomatic). Additionally, because locums (or in fact a new GP in the practice) frequently come from outside Tasmania, some participants noted the locums have had broader experience, seen more RD and were open to further investigations.

Patient/carer participants made the following recommendations:

- Patient/carers fully understood the challenge in attracting and retaining GPs in Tasmania; however, they want to see Government make continuing efforts to increase the number, particularly in regions where there are significant shortages.

4.2.4 Access to specialists

Tasmania has identified shortages in specialties such as psychiatry, emergency medicine, general surgery, and some allied health professions. These shortages can impact the provision of specialised care and lead to longer than clinically recommended waiting times for patients.[37, 38] Stakeholder consultations identified that there is limited system capacity in RD diagnosis to support the needs of Tasmanians, which primarily relates to shortages and poor access to the specialised clinical workforce, such as laboratory staff, geneticists, genetic counsellors, paediatricians, neurologists, rheumatologists, and other specialists. This has led to long wait lists for diagnostic testing.

These issues of access to specialists were highlighted by stakeholders, with key findings including:

- It should be firstly noted that many patient/carers fully understand the challenge in attracting and retaining specialists in Tasmania and particularly specialists with specific knowledge of RD.
- Patients/carers commonly raised the limited availability and subsequent extensive waiting time in Tasmania for neurology, paediatrics and rheumatology specialists. Wait times of 10 months from booking an appointment to the first appointment were shared by participants.
- This was also a key finding in the patient/carer survey where 20% of respondents (78/397) noted difficulties in accessing a medical specialist and/or long wait times as a major challenge. In addition, 40% of respondents (169 of 420) in the patient/carer survey identified the need to travel interstate as part of their diagnostic journey.
- Limited specialists in Tasmania means patients cannot seek a second opinion. This is exacerbated by specialists practising in both the public and private sector and therefore there may only be one specialist available, and many specialists are unwilling to review or re-diagnose a diagnosis that was made by that specialist's medical teacher or mentor.

> *"The registrars are more likely to listen and follow it up – but then they get shut down by the experienced specialist."*
> (Patient/Carer)

- Access to geneticists in Tasmania was identified by participants as limited and with long waiting times. However, it is telling that this was not raised by patients/carers to anywhere near the

degree as was the lack of access to other specialist services, potentially as a result of GPs not routinely thinking of a genetic condition as a possible explanation for the presenting symptoms.

- Patients/carers identified that interstate travel required when specialists are not available in Tasmania has associated challenges such as costs, time away from work (sometimes at a cost) and isolation from family and friends during times of extreme stress.
- The transition from paediatric to adult specialists was identified as a barrier to continuity of care by patients/carers. One participant stated that they had been referred to a paediatric specialist at age 13 and due to extended waiting lists was seen for the first time in the month prior to their 18th birthday. The paediatric specialist was then unable to treat the patient further and referred the patient on to an adult specialist. A further two patient/carer survey respondents identified the transition between children's and adult health services was a key challenge during diagnosis.
- The COVID-19 pandemic was considered by patient/carer participants as both positive and negative in relation to accessing specialists. Participants said that COVID-19 led to specialists offering telehealth appointments, and many participants said this approach has continued. Telehealth alleviates fatigue associated with travelling for multiple and regular medical appointments, and reduces costs for patients, all of which were viewed positively by participants. However, not all appointments can be done via telehealth, and some patient/carer participants stated that COVID-19 brought a stop to specialists travelling outside Hobart and patients now need to travel to see specialists.

Patient/carer participants made the following recommendations:

As noted above, many patient/carers fully understand the challenge in attracting and retaining specialists in Tasmania and particularly specialists with specific knowledge of RD and on that basis called for:

- Continuing efforts to increase access within Tasmania through employing more specialists
- Increased access to more specialists in Tasmania through telehealth appointments
- Ensure support is provided to access specialists in other jurisdictions for people with an RD and this may include changing PTAS eligibility criteria to ensure access to the most relevant specialist (e.g., in private practice) for the respective RD.

4.2.5 Knowledge of general practitioners and specialists

Many GPs have reported feeling overwhelmed when caring for RD patients.[61] For a positive interaction and timely diagnosis, practitioners require sufficient knowledge and information on testing and referral options.[61] GPs face a difficult task when caring for people with diagnosed or undiagnosed RD and the need for training, development, improved communication, and better awareness is highlighted in the literature.[5, 61] GPs caring for children and adults with RD have a crucial role in making appropriate referrals, providing care co-ordination, and linking families to psychosocial and other forms of support. It is important, therefore, that GPs are aware of information portals and educational resources that will assist them to help patients with an RD.[5]

The key to early diagnosis is also the specialist's knowledge of RD. Greater collaboration among GPs and specialists with expertise in RD may help to expedite the lengthy process to a correct diagnosis. Linking potential symptoms together rather than treating just one individual symptom may help to reduce the time to diagnosis.[63]

Specific feedback from stakeholders on GPs' and specialists' knowledge of rare disease include the following:

  - Clinicians indicated that certain RD, like Huntington Disease, are not that rare in Tasmania, meaning GPs are more familiar with it and can seek a diagnosis for their patients.
  - Depending on symptoms, many GPs start with the HealthPathways portal. It was indicated by health professionals that GPs in Tasmania use HealthPathways extensively. The key was to have knowledge of what's available and where. When asked about addressing the barriers in the diagnostic pathway the clinicians indicated:
    - Having a dedicated referral pathway
    - Increasing awareness among practitioners regarding when to refer on
    - For GPs, knowing about referral criteria is critical. Inappropriate referrals happen, hence referral criteria needs to be clear and communicated well.
- A key challenge in general practice is to have access to RD resources in one place and that they are easy to navigate.
- Some barriers for GPs were identified by health professionals as:
  - Need to go through the diagnostic process and narrow down the diagnosis but at some point, it is about understanding that the patient needs a referral and to whom to refer to. If symptoms are showing multiple organ involvement, it becomes more complex
  - The time pressure GPs experience means they may not have time and scope to even consider that genetics may be relevant.
- Patient/carer participants indicated that GPs and specialists do not understand RD. It is important to note that overwhelmingly, patient/carer feedback was that they did not expect GPs (and to some extent local specialists) to have knowledge of or consider a specific RD where they have never seen that RD previously (although they do expect them to consider a genetic condition might be present).
- Participants stated that GPs and specialists alike look for big symptoms rather than small symptoms and look for common and known symptoms.
- Further barriers to diagnosis were identified by patients/carers as:
  - GPs do not conduct thorough investigations because they do not understand the presenting symptoms.
  - GPs do not know where to refer patients for diagnosis, testing and specialists.
  - Patients are misdiagnosed with other conditions or with other RD due to lack of knowledge.
  - Patients remain undiagnosed due to doctors' lack of knowledge.
  - Co-morbidities are not monitored and managed.
- The patient/carer survey had similar findings, with lack of knowledge and awareness being the most commonly cited challenge experienced by survey respondents (excluding out-of-pocket costs). Of 310 responses, lack of knowledge and awareness among health providers was mentioned 88 times (approximately 28% of respondents).

> *"We hear about more medication options than the specialist does. The neurologist is knowledgeable but can't get hold of the medication we want to try."*
> (Patient/Carer)

- Participants, researchers and specialists all acknowledged that GPs cannot know everything about every RD. Patients mentioned that as long as a GP was willing to listen, do some research and keep exploring, then that was acceptable to them. Researchers/specialists indicated that GPs are required to have awareness of what is available and be able to refer to help with diagnosis.

"My GP described my situation as being 'odd' but that was where he left it. I think he just didn't know what to do and so just managed symptoms."
(Patient/Carer)

- Parent/carer participants identified that the level and continuity of care for RD was a very different and negative experience compared to the care received for other conditions such as asthma and maternity care.

"If the disease is rare, it doesn't mean we are not significant. Our lives are no less important than those who have diabetes or breast cancers. We also have families, children and that needs to be considered."
(Patient/Carer)

Patient/carer participants made the following recommendations:

- Raise awareness of GPs about how and where they can readily access information on RD from a range of sources.
- Establish a central body of RD information in Tasmania where GPs can call in.
- Raise awareness of GPs (e.g., through information, training, checklists) that an RD may be possible, the signs that a condition might be an RD, as well as the next steps to be taken when an RD is suspected.
- Increase training/awareness among GPs and specialists to increase their understanding of RD and their diagnosis.

4.2.6 Understanding and attitude of health professionals

The journey to identify an RD can be emotionally and psychologically taxing for patients and their families. It often involves multiple doctor visits, inconclusive tests, and uncertainty. The rare nature of the condition may lead to feelings of isolation and frustration. Complexity of rare conditions makes service navigation by families difficult. Research has identified that families affected by RD represent a medically disenfranchised population that falls through the cracks of every healthcare system in the world.[5] The Australian Paediatric Surveillance Unit indicates that people living with RD face significant challenges, including diagnostic delays, lack of available treatment and difficulty in finding the right health service. Families feel isolated, under-supported, and often face economic hardship.[74]

This was reinforced through the consultations where stakeholders cited that families often feel isolated, forgotten and fearful in their diagnostic journey. The TCGS CRG emphasised the importance of individuals knowing what to expect from a consultation with a practitioner.

There appears to be a strong relationship between the knowledge of GPs and specialists and the experiences and perceptions of patients, families and carers in their interactions with these health professionals when seeking diagnosis of an RD.

Specific feedback from patient/carer participants on the understanding and attitude of health professionals included the following:

- A doctor (GP or specialist) or other health professional who is willing to listen to the patient and to push for additional tests or additional referrals was considered extremely helpful to patients and carers. It was noted that this experience is the exception rather than the rule. There was a sense that the GP or specialist 'gives in' and reaches some diagnostic conclusion too early and this is proven to be the case later.

- Patient/carer participants overwhelmingly reported that they are not listened to and not believed by doctors. Participants stated that there is a 'doctor is the expert' mentality that means doctors cannot be challenged. Examples provided by patient/carer participants included:
    - Participants had to advocate hard to get genetic tests ordered. They said that doctors attribute symptoms to environmental factors and will not investigate genetics, even when there is a family history of a disease.
    - Participants want a diagnosis because it helps them; doctors do not always understand the value to patients of having a genetic diagnosis, particularly if there is no treatment available following diagnosis.
    - When a patient has co-occurring diagnoses, doctors focus on the existing or non-rare disease rather than new/additional symptoms. Further, when patients with an RD raise new symptoms with their doctor, they are told that the new symptoms are related to something else and not related to the RD.
    - People with RD are labelled as 'drug-seeking' when they ask to change medication or request more medication for pain management.

> *"Doctors tell me to 'take a big breath and it will go away'."*
> (Patient/Carer)

- Allied health professionals were identified by patient/carer participants as caring and willing to listen and to follow up when doctors were not willing to progress investigations and testing.

> *"One consultant treated us badly, they said I was hysterical and told me to 'spend more time mothering and less time reading PubMed'."*
> (Patient/Carer)

- The patient/carer survey also identified that a key challenge in their diagnostic journey was the lack of mental health/emotional support offered by providers and services.

> *"The specialist asked why I wanted a diagnosis – can you believe that?"*
> (Patient/Carer)

- Patient/carer participants who accessed the genetics service in Tasmania reported both positive and negative experiences. Positive experiences included timely, helpful, and thorough assistance. Negative experiences included being given a diagnosis without any accompanying information, support or referral; and requesting but being refused a genetic review.

> *"I had to do the research myself. I had to talk the specialists into believing me."*
> (Patient/Carer)

Patient/carer participants made the following recommendation:

- Patients/carers consider the dismissive attitude of GPs and to some extent specialists is largely related to limited knowledge and understanding of RD, as described above. Whilst they shared a view that training and education will assist to improve doctors' attitudes towards people

affected by an RD, they also consider that training should include the range of impacts that they experience.

4.2.7 Advocacy and support networks

Research conducted by researchers on RD care pathways in the European Union (EU) reinforced that the patient organisations and resource centres for RD have a special role in patient empowerment and self-management, and this needs to be further promoted.[49] In Australia, advocacy and support is an integral part of the the National Strategic Action Plan and RVA is a strong advocate for patients, but feedback suggests more is needed. Specific feedback from stakeholders on advocacy and support networks included the following:

- Support networks for RD are limited and lacking. Many patient/carer participants spoke of family and friends providing support, which can strain relationships. Aboriginal health services were identified by some participants as a major support, particularly for providing travel assistance to appointments. Others stated that they have no support people around them.
- The specific types of support mentioned by patients/carers were: emotional support, mental health support, transport to and from appointments, house and garden cleaning and maintenance.
- Personal circumstances impact the diagnostic journey of patients/carers. This includes social circumstances, such as family, friends and other supports available; resilience and a willingness to ask questions and challenge decisions; and health literacy, an understanding of how the health system works and where to find information.
- Social media platforms such as Facebook groups were identified by patients/carers as being a major source of support. Online support groups were preferred when participants were unable to leave their house due to mobility, transport, or anxiety. Online support groups were also preferred because other support groups require paid membership, which may be unaffordable.
- RVA was identified by participants as an organisation offering limited information and services. Patients/carers stated that RVA provided information on their specific disease when contacted, but also that RVA advised them that they (the individual) would need to become their own advocates; participants considered this an unsatisfactory response. Participants also identified that there are limited RD associations operating in Australia, and they are mostly based in Sydney or Melbourne and do not operate in Tasmania.
- Participants who joined focus groups spoke of the benefit of having the opportunity to share with people experiencing the same broad issues despite having a different diagnosis. The focus group facilitators noted attendees sharing strategies which they had used to overcome barriers and, in some instances, sharing of contact details. The need for a Tasmanian RD support group was suggested. Ideally this group would have an advocate that would contact a patient once a diagnosis has been made, to advise what to do next and who to contact and their contact details, for example social supports, occupational therapists for neurological conditions.

Patient/carer participants made the following recommendations:

- Mental health support is essential for patients and carers and family members.
- Support to access National Disability Insurance Scheme (NDIS) is needed.
- A broad RD support group with accessibility across Tasmania is needed. This would be established through government funding in the first instance and then responsibility could be assumed by the membership.

4.2.8 Access to information for people impacted by rare disease

Due to their rarity, many RD have not been the subject of significant scientific research and therefore there is limited information available on the underlying causes, mechanisms, and clinical presentations of these conditions. This scarcity of information makes it difficult for healthcare professionals to accurately diagnose an RD.[58] It also makes it difficult for patients and carers to access accurate, current, and locally relevant information.

During the stakeholder consultations, access to information about RD was identified by participants as a major challenge.

- Social media platforms such as Facebook groups were identified by patient/carer participants as being a major, or only, source of information about a RD. Online groups were also identified as a source of information about health professionals and for "finding out about good doctors".

> *"I go to the Facebook page because I'm not getting the information I need from doctors."*
> (Patient/Carer)

- International information and advocacy organisations in the UK and USA were identified as useful for providing information on specific conditions and RD. While the information about the condition was useful, the information about diagnostic and treatment options and advocacy services was not relevant as it was different from the Australian context.

Patient/carer participants made the following recommendations:

- Treat a RD diagnosis the same as a terminal diagnosis. That is, provide patients with access to a counsellor or social worker, provide information about the diagnosis and available support symptoms, and information on the likely impact of the diagnosis on the patient's and carer's mental health.

> *"I didn't know it was a rare disease and didn't know the impact it would have on my life. No one sat down and explained how my life would be different. I was told later that they did that on purpose. I lost my relationship, my house, friends, and family all due to my illness but that could have been prevented if the diagnosis had been given to me in a different way. I could have had different conversations with family, friends, and my workplace."*
> (Patient/Carer)

4.2.9 Navigation of health and psychosocial health systems

RD are often complex, serious, chronic and progressive conditions. Treatment of a rare disease often involves MDTs, including a variety of clinical, nursing and allied health specialists, including a geneticist. The system-level challenges to service provision in the Tasmanian context include overall low health literacy, rurality and socioeconomic status.

Problems in accessing and navigating the health system in Tasmania were highlighted by stakeholders during the consultations. Difficulties navigating the Tasmanian health system were also reported in other states. Specific feedback from stakeholders included the following:

- A general lack of clear pathways within the health system, and lack of support to navigate the health system, were reported by patient/carer participants as a challenge to their diagnostic journey.

- The importance of health literacy and persistence were emphasised by patient/carer participants. When asked what their journey might have looked like without health literacy, a common response

was “impossible”. Further, they stated they have become experts in their condition out of necessity, because there are no other experts available.

- Communication and coordination were identified as critical but needing improvement by clinician and patient/carer participants alike. Clinicians reported that improving care co-ordination by bringing health information and health professionals together is important to improve patient care. This challenge was also identified in the patient/carer survey with responses citing lack of coordination and communication among providers as a major difficulty in their diagnostic journey.

- Communication by health professionals was considered by patient/carer participants as a barrier within their diagnostic journey, in a number of ways:
    - Lack of communication between health professionals meant that participants had to share their story repeatedly with different health professionals. This was noted as a particular challenge when patients saw health professionals in other jurisdictions and regulations prevented sharing patient information across jurisdictions.
    - Lack of communication by health professionals with patients was frustrating. For example, doctors not providing information to patients and not setting realistic expectations of timeframes for receiving test results.

- Patients/carers identified that feedback and complaints mechanisms about health professionals and health services require strengthening as currently clear pathways are not readily available. One patient provided an example of receiving a response to their complaint after two months.

- Access to two parallel health support systems was discussed by patient/carer participants: the NDIS and the Patient Travel Assistance Scheme (PTAS).
    - Access to NDIS was identified as a major challenge by patient/carer participants. Many participants stated that they have been unable to apply for NDIS assistance because they do not have a diagnosis. Other participants stated that they have been able to access NDIS without a diagnosis. Participants also thought that assistance to apply for NDIS was essential. For example, completing application forms, understanding what paperwork is required for an application and where/how to get the paperwork. Where people have been able to access the NDIS, they have described this as a ‘game changer’.
    - Patient/carer perspectives on access to PTAS were varied. Many participants welcomed and appreciated the financial assistance for interstate travel provided by PTAS. On the other hand, the effort required to apply for PTAS was not seen to balance with the financial assistance received, and PTAS did not cover the true costs involved in medical travel. For example, PTAS may subside the airfare but not accommodation, food and other living costs. Some participants reported PTAS offering airfares for a child but not a parent, and in some situations, this meant the child could not travel because the parent could not afford the travel.

> *“PTAS won’t pay because they want people to see specialists in Tasmania, but there aren’t any here, or if they are here, they have long wait lists.”*
> (Patient/Carer)

Patient/carer participants made the following recommendations:

- Health professionals must use My Health Record consistently and thoroughly to enable continuity of care.
- MDTs are needed to aid in communication between health professionals, make the situation less complex for the patient and family/carer and shorten the diagnostic journey.

- Health professionals should provide information to new patients on pathways for providing feedback or making complaints.

4.2.10 Timeliness

Many RD are not well-known, even among healthcare professionals. The rarity of these conditions means that they are less likely to be encountered in routine medical practice. As a result, healthcare providers may not consider them initially, leading to delays in diagnosis. Unnecessary consultations cause substantial costs for the individual and for healthcare systems; before the correct diagnosis is made, patients see an average of 7.3 physicians.[56]

It is reported that 30 per cent of Australian adults living with a rare disease are impacted by a diagnostic delay of more than five years, while almost half have received at least one misdiagnosis. Both diagnostic delay and misdiagnosis can negatively impact the experience of care and support received by individuals.[2]

Specific feedback from stakeholders on timeliness included the following:

- Waiting for a diagnosis was identified by patient/carer participants as having a significant impact on their health and wellbeing, specifically the mental health burden, and feeling in limbo, while waiting for test results or waiting for specialists, and being unable to make plans. For participants without a diagnosis in the long-term, there are feelings of helplessness and shame. In addition, participants said they could not be referred to other health professionals and support networks without a diagnosis.

- As detailed earlier in these results, waiting times for health services have impacted participants' diagnostic journeys. For example, the wait time for:
    - an appointment with a GP was reported to be up to 6 weeks.
    - an appointment with a specialist, once referred, could range from 10 months to five years.
    - the completion of genetic tests once the test had been ordered was reported as two to three months.
    - receiving test results was reported to be between two and 14 months.

- Clinicians/researchers also indicated that the diagnostic delays are common for RD and as genomic technologies become more available and are lower in cost, it is putting stress on laboratories.

- Clinicians/researchers identified that the timing of reproductive carrier testing is too late when a child's diagnosis is made. An example was a patient who had two children with a RD as a result of the first child being misdiagnosed, leading to a delayed RD diagnosis, and therefore reproductive testing was not conducted in the second pregnancy.

Patient/carer participants made the following recommendation:

- Health professionals to be open and honest about wait times to allow the patient to decide if they will wait or go private or elsewhere.

> *"With a formal diagnosis you get help and legitimacy. Having a label is a positive thing."*
> (Patient/Carer)

4.2.11 Out-of-pocket expenses

The financial costs of the diagnostic odyssey may be high and unaffordable. Pursuing an accurate diagnosis can involve numerous medical tests and consultations. Additionally, without a diagnosis, health insurance might not cover specific treatments or therapies, leading to increased financial strain.

Out-of-pocket expenses were identified by patient/carer participants as impacting on their diagnostic journey. A summary of the findings is given below.

- Some participants indicated that having to pay for tests that incur out-of-pocket costs could be a deterrent to getting a test done.
- Participants identified the following examples of out-of-pocket expenses:
    - Pathology and having samples taken
    - Genetic tests e.g., $3400
    - Travel on multiple occasions, for example, the first trip to get tests done and then a return trip to receive the results
    - Travel to get to a specialist and to the geneticist, as the specialists are located in Hobart, Launceston and/or interstate
    - Medication costs, in particular compounded medications
    - Specialists' fees. One participant reported seeing 15 specialists so far in the diagnostic and treatment journey.
- Patient/carers identified that out-of-pocket costs during the diagnostic journey required significant financial outlay for many patient/carers. Approximately 42% of respondents (152 of 358) had out-of-pocket expenses of more than $2,000, and 8% of respondents (28 of 358) had incurred out-of-pocket expenses over $10,000.

Patient/carer participants made the following recommendation:

- Provide financial support that recognises individual circumstances (e.g., PTAS eligibility criteria need to be changed to ensure access to the most relevant specialist for the respective RD for private practitioners in other jurisdictions [i.e. in private practice]) and the fluctuations in symptoms (challenging to access NDIS when symptoms are intermittent).

"Radiology now provides a log that we can access of how many scans we've had – it was surprising to see how many scans I've had - they are all expensive and some I feel were not necessary; the specialists don't consider our financial position for affordability, and they need to do more comprehensive but fewer scans to reduce costs."
(Patient/Carer)

"I've been really lucky with the Aboriginal Health service, some of the costs are covered; but they do have the long wait lists."
(Patient/Carer)

"I went ahead and got private health insurance when the diagnostic journey started but had the 12 months wait before we could claim any benefits; so seeing both public and private. we have hit the Medicare threshold and still out of pocket a large amount of money."
(Patient/Carer)

4.2.12 System and process improvements

Clinician and patient/carer participants alike identified system and process improvements that would enable more efficient and effective diagnosis of RD. A discussion of these system and process improvements and associated recommendations are listed below.

- **Systematic data gathering**: Clinician participants identified that systematic data gathering on RD and the use of registries would have several benefits. At the patient level, greater data repositories could be used for quicker diagnosis in the future. At the epidemiological level, increased and improved data could be used as evidence to attract funding.

- **Access to diagnostic tools:** Patient/carer participants stated that greater availability of diagnostic tools in Tasmania would have helped to detect and address their condition much earlier. Some participants sent samples to interstate and international laboratories and in several instances the samples or the results were misplaced, leading to extra costs, extra time delays, and unnecessary worry. Other examples provided were: MRI not being available in Tasmania; and a 4-day wait for a test kit to be sent from Melbourne to Hobart. Clinician/research participants stated that genomics is a specialised resource and the teams do not necessarily need to be location bound and acknowledged that the process of sample transfer and reporting could be improved.

- **Funding for rare disease research, diagnosis, and treatment:** Clinician/researcher participants indicated that the size of the Tasmanian genetics service is an issue as there are not enough clinicians to meet community needs, and there is a lot of fear around genomics and what it means. Additional funding could be allocated to expand genetics services, to create a system that helps to improve patient understanding of genomics, and to train more geneticists and to upskill general paediatricians and other health professionals to be more genomics literate. Specialists that have been trained more recently have greater genomics literacy and are better able to suspect a genetic condition.

4.3 Summary of findings

A summary of key findings is presented here.

What We Heard	
What makes diagnosis challenging?	**What helps diagnosis go well?**
– GP shortage (especially North West, rural) & limited capacity – Limited knowledge/understanding of RD among doctors (noting no expectation from patients/carers that doctors can know everything) – Lack of listening & empathy from many doctors – Limited access to local specialists & diagnostic tools – Long wait times for specialists (e.g., neurology, paediatrics & rheumatology) – Impact of interstate travel (financial, work, family/friends); some having to relocate – Moving from child-specific to adult health services – Lack of support to navigate & understand service system, including genetics service – Poor communication & co-ordination from/between care providers – Financial barriers (out-of-pocket expenses).	– Strong trust with a consistent GP – GP has experience with RD – GP listens and is committed to finding a diagnosis – MDT approach – Caring allied health professionals – Emotional/mental health support from family/friends, others with RD – Practical support (e.g., transport, PTAS) – Patient/carer health literacy and commitment to finding a diagnosis – Coordinated support to navigate and access health system and associated supports

What Patients and Carers Said They Need	
• **Health professionals who** – listen – consider all symptoms, whole context, family history – research and explore the condition – investigate and refer for testing/specialist review as needed – communicate well and regularly – communicate and coordinate with other health professionals.	• **Less time** between testing and diagnosis • **More information** about RD • **More support** for RD through patient networks/groups • **More assistance** to access health services and social/disability supports, including access to NDIS – e.g., advocacy, referrals, support networks within Tasmania.

4.4 Dissemination of findings

A summary of the findings of the stakeholder consultations was shared with all participants who provided contact details in the form of a two-page summary of this chapter.

All qualitative information gathered via the consultations along with the survey data was assessed against the current diagnostic pathways established by other countries or states in Australia, and used to map the current diagnostic pathway/s for RD in Tasmania (Chapter 5), and to develop innovative models of diagnostic care for people with RD in Tasmania (Chapter 6).

5 Current diagnostic pathways in Tasmania

The diagnostic journey for a person with RD is characterised by multiple touchpoints within the health system. Health system touchpoints can refer to the various interactions or points of contact between individuals and the healthcare system. These touchpoints encompass a wide range of experiences, including visits to healthcare providers, hospitals, clinics, or interactions with healthcare professionals via telemedicine or digital platforms. They can also involve administrative processes such as scheduling appointments, obtaining test results, receiving medical advice, or accessing health-related information. This diagnostic journey of a patient with an RD is depicted in Figure 9 below.

From examining global evidence, reviewing the current pathways in Tasmania, and consulting with stakeholders, the following touchpoints were identified: General practitioners, medical specialists, MDT, rare disease specialists and geneticists, investigations, and research/clinical trials. See Table 5 for the details regarding these touchpoints, the challenges patients face at each point on the path, as reported by stakeholders, and key considerations for health service provision. Stakeholder consultations also elicited several features that were common to all touchpoints. These included: communication & coordination, timeliness, out of pocket expenses, information sharing, diagnostic limbo, fear of genetic testing, own expert from necessity.

Figure 7. Burden of diagnostic odyssey on people with rare disease

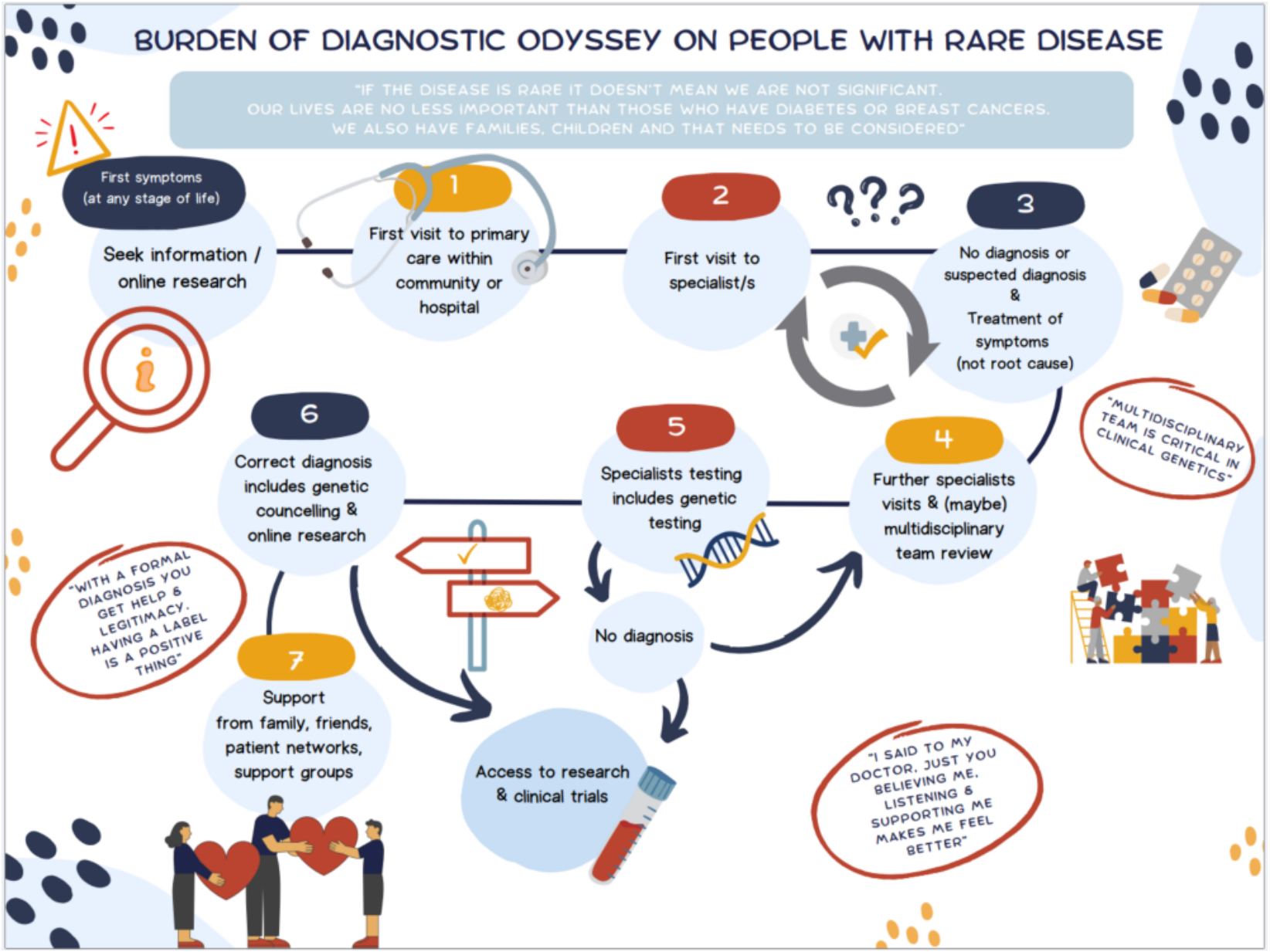


Table 5. Summary of pathways with key touchpoints along the journey, Tasmania

	General Practitioners	Specialists	Multidisciplinary Team Access
Key Issues	**Access** – Waitlist and up to 6 weeks wait – Workforce shortages – Continuity of care with the same GP/practice **Knowledge and awareness** – Familiarity with most common RD such as Huntington but lack of or little knowledge about other RD, resulting into delayed referrals, misdiagnosis or delayed diagnosis **Attitude** – Dismissive attitude – Lack of willingness to listen to the patient and suspect a rare condition – GPs Considering repeated complaints as a result of mental health issues and not believing in patients	**Access** – Waitlist can be 10 months to one year – Workforce shortages – Lack of communication between specialists – Cost and travel to see a specialist **Knowledge** – Lack of comprehensive or holistic view due to limited knowledge of RD **Attitude** – Dismissive attitude due to lack of knowledge and awareness	**Access** – Workforce shortages – Limited specialist availability – Lack of care co-ordination and communication – Multiple specialists visited separately by patients adding to diagnostic delays
Patient Perspectives	– Patients/families understand that not all practitioners will have knowledge regarding all RD, but expect that they will know where to find that information and provide guidance to the patients – Locum doctors fill the gap in the lack of workforce and bring in fresh approach but in some cases add to the lack of continuity of care	– Many patient/carers fully understood the challenge in attracting and retaining specialists in Tasmania and particularly specialists with specific knowledge of RD	– Difficult to navigate the system hence need holistic guidance
Key Considerations	– Develop RD pathways with clear and well communicated referral criteria and processes for inclusion in the HealthPathways portal – Promote the availability and location of existing RD resources that provide information and guidance to health professionals including GPs, paediatricians, and other specialists – Provide core training and education material to Tasmanian GPs on RD	– Increase access to specialists through telehealth appointments – Ensure support is provided to access specialists in other jurisdictions for people with an RD – Recruit and retain specialists in Tasmania	– Establish a Tasmanian-based MDT within a Rare Care Centre for patients with suspected RD for diagnostic purposes and complex post-diagnostic care as required
Feedback Quotes	*"My GP described my situation as being 'odd' but that was where he left it. I think he just didn't know what to do and so just managed symptoms"* *"Doctors tell me to "take a big breath and it will go away"*	*"The specialist asked why I wanted a diagnosis – can you believe that?"* *"I had to do the research myself. I had to talk the specialists into believing me"*	*"Patients end up with various specialists who don't see the whole picture – which is important in the role of a GP. For GPs, navigation of the system is the challenge, as the GPs best option is to refer to various specialists and write multiple referrals. GPs rely on specialists to be coordinated/communicate to work out what's the best option for their patient holistically and therefore a multi-disciplinary team approach is critical"* (GP)

	Investigations	Rare Disease Specialists/Geneticists	Research/Clinical Trials
Key Issues	**Access** – Longer wait to accessing investigations due to delay in suspecting RD – Investigations not available across Tasmania and patients must travel interstate – Cost associated with some investigations can be a barrier – Unclear referral pathways **Workforce** – Skilled workforce – Workforce shortages **Equipment** – Availability of equipment for required tests	**Access** – Workforce shortages – Wait times including a 12-month wait to see a geneticist in Tasmania – Limited availability of RD specialists or locations with experts in RD – Cost and travel to see an RD specialist as mostly work outside Tasmania	**Knowledge** – Health professionals having knowledge of available research and experimental diagnostic tools
Patient Perspectives	– GP awareness re family history for access to prenatal investigations – Parents understanding regarding NBS and participation in undertaking available genetic tests	– Fear of genetics and/or lack of understanding of the impact of the genetic diagnosis – Long wait times/lack of access to RD specialists resulting in diminishing quality of life and increased burden of treatment, with in some cases, a reduced lifespan	– Lack of knowledge and understanding regarding the consent process results in lack of participation
Key Considerations	– GPs, specialists and navigators to promote and/or facilitate access to a range of psychosocial supports (e.g., mental health, NDIS, financial counsellors) for patients, carers and family members – Improve workforce and laboratory capacity to undertake genetic and genomic testing and analysis used for suspected RD – Improve procedures for sample handling and return of results within Tasmania and to other jurisdictions	– Establish a Tasmanian Rare Care Centre for people with RD to avoid siloed health care	– Establish a registry of patients with undiagnosed RD to improve access to undiagnosed disease and RD research and programs – Improve access for individuals to participate in clinical trials and research studies aimed at developing new therapies or improving existing ones
Feedback Quotes	*"I didn't know it was a rare disease and didn't know the impact it would have on my life. No one sat down and explained how my life would be different." (Patient/carer)*	*"With the new complex respiratory team commencing [in Tasmania], my experience with the system has never been better and my child is getting much better outcomes' (Parent)* *"PTAS won't pay because they want people to see specialists in Tasmania, but there aren't any here, or if they are here, they have long wait lists" (Patient/carer)*	*"We hear about more medication options than the specialist does. The neurologist is knowledgeable but can't get hold of the medication we want to try" (Patient/carer)*

6 Models of care

This chapter details MoC components developed based on all the evidence gathered throughout this project. The National Strategic Action Plan for Rare Diseases prioritises the development of MoC through priority action 2.1.1.1[2]:

> Action item 2.1.1.1: Establish standards for care and support that are integrated and incorporate clear pathways throughout all systems. Ensure these are informed by clinical and consumer rare disease experts and that such consultation informs policy development.

It is anticipated that the innovative diagnostic care models developed by this project will influence the operation of any future Undiagnosed Disease Program - Tasmania (UDP-Tas), particularly how the clinical model of UDP-Tas in the hospital system would interact with primary and community health care for patients with undiagnosed diseases and RD. To address the contextual nuances in Tasmania, the key components of a MoC for people with RD in Tasmania have been developed. Based on the information gathered in the previous chapters, seven MoC components were identified that capture health system touchpoints, and reflect a continuum of care that supports the patient journey. The 7 components are:

1. Accessing information
2. Accessing the health system
3. Knowledge and attitudes of clinicians
4. Coordination and integration of care
5. Timely and appropriate investigations
6. Accessing research and clinical trials
7. Accessing support

Each of these MoC components includes three sections: Context, the Current Tasmanian situation, and Options for consideration.

- **Context**: The Context section of each MoC component is based on background evidence and sets out accepted standards and guidelines where available.
- **Current Tasmanian situation**: The current situation section reflects the key findings from the stakeholder consultations including identification of need from patients (and carers) and supplemented by data on workforce availability.
- **Options for considerations**: The options are based on information gathered from the range of stakeholder consultations (surveys, interviews and focus groups) and research undertaken in relation to best practice diagnosis and management of RD.

6.1 Accessing Information

Context:

Due to their rarity, many RD have not been the subject of significant scientific research and therefore there can be limited information available on the underlying causes, mechanisms, and clinical presentations of these conditions. This scarcity of information makes it difficult for healthcare professionals to accurately diagnose RD.[58] It also makes it difficult for patients and carers to access accurate, current, and locally relevant information. Some available resources in Australia include: 1. Genetic Support Network Victoria (GSNV)[28] has a Patient Pathways Program[29] where a Telehealth Patient Pathways Nurse provides free information and support services and links patients with genetic, undiagnosed and RD communities within Australia; 2. RD patient advocacy groups in Australia, such as RVA and Syndromes without a Name provide individualised support and information to patients and their families, while others focus on health system advocacy.

Current Tasmanian Situation:

Access to information about RD was identified by participants as a major challenge. Social media platforms such as Facebook groups were identified by patient/carer participants as being a major, or only, source of information about an RD. Online groups were also identified as a source of information about health professionals and for *"finding out about good doctors".*

International information and advocacy organisations in the UK and USA were identified by patients/carers as useful for providing information on specific conditions and RD. Stakeholders indicated that there are ad-hoc data, information, and resources available relating to different RD, but these resources are fragmented, and general awareness of them is low. Having this information across multiple platforms and not a singular platform that will guide the patients/carers is noted as a challenge for navigation through to diagnosis.

Options for consideration:

- Promote existing (e.g., My Health Record) and explore other systems and processes for improving communication between health professionals treating patients with RD
- Promote the availability of existing resources in Australia
- Establish a Tasmanian-specific RD support group (short to medium-term):
    - Seed funding from government to establish, and then membership driven
    - Include an advocate to contact patients with diagnosed and undiagnosed RD, to advise what to do next and who to contact, for example social supports, occupational therapists for neurological conditions
- Establish a Tasmanian Rare Care Centre for people with RD to avoid siloed health care (long-term)
    - Undertake a feasibility study for the establishment of a Tasmanian Rare Care Centre (short to medium-term)
    - If feasibility is proven, develop a business case and seek resources for establishment (medium-term)
- Encourage collaboration and data sharing among researchers, healthcare providers, and patient advocacy groups via existing resources (short to medium-term).

Other lead agencies could include:

RVA, Consumer-led organisations, Patient advocacy groups, health professionals

6.2 Accessing the Health System

Context:

Early diagnosis of disease is essential as it results in longer periods of higher quality of life, better patient outcomes and reduced expenditure on hospital admissions.[45, 46] Delayed diagnosis can lead to a higher overall burden on the healthcare system including longer hospital stays, increased emergency department visits, and extended outpatient care.[73]

GPs play a critical role in RD diagnosis, especially in Australia where GPs act as gatekeepers in the healthcare system.[59] Many patients with RD will present to a GP with their symptoms as a first step into their diagnostic journey. They will also see their GP in between visits to a specialist, and for a range of other care services. This ongoing relationship that also has a relationship-based advocacy and support role can be a foundation for positive experiences of patients with RD.[59] There are no standards in Australia to guide timeliness around access and wait times to see GPs. There are many factors to consider with regards to GP accessibility including routine versus urgent appointments, after hours services, bulk billing and out of pocket costs, telehealth services, and location, in particular rural and remote locations where limited GPs lead to longer wait times. Most recent data from the ABS indicates that across Australia 39% of people who saw a GP for urgent medical care waited for 24 hours or more and 23% of people indicated that they waited longer than they felt was acceptable to get an appointment with a GP.[84] These percentages were much higher for all rural and remote areas including Tasmania.

In the Australian health system, a medical specialist visit is the next step after the GP for many patients requiring a diagnosis for any complex condition. While most recent data from the ABS indicates that across Australia specialists listened carefully (79% of people reporting), showed respect (83%) and spent enough time with the person (79%), around 26% of people indicated that they waited longer than they felt was acceptable to get an appointment with a specialist.[84] These wait time figures were much higher for all areas compared to metropolitan cities. There are clear clinically recommended timeframes for patients to access outpatient care and support from specialists in the public hospital system, dependent on categorisation of urgency, these being 30, 90 and 365 days for categories 1, 2 and 3, respectively. There is no specialty training for RD practitioners, however clinical geneticists tend to be considered RD specialists. In addition, there are specialists with an interest in particular RD; these specialists may be attached to a centre of excellence or an RD clinic that is location-specific. There are more than 5 centres of excellence or specialised clinics that focus on RD in Australia. For example, there is a clinic in Queensland that focuses on silicosis. Other clinics or centres may be connected to specific patient groups or clinical trials, which drives a patient's desire to see these RD specialists.

Current Tasmanian Situation:

In 2018 Tasmania had 105.4 GPs per 100,000 population compared to 113.4 across Australia. The majority of Tasmanian GPs were located in inner regional locations (112.4 GPs), followed by outer regional, remote and very remote (93.8 GPs) and outer regional (92.3 GPs).[85] According to the Medical Board of Australia, at 30 June 2023 Tasmania had 3060 registered medical practitioners and 883 (28%) were GPs.[86] Local access to general practice, allied health and community nursing services is lower for people living outside urban population centres in Tasmania. Communities often rely on visiting services, which present challenges in delivering continuity of local primary care.[38]

Attracting and retaining healthcare professionals, including doctors, nurses, and allied health professionals, has been a persistent challenge for Tasmania. A shortage of GPs in Tasmania was

identified in our research as impacting a patient's diagnostic journey, and this is amplified in rural areas, particularly in North West Tasmania.

Access to specialists via telehealth increased during the COVID-19 pandemic and that access has been sustained for some patients. Telehealth alleviates fatigue associated with travelling for multiple and regular medical appointments, and reduces costs for patients, all of which were viewed positively by participants. Some patients/carers stated, however, that COVID-19 stopped specialists travelling outside Hobart and patients now need to travel to see specialists.

Clinician/researcher participants indicated that the size of the Tasmanian genetics service is an issue as there are not enough clinicians to meet community needs. Patients and carers identified that they travelled interstate for consultation, diagnosis, and/or treatment with RD specialists and access to an RD specialist requires travel which has associated costs. Fear around genomics and what it means was also reported amongst patients and carers.

Patient/carer participants understand the challenge in attracting and retaining specialists in Tasmania, but want to see Government make continuing efforts to increase the number, particularly in regions where there are large shortages. Tasmania has also identified shortages in specialties such as psychiatry, emergency medicine, general surgery, and some allied health professions. These shortages can impact the provision of specialised care and lead to longer than clinically recommended waiting times for patients.[37] The Tasmanian health system is currently failing to meet many of the clinically recommended timeframes for access to outpatient care under most specialties. Patients/carers understand the challenge in attracting and retaining specialists in Tasmania, particularly specialists with specific knowledge of RD. Patients/carers identified the limited availability and subsequently long waiting times for neurology, paediatrics and rheumatology specialists in Tasmania was commonly raised. Wait times of up to 10 months from booking an appointment to the first appointment were shared by participants. Limited specialists in Tasmania means patients cannot seek a second opinion. The transition from paediatric to adult specialists was identified as a barrier to continuity of care by patients/carers in the consultations and survey.

Stakeholders reported that navigation of the health system was incredibly challenging for patients and families, both during the diagnostic journey and following diagnosis. Some people felt that they were not appropriately informed of available service options and that in some cases health services failed to communicate appropriately with each other and the patient. Some stakeholders, including members of the CRG, indicated that care coordination services would be of significant benefit to patients, particularly in the diagnostic and early stages of post-diagnostic care.

Options for consideration:

- Develop RD pathways with clear and well communicated referral criteria and processes for inclusion in the HealthPathways portal (short to medium-term)
- Investigate the options for a roster of regular GP locums for servicing shortages across Tasmania, to improve communication and continuity of care (short to medium-term)
- Review telehealth policies and procedures to increase access to more GPs in Tasmania through telehealth appointments; particularly in the North West (short to medium-term)
- Recruit and retain additional GPs and specialists in Tasmania (medium to long-term)
- Increase access to specialists through telehealth appointments (short to medium-term)
- Include RD specialists and centres of excellence in documented care pathways for those patients who have complex conditions/diagnoses requiring highly specialised treatment (short to medium-term)

- Ensure support is provided to access specialists in other jurisdictions for people with a RD; this may include changing PTAS eligibility criteria to ensure access to the most relevant specialist (e.g., in private practice) for the respective RD (short to medium-term)
- Establish a Tasmanian Rare Care Centre for people with RD (long-term)
    - Undertake a feasibility study for the establishment of a Tasmanian Rare Care Centre (short to medium-term)
    - If feasibility is proven, develop a business case and seek resources for establishment (medium-term)

Other lead agencies could include:

General practices, Australian Government Department of Health and Aged Care (DoHAC), Primary Health Tasmania (PHT), Rare Care Centres in other jurisdictions

6.3 Knowledge and Attitudes of Clinicians

Context:

One of the most important factors causing the delayed diagnosis of RD is the lack of suspicion of an RD or under-recognition by patients and their medical professionals.[55] It is important to focus on the interventions that are needed to facilitate patients and primary care clinicians to suspect a rare condition (for example via a detailed family history or noticing a clinical presentation consistent with genetic conditions) and its early symptoms along the patient journey.[55]

Genomics is a rapidly advancing area of science. While genetics is part of the core training curriculum for many health professionals,[87] this training is limited and many graduates are still ill equipped to recognise an RD. Many practising health professionals were trained at a time when genetics knowledge and technology was much less advanced than today. Augustine *et. al.* (2017)[88] developed the Care Continuum Model in recognition that traditional health care and local delivery of care *"does not meet the needs of people with rare diseases".* The Model focused on three concepts: 1) telehealth, 2) integration, and 3) improving patient-clinician-researcher collaboration. As a patient-centred model it acknowledges that patients are experts in their RD and encourages a partnership between the patient and their medical practitioners.

Current Tasmanian Situation:

Patients/carers, researchers and specialists all acknowledged that it is not practical for GPs to know about all RD. Patients mentioned that as long as a GP was willing to listen, do some research and keep exploring, including considering that a genetic condition might be present, then that was acceptable to them. Researchers/specialists indicated that GPs can be aware of available resources/support and be able to refer to help with diagnosis.

There were two key aspects that stakeholders felt GPs might need: a) when to think that it might be an RD, and b) who to refer to. Another key challenge in general practice is to have access to RD resources in one place that are easy to navigate. It was indicated by health professionals that GPs in Tasmania use HealthPathways extensively and this was identified as being a key resource to consider developing.

Stakeholders involved in rare care in other jurisdictions advised that while text mining of general practice records can identify certain RD, GP software alerts are often found to be unhelpful, and that general practice solutions need to be considered in collaboration with primary care health professionals.

Patient/carer participants overwhelmingly reported that they are not listened to and not believed by doctors. Several patients/carers reported this was the primary reason for their delayed diagnosis, as it resulted in lower specialist referral rates (including access to a second opinion) and fewer diagnostic investigations being performed. Allied health professionals were identified by patient/carer participants as willing to listen and to follow up when doctors were not willing to progress investigations or referrals.

A doctor (GP or specialist) or other health professional who is willing to listen to the patient and to push for additional tests or additional referrals was considered extremely helpful by patients and carers. There was a sense that often a GP or specialist 'gives in' and reaches an incorrect diagnostic conclusion too early when treating a patient with RD, and then suggests mental illness as a causative factor or the need for mental health support.

Options for consideration:

- Promote the availability and location of existing RD resources that provide RD information and guidance to health professionals including GPs, paediatricians, and other specialists (short-term)
- Develop and promote resources that guide clinicians to troubleshoot when RD is suspected, such as where to go or where to refer (short-term)
- Raise the profile of the TCGS and promote it across Tasmania (short-term)
- Conduct outreach about public clinical genetics services to GPs, primary health practitioners and specialists to improve awareness of services (short-term)
- Provide core training/awareness raising among Tasmanian GPs and specialists to increase their understanding of (short to medium-term):
    - RD and their diagnosis
    - RD referral criteria and processes
    - psychosocial impacts of an RD diagnosis on patients
    - engaging and communicating with a patient/carer with an RD including:
        - providing information to patients on investigations and outcomes of a suspected rare condition
    - connecting patients with the appropriate advocacy group.
- Develop RD pathways for inclusion in the HealthPathways portal (short to medium-term)
- Promote existing hospital systems and explore other options for identifying patients with RD at the point of, or during, their admission, and for improving communication on discharge(short to medium-term)
- Work with primary care and software vendors to improve RD patient identification and management via existing primary practice software and other health ICT solutions (short to medium-term)
- Promote the increased use of My Health Record among specialists to contribute to continuity of care and capturing and retaining all relevant information for timely diagnosis (short to medium-term)
- Develop and implement workshops in genomics for clinicians such as GPs and general paediatricians (short to medium-term)
- Promote the availability and location of existing RD resources that provide RD information and guidance to health professionals including GPs, paediatricians, and other specialists (short-term)
- GPs, specialists and navigators to promote and/or facilitate access to a range of psychosocial supports (e.g., mental health, NDIS, financial counsellors) for patients, carers and family members

Other lead agencies could include:

PHT, RVA, Royal Australian College of General Practitioners (RACGP), Rare Care Centres in other jurisdictions, Australian Genomics

6.4 Coordination and Integration of Care

Context:

Integrated care aims to improve the coordination of care between different services and providers. Various specialists, such as geneticists, neurologists, and GPs and laboratory experts, work collaboratively to discuss complex cases. This multidisciplinary approach ensures shared insights, opinions, and coordinated decision-making, aiming to reach an accurate and timely diagnosis.[17, 18]

Structured multidisciplinary CPW can guide diagnosis, treatment, and care of patients with RD, linking scientific evidence from clinical practice to clinical outcomes whilst optimising clinical efficiency.[17] The application of the CPW in Germany has been shown to result in reducing the number of days required to establish a confirmatory diagnosis: at 109 days it was much shorter than diagnostic delays reported elsewhere in the literature.[43] In Australia, the UDP-WA adopts a multidisciplinary approach to RD: a multidisciplinary panel review is held monthly, with only one new patient being reviewed. There are also MDTs operating in clinical genetics services in Victoria.

Current Tasmanian Situation:

Improving the coordination and integration of care in Tasmanian was identified as a key priority among patients/carers and other stakeholders. The limited availability of specialists within Tasmania results in few formal MDT approaches or independent multidisciplinary input in patient diagnosis and care. Currently, MDTs in Victoria and other Australian jurisdictions (often based on the focal point of an RD) support Tasmanian health professionals in clinical decision making and planning the next steps to take in a patient's diagnostic journey. All stakeholders consulted agreed that an MDT approach is needed in Tasmania and where this occurred elsewhere better outcomes for the patient and health system had resulted. This is all but non-existent in the Tasmanian context for someone with an RD. The lack of access to Tasmanian-based MDT care impacts the timeliness of diagnosis and adds delays to diagnostic testing for example, through siloed medical care.

Options for consideration:

- Establish a Tasmanian-based MDT for patients with suspected RD for diagnostic purposes and complex post-diagnostic care as required (short to medium-term)
- Explore the potential for accessing MDTs within Rare Care Centres and other specialist areas (e.g., complex conditions) in other jurisdictions for Tasmanian patients (short to medium-term)
- Establish a Tasmanian Rare Care Centre for people with RD (long-term)
    - Undertake a feasibility study for the establishment of a Tasmanian Rare Care Centre (short to medium-term)
    - If feasibility is proven, develop a business case and seek resources for establishment (medium-term)

- Work with primary care and software vendors to improve RD patient identification and management via existing primary practice software and other health ICT solutions (short to medium-term)
- Utilise/promote utilisation of electronic record systems to document and safely share patient information, including My Health Record (short to medium-term)
- Promote existing (e.g., My Health Record) and explore other systems and processes for improving communication between health professionals treating patients with RD (short-term)
- Provide a dedicated resource such as nurse navigator for system navigation and care co-ordination for people with a RD (short to medium-term)
- Explore the options to employ a nurse navigator role at a RD telephone helpline (short to medium-term)
- GPs, specialists and navigators to promote and/or facilitate access to a range of psychosocial supports (e.g., mental health, NDIS, financial counsellors) for patients, carers and family members (short to medium-term)
- Establish a Tasmanian-specific RD support group (short to medium-term):
    - Seed funding from government to establish and then membership driven
    - Include an advocate to contact patients with diagnosed and undiagnosed RD, to advise what to do next and who to contact, for example social supports, occupational therapists for neurological conditions
- Encourage collaboration and data sharing among researchers, healthcare providers, and patient advocacy groups via existing resources (short to medium-term).

Other lead agencies could include:

RVA, Consumer-led organisations, patient advocacy groups, health professionals, general practices, DoHAC

6.5 Timely and Appropriate Investigations

Context:

The diagnostic journey for RD often involves multiple specialists, extensive testing, and a process of ruling out more common conditions. Diagnostic tests specific to RD may not be readily available or may require specialised laboratories. Moreover, some RD have overlapping symptoms with other conditions, leading to misdiagnosis or delayed diagnosis.[22]

Current Tasmanian Situation:

Stakeholder consultations identified that there is limited system capacity in RD diagnosis to support the needs of Tasmanians. These limitations in the clinical workforce, laboratory staff, geneticists, genetic counsellors, paediatricians, and genomics have led to long wait lists for diagnostic testing.

Patient/carer participants stated that greater availability of diagnostic tools in Tasmania would have helped to detect and address their condition much earlier. Clinician/researcher participants stated that genomics is a specialised resource and the teams do not necessarily need to be location bound and acknowledged that the process of sample transfer and reporting could be improved.

Some stakeholders reported having multiple investigations undertaken by different health professionals, sometimes repeating diagnostic tests, the results of which were unavailable to others, resulting in a lack of a total picture of the patient's symptoms, potentially resulting in

lack of suspicion of RD or misdiagnosis. Some indicated that public support for preventive health diagnostic testing could be improved for patients with family history of RD.

Options for consideration:

- Improve procedures for sample handling and return of results within Tasmania and to other jurisdictions (short-term)
- Assess to ensure that the Medicare item numbers are available for diagnostic testing, diagnosis and treatment of people with RD (medium to long-term)
- Improve workforce and laboratory capacity to undertake genetic and genomic testing and analysis used for suspected RD (medium-term)
- Utilise/promote utilisation of electronic record systems to document and safely share patient information, including My Health Record (short-term)

Other lead agencies could include:

Laboratory bodies, RACGP (Guidelines), Accredited education institutes

6.6 Accessing Research and Clinical Trials

Context:

Currently, high quality national data on congenital anomalies, including RD, is unavailable, despite recognition that they are a significant public health problem in Australia.[2] This lack of data further exacerbates the challenges in providing a consistent approach to diagnosis. Commonwealth, state and territory approaches to RD remain fragmented.[70]

Elsewhere, the National Rare Disease Plan for Ireland 2014-2018 included key options in improving access to research and innovation.[13]

- Enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of RD, informing care management, research and health system planning
- Develop a national research strategy for RD to foster, support and drive all types of research for RD, contributing to agreed priorities and systematically addressing gaps
- Ensure research into RD is collaborative and person-centred
- Translate research and innovation into clinical care: clinical care informs research and innovation.

Current Tasmanian Situation:

Stakeholders indicated that there are ad-hoc data, information and resources relating to different RD, and that having resources or information links across multiple platforms is a challenge for navigation through to diagnosis. Across Australia, there are many initiatives and resources available to gather information; however, how to gather information effectively so that it can be used for health service planning and how to promote the information to ensure doctors and consumers are accessing it, are two key concerns.

Clinician participants identified that systematic data gathering on RD, and the use of registries would have several benefits. At the patient level, greater data repositories could be used for quicker diagnosis in the future. At the epidemiological level, increased and improved data could be used as evidence to better distribute resources for health services.

There are limited funds dedicated to support access for Tasmanian patients to participate in research and clinical trials via the public genetics service in Tasmania. Present support relies on

time-limited research funding and clinical staff within the public genetics service have a full clinical load, with no dedicated time for supporting research.

Options for consideration:

- Establish a registry of patients with undiagnosed RD to improve access to undiagnosed and RD programs (short to medium-term)
- Additional funding could be allocated to expand public genetics services, to enable greater clinical capacity and dedicated clinician time and associated support to enable patient participation in research and clinical trials.
- Improve access for individuals to participate in clinical trials and research studies aimed at developing new therapies or improving existing ones. For example, by:
    - Generating a local directory of available research opportunities or clinical trials related to RD and promoting the location of such amongst health professionals (medium-term)
    - Considering other ways to notify health professionals of RD research, the registry and opportunities for patients (medium-term)

Other lead agencies could include:

Researchers/universities, data informatics from various stakeholders

6.7 Accessing Support

Context:

Research conducted on RD care pathways in the EU reinforced that patient organisations and resource centres for RD have a special role in patient empowerment and self-management, and this needs to be further promoted.[49] The Australian National Strategic Action Plan for Rare Diseases comprises 3 core pillars, with each pillar outlining priorities, actions and implementation areas: (1) Awareness and Education, (2) Care and Support and (3) Research and Data.

Current Tasmanian Situation:

Patient/carer participants identified that personal circumstances impact the diagnostic journey. This includes social circumstances, including family, friends and other supports available; resilience and a willingness to ask questions and challenge decisions; and health literacy and an understanding of how the health system works and where to find information.

Patient/carer participants said that support networks for RD are limited and lacking; many participants spoke of family and friends providing support, which can strain relationships. Patient/carer participants identified the types of support needed were: emotional support, mental health support, transport to and from appointments, house and garden cleaning and maintenance. They also identified social media platforms such as Facebook groups and other online support groups as being a major source of support, and suggested the need for a Tasmanian specific RD support group.

Patient/carer participants stated that RVA provided information on their specific disease when contacted, but also that RVA advised that the individual would need to become their own advocate; participants considered this an unsatisfactory response. Participants also identified that there are limited RD associations operating in Australia, and they are mostly based in Sydney or Melbourne and do not operate in Tasmania.

Options for consideration:

- Establish a Tasmanian-specific RD support group (short to medium-term):

- Seed funding from government to establish, and then membership driven
- Include an advocate to contact patients with diagnosed and undiagnosed RD, to advise what to do next and who to contact, for example social supports, occupational therapists for neurological conditions

- Establish a Tasmania Rare Care Centre for people with RD (long-term)
  - Undertake a feasibility study for the establishment of a Tasmanian Rare Care Centre (short to medium-term)
  - If feasibility is proven, develop a business case and seek resources for establishment (medium-term)

Other lead agencies could include:

General practices, DoHAC, PHT, Rare Care Centres in other jurisdictions

7 Recommendations

The options for consideration detailed in Chapter 6 have been informed by the data gathered from the range of stakeholder consultations (survey, interviews and focus groups) and research undertaken in relation to best practice diagnosis and management of RD. These options were refined into draft recommendations which were reviewed by the TCGS CRG as well as TRUDN members, and all feedback has been incorporated into these final recommendations. In considering the draft recommendations, the reviewers were asked to consider the following factors:

- Complexity: significantly challenging and are already the subject of strategies to try and address or mitigate the problem
- Responsibility: beyond the remit of the Tasmanian Government and thus require collaboration
- Feasibility: Some may not be feasible
- Stakeholders were also advised that none of the recommendations had been considered by the Tasmanian Department of Health.

A recurring recommendation provided by all stakeholders, and supported by evidence from literature, was the establishment of a Tasmanian Rare Care Centre. This centre would be a single point of care for people with RD, to avoid siloed health care. This centre would ideally include:

- ethos based on patient centred care
- overall guidance from a clinical geneticist
- a navigator role or lead care coordinator
- an MDT, including allied health professionals
- telehealth services, particularly if the centre is located outside Tasmania.

A summary of the recommendations is provided in Table 6. These recommendations are not ranked according to timing, priority or importance.

Table 6. Recommendations

Patients	Health Professionals	Health System
Establish a Tasmanian-specific RD support group	Conduct outreach about public clinical genetics services (TCGS) to GPs and primary health practitioners to improve awareness of services	Establish a Rare Care Centre in Tasmania
Establish a registry of patients with undiagnosed RD to improve access to undiagnosed disease and RD programs, research based treatments and diagnostics and improve health system data in relation to burden of rare disease	Provide core training and education material to Tasmanian GPs to increase their understanding of RD referral criteria and processes, psychosocial impacts of an RD diagnosis on patients, investigations, and connecting patients with the appropriate advocacy/support group	Establish a Tasmanian-based MDT within the Rare Care Centre for patients with suspected RD for diagnostic purposes and complex post-diagnostic care as required, including GP, a resource for system navigation and care co-ordination for patients, provide telephone hotline for patients and clinicians,
Develop resources that assist GPs, specialists and navigators to promote and/or facilitate access to a range of psychosocial supports (e.g., mental health, NDIS, financial counsellors) for patients, carers and family members	Develop and implement workshops in genomics and RD for clinicians such as GPs and general paediatricians, include RD and genomics during hospital training days	Explore the potential for accessing MDTs within Rare Care Centres and other specialist areas (e.g., complex conditions) in other jurisdictions for Tasmanian patients
Raise the profile of the TCGS and promote it across Tasmania	Develop RD pathways with clear and well communicated referral criteria and processes for inclusion in the HealthPathways portal, including information about RD specialists and centres of excellence	Assess to ensure that the Medicare item numbers are available for diagnostic testing, diagnosis and treatment of people with RD
Promote availability and location of existing RD resources for patients and carers	Promote the availability and location of existing RD resources that provide RD information and guidance to health professionals including GPs, paediatricians, and other specialists	Register the Tasmanian Rare Care Centre as an accredited GP training centre, to enable GP professional development and training
	Work with PHN and RACGP to identify GP RD champions	Promote existing hospital systems and explore other options for identifying patients with RD at the point of, or during, their admission, and for improving communication on discharge
	Utilise/promote utilisation of electronic record systems to document and safely share patient information, including My Health Record	Improve workforce and laboratory capacity to undertake genetic and genomic testing and analysis used for suspected RD
	Encourage collaboration and data sharing among researchers, healthcare providers, and patient advocacy groups via existing resources	Improve access to GPs and spcecialists through recruitment, retention, increase access to telehealth appointments, a roster of regular GP locums for servicing shortages across Tasmania to improve communication and continuity of care; support patients with RD to access appropriate specialists in other jurisdictions
		Improve procedures for sample handling and return of results within Tasmania and to other jurisdictions
		Work with primary care and software vendors to improve RD patient identification and management via existing primary practice software and other health ICT solutions

8 Appendices

Appendix 1: Literature review and documentation review methodology

The literature review was conducted using keywords “rare disease” OR “orphan disease” OR “undiagnosed disease” AND “diagnostic care pathways” OR “care pathways” OR “model/s of care”. Possible alternative terms and MeSH terms were also used. Key databases used for the literature searches included PubMed, Medline, Web of Science and Google Scholar. More specific resources directly related to rare diseases included Orphanet Journal of Rare Diseases and portal for rare diseases and orphan drugs (https://www.orpha.net/).

The literature search was not limited to “Australia” or “Tasmania” as it results in very few publications. By obtaining broader international evidence on designing effective patient diagnostic care pathway models in rare and undiagnosed conditions, we were able to identify important elements of effective diagnostic care pathways. These elements also served as an overarching structure/framework for understanding current diagnostic and clinical care pathways, community needs and designing innovative and effective service models to better meet those unmet needs.

We have also explored Australian published and non-published resources that focused on studying existing practice in diagnosing and referring patients with rare and undiagnosed conditions; the issues and challenges in implementing National Strategic Action Plan for Rare Diseases; and initiatives or projects on improving access to early diagnosis.

Our focus was on current literature (no older than 15 years) with information about rare and undiagnosed conditions in regard to the following broader categories and subcategories:

- Prevalence, data and its registries
    - Common conditions in Australia/Tasmania
    - Why it is important to have diagnostic pathways for rare diseases?
- Diagnostic Care pathways for rare diseases
    - Reviews of existing diagnostic pathways for specific conditions
    - Common challenges in diagnosis
- Evidence of factors that would assist early diagnosis:
    - Identification and awareness of red-flags (any existing diagnostic/care pathways for suspected conditions)
    - Promotion of person-centred care approaches
    - Effective relationships between health care providers and patients/carers
    - Skilled workforce and provision of integrated health care/care coordination
    - Other contextual factors such as consumers and carers knowledge and cultural acceptance of referrals, testing methods such as genetic testing; availability of testing capacity.

These categories and subcategories were developed and further expanded as we performed the search and initial synthesis. We did not limit it to our pre-established themes, as various reports and publications about the care pathway implementation informed emerging themes and interrelated topics that need to be considered in the review. As we also interviewed key informants simultaneously, some themes for further exploration emerged from those interviews.

From initial search results, a snowball approach was used to extract further relevant publications with regards to rare disease pathways. The preliminary literature review process assisted us in understanding the general breadth and depth of the issues and challenges in diagnosing these conditions and gaps in the provision of coordinated care both in Australian context as well as in other settings. This was followed by synthesis of key informant interviews that assisted us in identifying current issues and challenges, key

elements of the diagnostic care pathways and identifying potential opportunities and factors for implementing the model of care, especially in rural and remote and hard to reach populations.

Types of documentation reviewed and used in the literature review include the following:

- Research articles on rare disease diagnosis and care in Australia and other comparable countries. We have primarily used high quality research papers published on peer reviewed Australian and International journals.
- Rare disease project documentation provided by Undiagnosed Diseases Program (UDA), WA
- Policy documents and strategic plans on rare disease by the Department of Health, Australia
- Rare disease publications by Australian Genomics
- Other relevant resource links and web-based publications suggested by the clinicians and researchers interviewed as key informant interviewees.

Appendix 2: Summary of the stakeholder interviews

Key findings from the stakeholder interviews are summarised here, but are also integrated throughout this report.

Timeliness of diagnosis

- Variable time and processes for requesting diagnostic tests – dependent on a range of factors:
    - Information gathering, patient profile/context, complexity of conditions, diagnosed mental health condition, family history, presenting features/conditions, symptom development over time, openness of family to support diagnostic process, experience of the provider
    - Nature of condition informs complexity of elimination process for diagnosis and referral (e.g. presents slowly versus urgent acute presentation; increased complexity of multiple organ systems condition; diagnosed mental health condition)
    - Resource capacity of diagnostic laboratories (noting demand has increased as access to genomic technology has increased and costs have reduced)
    - Reliance on knowledge and availability of specialist services
    - No family history means that diagnosis can take longer
    - Rural patients wait time can be reduced if accessing remotely (telehealth)
- Inequitable access for clinical genomic testing across various cohorts – intellectual disability, Aboriginal and Torres Strait Islanders, culturally and linguistically diverse, rural/remote.
- Approximately 50% of rare disease genetic tests do not yield a diagnosis – increasing need for further functional studies (often in research setting - RNA sequencing, cell-based functions) over extended timeframes
- Extensive consent process requires time and paperwork
- Currently accessing international diagnostic testing (i.e. not limited to Tasmanian capacity, expertise) – international testing can lead to data collection challenges
- Identified opportunities to increase timeliness of diagnosis:
    - Automated genomic reanalysis projects (research), rapid genomics testing, and transferring of research technology such as RNA sequencing to a second-tier clinical diagnosis pathway.
    - Resourcing – funding, workforce.

Educational needs of health workforce

- Limited knowledge among primary care and referral providers (including psychiatrists, ophthalmologists) of key issues and availability of diagnostic tests and referral services - reduces referrals to clinical genetics services and/or leads to limited referral information for specialists
- Limited access to clinical genetics networks in Tasmania leads to isolation, limited knowledge sharing
- Identified opportunities to address educational needs of health workforce:
    - Improve training of GPs, generalists / non-genetic workforce: referrals, communication with families, availability of research studies; enhance focus on diagnosis of rare disease within genetics training
    - Training in and embedding clinical ethics consultation
    - Upskill paediatric generalists
    - Upskill adult genetic specialists on genomics in adults (including cardiac genetics)
    - Enhance funding / access to genetic specialists
    - Focus on GPs supporting vulnerable populations
    - Recognition by AHPRA of genetic counsellors
    - Build capacity / upskill laboratory scientists in genomics
    - Increase awareness of HealthPathways as a resource
    - Continue to work toward upgrading university curriculums (non-genomic clinicians)
    - Enhance capacity building for Tasmania through UDP in WA (e.g. collaboration, research case reviews)

Fragmentation of diagnostic pathways

- Current disjointed diagnostic outcomes pathways and referral system:
    - GPs often refer (early) to clinical genetics to avoid complexities of diagnosis, consent
    - Examples cited of participants feeling isolated, fearful, forgotten through diagnosis process
    - Barriers to funding genetic testing (access to MBS items, MSAC reviews ongoing)
- Limited system capacity to meet needs of community (long wait lists), including clinical workforce, laboratory staff, geneticists, genetic counsellors, paediatricians, genomics.
    - NSW: non-urgent patients can wait up to 12 months to access clinical genetics services
- Challenges for GPs include:
    - Limited existing diagnostic/ referral and specialist networks (e.g. within Tasmania), and challenges accessing diagnostic services outside of Tasmania
    - Limited continuity of patient's primary care providers and limited patients with rare disease
    - Limited awareness of when genetics/referral is appropriate
    - Limited awareness of availability of funding / Medicare eligibility of tests, and access to genomic sequencing / accredited genetic testing.
    - Pressure of limited time with patients, including for consent processes
- Poor quality coding and collection of diagnostic data, limited admin support to improve systems
- Challenges at transition between paediatric and adult services due in part to generalist specialty of paediatrics, and specific medical speciality of adult services (e.g. nephrology)
    - Some cohorts (including older people, adolescents) are 'falling through the gaps' as services have tended to support / focus on diagnosis in children (hospital paediatrics)
- Challenges around patient consent to participate in research where focus is clinical diagnosis.
- Identified opportunities for streamlining diagnostic pathways across the health system:
    - Enhance clarity and key messaging among providers for: referral criteria, available services, supporting families through the diagnostic process and accessing services (with resources, care planning, follow-up services such as clinical phenotyping, genomics)
    - Support care continuity, and comprehensive patient records and communications
    - Explore opportunities for national data sharing (noting ethics, privacy, safety) mechanism and governance structure (e.g. Australian Genomics has potential model)
        - Victoria: focus on electronic records has improved communication among MDC team
    - Further enhance HealthPathways for diagnostic pathways and genetics services
    - Curate and bring key information and guidelines together – currently, a range of sources of information, diagnostic and referral guidelines used (see Appendix 2)
    - Integrate genetics earlier (e.g. reproductive carrier testing)
    - Increase access to clinical genetic testing as part of diagnosis (rather than as part of research)
        - Victoria: Genome sequencing program available if no diagnosis after genetic testing
    - Promote MBS item numbers for genetic testing
    - Increase clarity of standards for patient privacy, autonomy whilst supporting participation in research
    - Explore electronic consent processes for genetic services.

System-wide challenges and opportunities

- System wide challenges identified:
    - Lower education / literacy levels in Tasmania compounds low community knowledge
- System wide opportunities identified:
    - There is a need for clear government priorities (e.g. screening or diagnostic program) and supporting policy interface as sector evolves beyond symptomatic screening. This must include a focus on the

whole system (registries, data, health insurance, health system efficiency); and the down-stream implications of diagnosis.
    - Advocate for societal acceptability
    - Enhance funding for reproductive carrier screening (noting this responsibility sits with GPs)
- Future – use of genomics for establishing polygenic risk score (combination of risks based on genomics, environment, family history) for common genetic based conditions (e.g. diabetes)

Models of care

- Providers and families are challenged to navigate complex services system
- Learning from other program models (e.g. WA UDP, NIH Utah USA, Ireland), noting variation in traction, resource intensity and model success:
    - WA - high level of attention/resource per patient (1-2 per month), initial focus on paediatrics
    - NSW - challenged in establishment phase; Gene Equal project (equity – intellectual disability)
    - Victoria - multiple patients per month; success supported by advocacy, patient stories
- Australian Genomics has enhanced efficiencies and impact for work in this area
- Development of state-based genomics programs boosted genomic technologies
- Identified opportunities to improve diagnostic experience and efficiency through models of care:
    - Continue to learn from other program models
    - GP needs to plays critical role in diagnosis and understanding the needs and preferences of families (e.g. transport, telehealth) – but GPs must feel supported
    - MDC team for diagnosis (person-centred approach, shared knowledge, case review, collaborative decision-making, interpretation of genetic tests) – this could include GP, clinical specialist, genetic counsellor, geneticist, radiologist, administrative support
    - Consider efficient use of each MDC role (e.g. more efficient to use genetic counsellors and administrative staff with targeted input from geneticist as required)
    - Care coordination / navigator role (between primary and specialist health providers)
    - Recognise pan-disciplinary nature of genetics, genomics technology (and options for remote delivery)
    - Explore model for GPs with special interest
    - Facilitate translation of genomics in health care, and explore options for genetic cause research to inform genetic testing (e.g. acute risk prediction)
    - Embed clinical ethics consultation process into the diagnostic pathway.

Appendix 3: Survey Reports

The following provides an analysis of the responses to the Patient Survey and the Clinician/Researcher/ Advocacy Organisation survey.

Patient Survey

The Patient, Family or Carer survey was developed and approved by the University of Tasmania Human Ethics and Research Committee. The survey was promoted via various advocacy organisations as well as on social media (Facebook page) by the Department of Health, Tasmania. It was open for two months (Aug and Sep 2023) with 1443 responses received. Responses with incomplete data were excluded, resulting in 880 eligible responses available for analysis. The rationale applied for excluding responses were related to:

- Participants who started to complete the survey multiple times but had only one complete survey; incomplete duplicates were excluded
- Surveys that had limited demographic data and no response for other questions and as such insufficient information.

Demographics

This section provides a summary of the analysis of demographic questions in the survey relating to type of respondent, current diagnosis status (at time of survey response), gender, age and geographic region of residence.

Type of respondent

From the total of 880 survey respondents, 546 (62%) have a rare disease themselves while 334 (38%) respondents support or are related to someone with a rare disease, as illustrated in Table 1 below.

Table 1:Type of respondents, all respondents n=880

Type of Respondent	Number of participants	% of participants
I have a rare disease	546	62%
I support/am related to someone with rare disease	334	38%
Grand Total	880	100%

Diagnosis Status

Of the 880 total respondents, 797 (91%) reported having a diagnosis at the time of completing the survey, and 83 (9%) were awaiting diagnosis.

Of the 797 respondents with a diagnosis, 495 (62%) were patient respondents and 302 (38%) were carer/family respondents, reflecting the same distribution of all respondents (with or awaiting a diagnosis). Diagnosis status of patient and carer/family respondents is further detailed in Table 2 below.

Table 2: Diagnosis status of person with rare disease

Respondent Type	Await diagnosis		Have diagnosis		Total
	N	%	N	%	
Patient respondent	51	9%	495	91%	546
Carer/family respondent	32	10%	302	90%	334
Total	**83**	9%	797	91%	880

Note that in the analysis respondents with a rare disease are referred to as 'patient respondents' and respondents that support or are related to someone with a rare disease are referred to as 'carer/family respondents'.

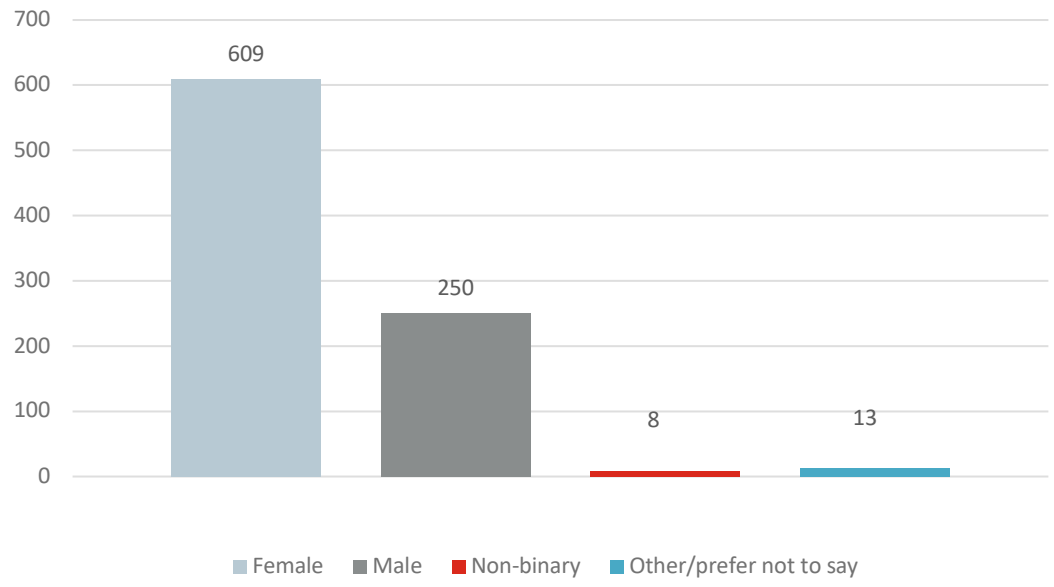
Gender distribution

Of the 880 respondents:

- 609 (69.2%) were female
- 250 (28.4%) were male
- (0.9%) were non-binary
- 13 (1.5%) were another gender, unsure or preferred not to say.

This distribution is detailed below in Figure 1.

Figure 1: Number of respondents by gender, n= 880



The distribution of gender by respondent type was different to the overall distribution:

- For patient respondents, 465 (85%) were female and 69 (13%) were male
- For carer/family respondents, 144 (54%) were female and 181 (54%) were male.

The survey questions were worded to ensure that even if the carer/family member answers the questions, the answer sought information related to the person in their care.

Age group of persons with rare disease

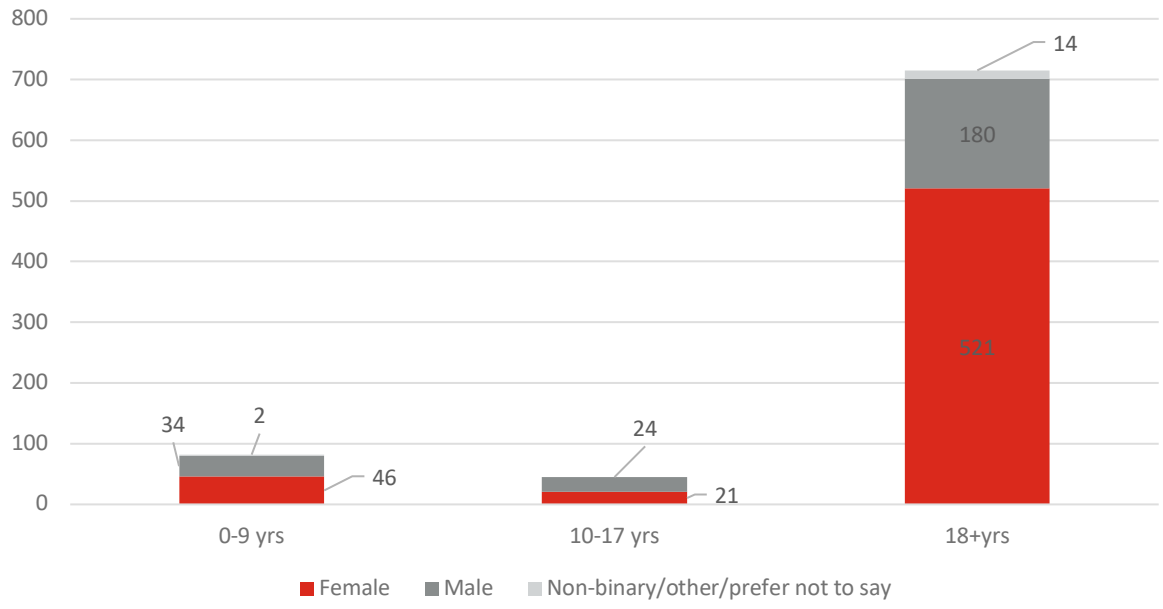
The age of the person with the rare disease (i.e. age of patient respondents, and age of the person the carer/family respondent were responding on behalf of) ranged from 1 month to 86 years. The distribution of age groups (n=842) included:

- 715 (85%) aged 18 years and over
- 45 (5%) aged 10-17 years
- 82 (10%) aged 0-9 years.
- The distribution of gender by age group was:
- 18 years and over – 73% female / 25% male / 2% non-binary, unsure, prefer not to say
- 10-17 years – 47% female / 53% male
- 0-9 years – 56% female / 42% male / 2% non-binary, unsure, prefer not to say
- This is described in Table 3 and Figure 2 below.

Table 3: Age distribution of persons with rare disease participating in the survey or for which answers were provided on their behalf by a carer or family member, n=842

Gender	0-9yrs	10-17yrs	18+yrs	Total
Female	46 (56%)	21 (47%)	521 (73%)	588 (70%)
Male	34 (42%)	24 (53%)	180 (25%)	238 (28%)
Non-binary/prefer not to answer/unsure	2 (2%)	0 (0%)	14 (2%)	16 (2%)
Grand Total	82 (100%)	45 (100%)	715 (100%)	842 (100%)

Figure 2: Age and gender distribution of persons with rare diseases

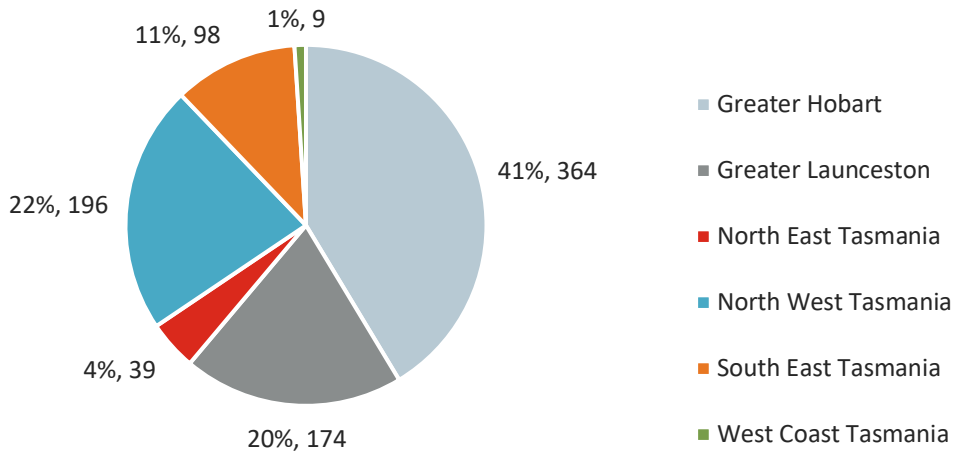


Geographical distribution

The largest number of respondents (n=880) resided in Greater Hobart and Greater Launceston. The distribution of respondents in Greater Hobart (approximately 40%) and outside of Greater Hobart is in accordance with the overall population distribution across Tasmania[23]. Key findings of geographic distribution of respondents:

- 364 (41%) reside in Greater Hobart
- 196 (22%) reside in North West Tasmania
- 174 (20%) reside in Greater Launceston
- 98 (11%) reside in South East Tasmania.

Figure 3: Location of respondents across Tasmania, n=880



Diagnostic Journey

This section summarises the survey findings relating to the diagnostic journey of the person with rare disease, including time between first symptoms and diagnosis, age at diagnosis, and the health services sought in their diagnostic journey. Unless otherwise specified, this analysis includes respondents with diagnosis and awaiting diagnosis.

Time between first symptoms and diagnosis

Please interpret this data with caution as not all respondents might have interpreted and responded appropriately to the survey question. Some numbers exceed the evidence/expectations and are likely outliers. It is also plausible that some of the time or time periods may reflect issues such as:

1. When a child is diagnosed then a parent or relative might be tested and also diagnosed. The parent might get diagnosed at 50 years of age but retrospectively identify that the symptoms started in their teens making the time between symptoms and diagnosis more than 30 years
2. Genetic testing for a condition was not available 20 years ago when symptoms were first identified but now is available making the time between first symptoms and diagnosis greater than expected at 20 years.

This analysis only includes responses where the survey respondent indicated they already have a rare disease diagnosis and excludes responses that identified 'awaiting diagnosis'.

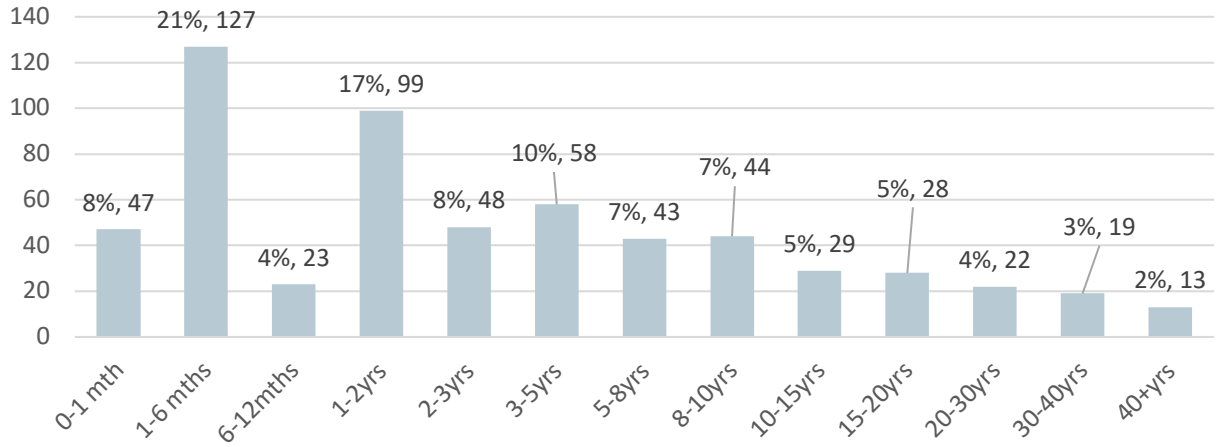
[23] https://www.population.net.au/hobart-population/#:~:text=Hobart%20is%20Tasmania's%20state%20capital,Tasmania%20people%20lives%20in%20Hobart.

Key findings (n=600) relating to time between first symptoms and diagnosis include:

- Almost a third (174 or 29%) received a diagnosis within 6 months of first observing symptoms
- Almost half (296 or 49%) received a diagnosis within 2 years of first observing symptoms
- A further third (193 or 32%) received a diagnosis between 2 and 10 years of first observing symptoms
- 10% (57) received a diagnosis between 10 and 20 years of first observing symptoms
- 9% (54) received a diagnosis between more than 20 years after first observing symptoms.

This is illustrated in Figure 4 below.

Figure 4: Time between first symptoms and diagnosis (n=600)



Age when symptoms first noticed

Please interpret this data with caution as not all respondents might have interpreted and responded appropriately to the survey question. Some numbers exceed the evidence/expectations and are likely outliers. This question might also be impacted by the recall bias for some of the participants.

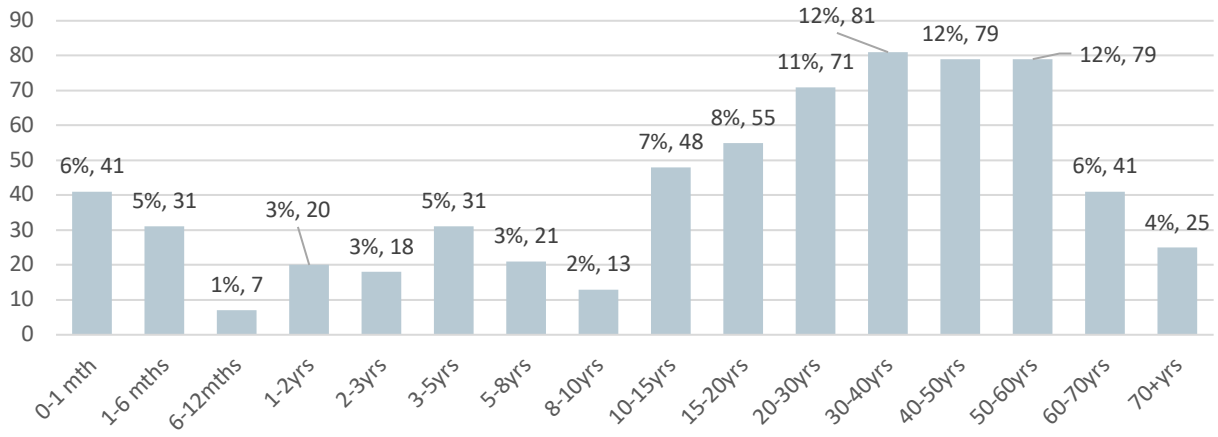
The reported age when symptoms were first noticed varied widely across respondents (n=662), from birth to 82 years of age. The distribution of age when symptoms were first noticed did not differ significantly for those with a diagnosis compared to those awaiting diagnosis. Key findings by age are included in Table 4.

Table 4: Age distribution of persons when symptoms first noticed, n=660

Age	% and number of respondents
0-1 month	6% (41)
1-12 months	6% (38)
1-5 years	10% (69)
5-20 years	21% (137)
20-40 years	23% (152)
40-60 years	24% (158)
60 years or older	10% (66)
	100%/660 respondents

This is illustrated in Figure 5 below.

Figure 5: Age of the patient when the symptoms were first noticed, n=660



Age of person at diagnosis

This analysis only includes responses where the survey respondent indicated they already have a rare disease diagnosis and excludes responses that identified 'awaiting diagnosis'.

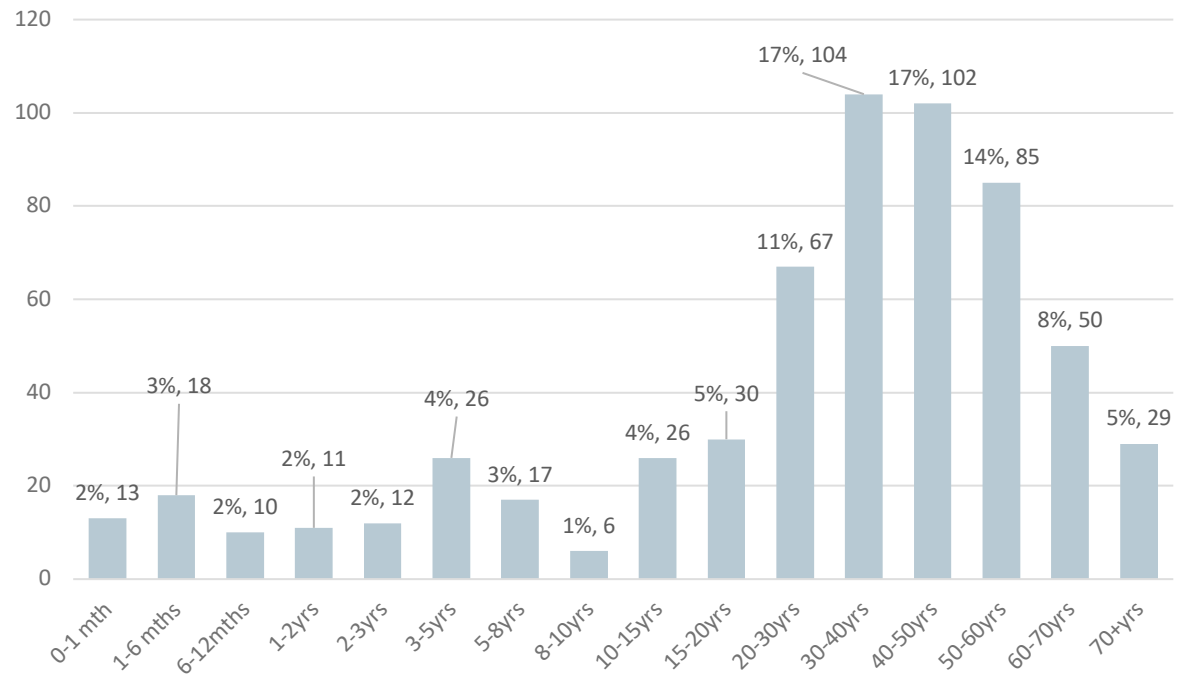
The reported age at the time of diagnosis varied widely across respondents (n=606), from 0 to 82 years. The majority were adults at the time of diagnosis. The distribution of age at diagnosis is illustrated in Table 5.

Table 5: Age distribution of persons when diagnosed with a rare disease, n=606

Age	% and number of respondents
Less than 1 year old	7% (41)
1-5 years	8% (49)
5-15 years	8% (49)
15-20 years	5% (30)
20-40 years	28% (171)
40-60 years	31% (187)
60-70 years	8% (50)
70 years or older	5% (29)
	100%/606 respondents

This is illustrated in Figure 6 below.

Figure 6: Age of the patient at diagnosis, n=606



Health professionals visited when symptoms were first noticed

Survey respondents were asked to list all the health professionals that they visited when symptoms were first noticed. Only 680 participants responded. A total of 866 health professionals were visited by 680 patients when symptoms were first noticed. The most visited health professional was the GP (493, 57% of responses), followed by the hospital emergency department (17%), paediatrician (9%) and neonatal unit (3%). Note: See full list of health professionals in Appendix 6. Further detail is provided in the table below.

Table 6: List of health professionals visited when symptoms were first noticed (where identified by more than 5 respondents)

Health Professional/location	Number	%
Local Doctor / GP	493	57%
Hospital emergency	150	17%
Paediatrician	77	9%
Neonatal unit	22	3%
Specialist (not specified)	12	1%
Other/not specified	11	1%
Rheumatologist	10	1%
Neurologist	10	1%
Hospital	7	1%
Optometrist / Optician	6	1%
Physiotherapist	5	1%
Pulmonary / lung / respiratory specialist	5	1%

Medical specialists seen for diagnosis

Respondents were asked to list the type of medical specialists seen to get a diagnosis. There were 652 responses that identified seeing one or more specialist. There were 450 respondents who indicated seeing two or more medical specialists, and 286 respondents who identified seeing three or more providers to obtain a diagnosis. The list of the top 10 medical specialists identified by respondents is included in Table 7 below and the full list is in Appendix 6.

In this question, respondents included allied health professionals, specialist doctors and general practitioners in their responses. Allied health professionals (across a broad range of disciplines) and neurologists had the highest number of responses with 121 (11%) and 120 (11%) respectively. Other common responses included surgeon (across a range of surgery types) with 76 (7%), cardiologist with 74 (7%), rheumatologist with 66 (6%) and gastroenterologist with 60 (6%) responses. A further 5% (56) responses identified a geneticist as a medical specialist seen for their diagnosis. Further detail is provided in the table below.

Table 7: Top 10 'medical specialists' seen to get a diagnosis (n=652, total 1,080 medical specialists identified)

Health Professional	Number	%
Allied health	121	11%
Neurologist	120	11%
Surgeon	76	7%
Cardiologist	74	7%
Rheumatologist	66	6%
Gastroenterologist	60	6%
Geneticist	56	5%
Paediatrician	42	4%
Endocrinologist	34	3%
Haematologist	30	3%
Immunologist	30	3%

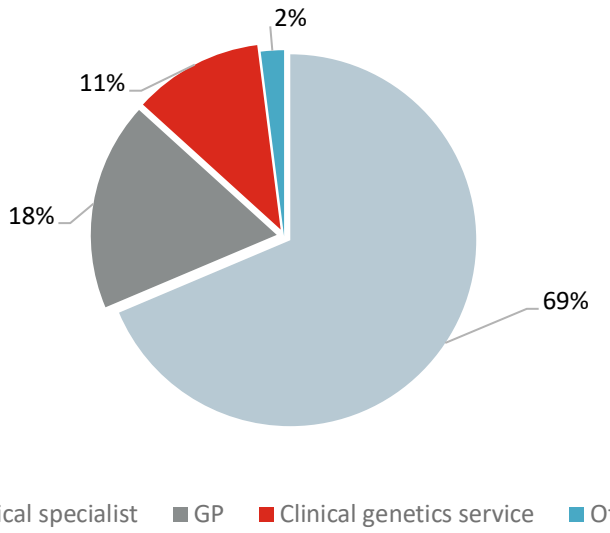
Medical professional/s providing the diagnosis

Survey respondents were asked "who gave you or the person you support a diagnosis?". A total of 619 responses identified 746 providers that provided the diagnosis. There were 20 patients who indicated seeing more than two professionals who gave them the diagnosis. Note: interpret this with caution as the medical specialist category may include geneticist also.

Key findings include:

- Medical specialist (broad range of disciplines) was identified in 69% (512 of 746) of responses
- GP was identified in 18% (135 of 746) responses
- Clinical genetics service was identified in 11% (84 of 746) responses
- Other responses (2%, 15) included allied health, nurse, family link, unsure.

Figure 7: Type of medical specialist/service providing the rare disease diagnosis



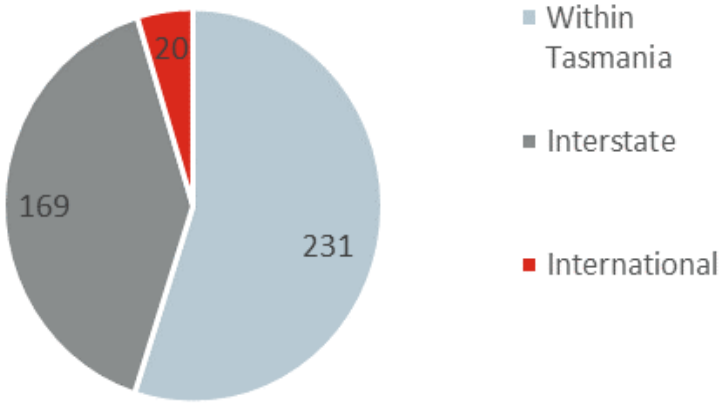
Diagnosis related travel, costs and other difficulties

Travel

Survey respondents were asked about their need to travel whilst seeking a diagnosis including where travel was more than 50 kilometres; and subsequently if travel was within Tasmania, interstate and/or overseas. From 688 responses, 357 patients (52%) indicated that they travelled more than 50 kilometres when seeking their diagnosis, followed by 309 (44%) indicating they didn't travel more than 50 kilometres. The remaining 26 (4%) had not travelled more than 50 kilometres but were still undiagnosed. Note that of the 26 without a diagnosis and that had not travelled more than 50 kilometres, 65% (17) lived in Greater Hobart, and a further 27% (7) lived in Greater Launceston.

The destination category (for those that travelled >50 kilometres to seek a diagnosis) was identified by 327 survey respondents with a total 420 destinations identified. Approximately 40% (169 of 420) of responses included interstate, and 5% (20 of 420) of responses indicated international travel was a feature when seeking a diagnosis.

Figure 8: Destination (for those that travelled >50 kms for their diagnosis), n=327 (total 420 destinations identified)



Survey respondents were also asked about the number of times they had travelled to the various destinations when seeking a diagnosis. Key findings include:

When travelling within Tasmania (n=223):

- 22% (49) travelled up to 2 times
- 27% (60) travelled between 3 and 5 times
- 20% (44) travelled between 6 and 10 times
- 21% (47) travelled between 11 and 20 times
- 17% (37) travelled more than 20 times.

When travelling interstate (n=164):

- 48% (79) travelled up to 2 times
- 24% (40) travelled between 3 and 5 times
- 6% (10) travelled more than 20 times.

When travelling overseas (n=5), respondents travelled between 1 and 5 times to:

- United States (n=3)
- England (n=1)
- New Zealand (n=1)

This is illustrated in the figures below.

Figure 9: Number of times survey respondents travelled within Tasmania when seeking a diagnosis (n=223)

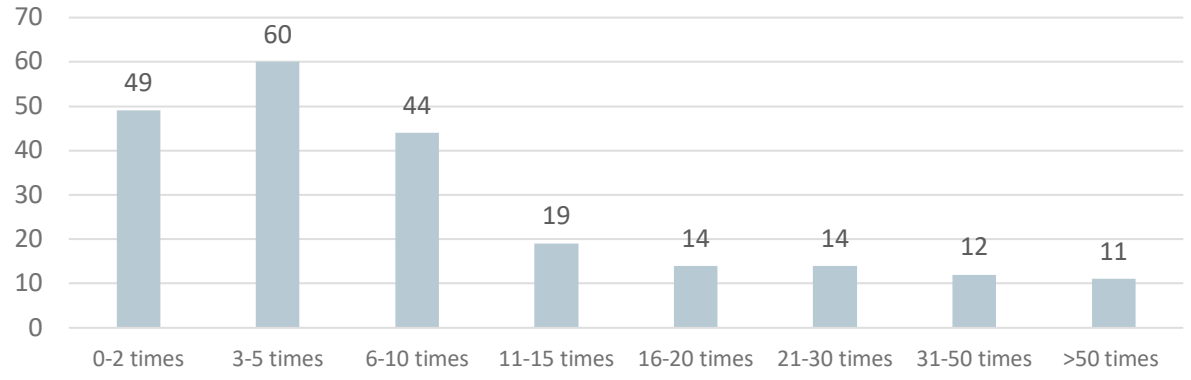
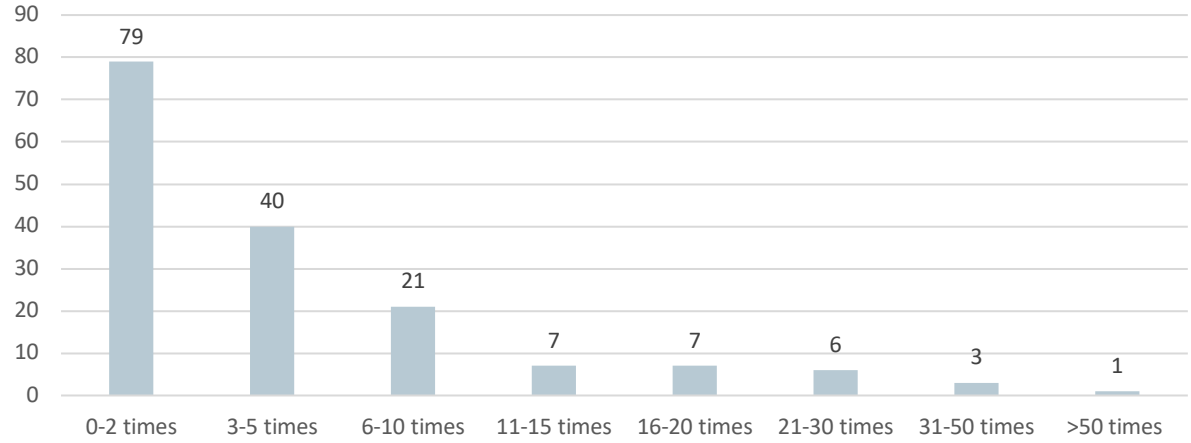


Figure 10: Number of times respondents travelled interstate (n=164)



Out-of-pocket expenses

Survey respondents were asked if there were out-of-pocket expenses for them when seeking a diagnosis, and if out-of-pocket expenses were a barrier to seeking a diagnosis. Key findings include:

- 438 respondents (70% of total 629) incurred out-of-pocket expenses
- 230 (52% of 431) respondents who incurred expenses indicated the expenses were a barrier to their diagnosis
- Overall, of 657 responses, 273 (42%) of a total 657 respondents identified out-of-pocket costs were a barrier for them, and 384 (58%) indicated expenses have not been a barrier.
    - 23 (or 12% of total 191) respondents indicated they had not incurred out-of-pocket expenses, but that out-of-pocket expenses had been a barrier to seeking a diagnosis. Of this group of 23, 4 (17%) were awaiting a diagnosis.
    - Of the 230 respondents that indicated they have incurred out-of-pocket expenses that were a barrier to seeking their diagnosis, 32 (14%) were awaiting a diagnosis.

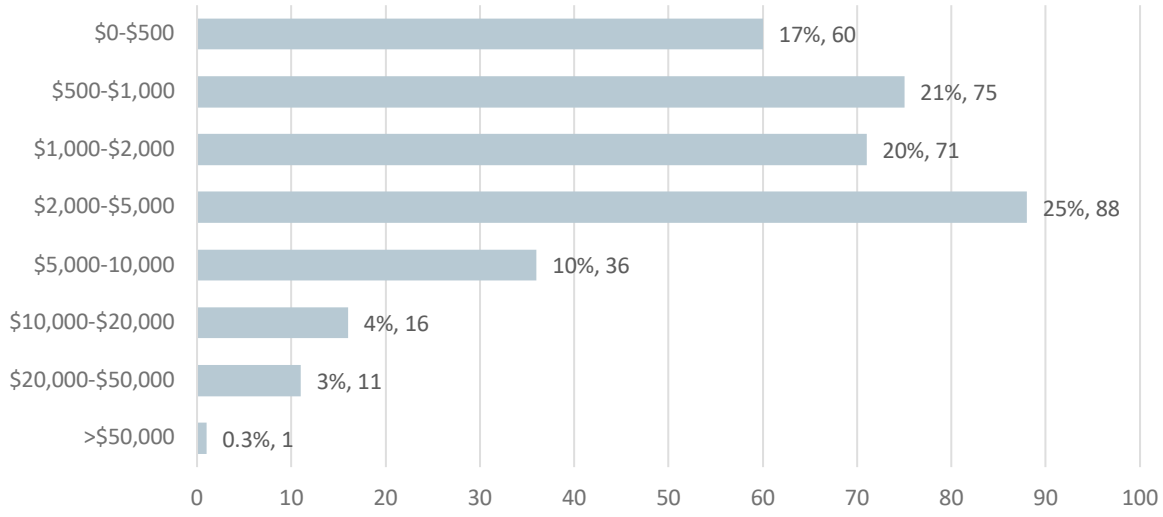
The range of out-of-pocket expenses (for those incurring these costs) ranged widely (n=358). As of 25 September 2023, these findings are yet to be cross-referenced with timing of diagnostic journey (this analysis will be updated).

Key findings (yet to be cross-referenced, excludes 'unsure/not specified responses):

- Majority (58%, 206) incurred out-of-pocket expenses up to $2,000
- 25% (88) incurred out-of-pocket expenses between $2,000 and $5,000
- 10% (36) incurred out-of-pocket expenses between $5,000 and $10,000
- Approximately 8% (28) incurred out-of-pocket expenses over $10,000.

This is illustrated in Figure 11 below.

Figure 11: Out-of-pocket expenses during the diagnostic journey, n=358



Other difficulties in a diagnostic journey

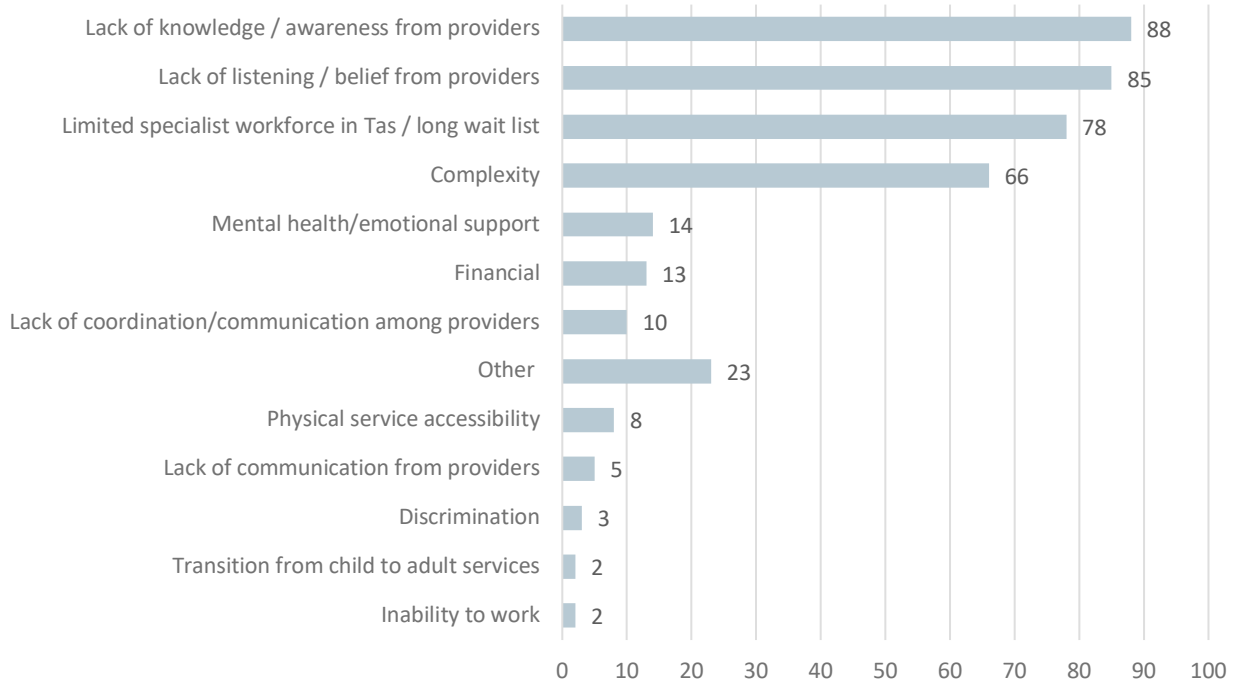
Survey respondents were asked if there were difficulties experienced other than out-of-pocket expenses, when seeking a diagnosis. Of the 647 respondents, 350 (54%) indicated that they had experienced other difficulties, with 310 respondents describing the difficulties faced. These were thematically grouped (with up to three difficulties for each respondent) and are represented in the figure below.

The most common cited challenges include:

- Lack of knowledge and awareness from providers
- Lack of listening / belief from providers (including examples of lack of empathy, poor attitude, discrimination and 'gaslighting')
- Limited specialist workforce in Tasmania and/or long wait lists to see a specialist or other health provider.

This is illustrated in Figure 12 below.

Figure 12: Other difficulties experienced in the diagnostic journey (n=310, with total 397 difficulties listed)



Survey respondents were also asked "did you change or do you plan to change your residential address in relation to making it easier for you to get a diagnosis?", and additionally, the reasons for moving or planning to move. Approximately 9.3% (57 of 615) respondents have changed their residential address to facilitate seeking a diagnosis or for management/treatment of their condition. The reasons for changing residential address were noted by 53 respondents as:

- To get improved access to support / specialists due to limitations in Tasmania
- To move closer - proximity to hospital / services
- To access specialists
- For physical accessibility (mobility)
- Responses also included reasons not able to move (e.g. supporting a person; needing to be near city/services, affordability, waiting for a Housing Tasmania place).

Clinician/Researcher/advocacy organisation survey

The survey for clinicians, researchers and advocacy organisations working with rare and undiagnosed disease (Appendix 2) was approved by the University of Tasmania HREC. It was distributed by:

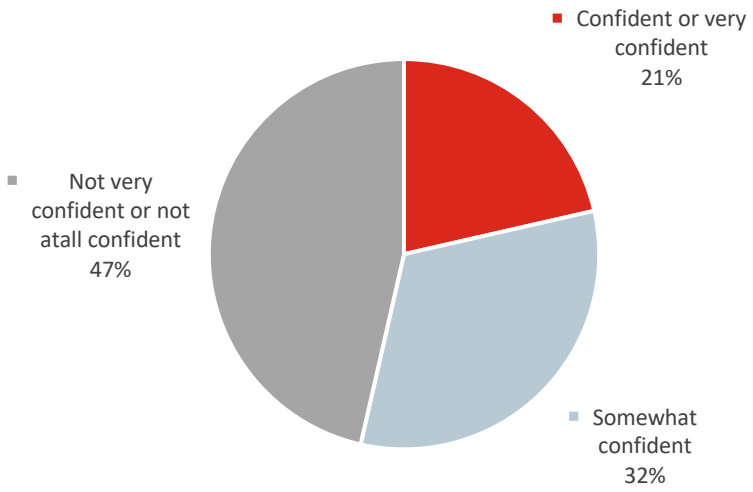
- the Department of Health via the newsletter that is circulated to all clinicians.
- Abt Associates to the advocacy organisations with valid email addresses.
- Primary Health Tasmania via their newsletter to primary health professionals

Currently (September 2023), there are a total of 32 valid responses and 31 responses for clinical information.

- From 32 participants who responded 13 were nurses (41%) followed by nine specialists (28%), seven allied health professionals (22%), two GPs (6%) and one representative of an advocacy organisation (n=3%).
- Most respondents worked in Hobart (n=13) followed by North West Tasmania (n=10) and Greater Launceston (n=7)
- From 32 participants, 28 identified their confidence levels in seeing / supporting people with rare disease with 'Confidence' being low in almost 47% of respondents.

This is illustrated in the figure below.

Figure 138: Distribution of confidence levels is supporting people with rare disease across survey respondents (n=28)



Health professional respondents were asked to identify the number of patients with rare disease they see each year:

- 39% (n=12) of health professional respondents reported seeing one to five patients with rare disease per year and 29% (n=9) see / support up to 20 patients in a year in their practice.

The distribution by the type of health professional is included in table 8 below.

Table 8: Distribution of number of patients with rare disease seen annually, by health professional (n=30)

	Allied Health	GP	Medical Specialist	Nurse	Total N	%
None	0	0	1	1	2	6.7
one to five per year	3	1	0	8	12	40.0
Six to 20 per year	2	0	6	1	9	30.0
21 to 50 per year	1	0	2	1	4	13.3
More than 50 per year	0	0	0	1	1	3.3
Don't know	0	1	0	0	1	3.3
Many	0	0	0	1	1	3.3
Uncertain on definition	1	0	0	0	0	0.0
Total	7	2	9	13	30	100.0

Respondents were asked about the frequency of reviewing a patient with rare disease. A total of 20 responses were collected with details included in table 9 below. Key findings include:

- Approximately 30% of health professionals said that they review patients with rare disease weekly or monthly.

Table 9: Distribution of frequency of review for patients with rare disease, by health professional (n=20 Responses)

	Allied Health	GP	Medical Specialist	Nurse	Total	%
Weekly				1	1	5.0
Monthly	1		1	3	5	25.0
6-monthly	1	2	3	1	7	35.0
6-12 monthly			3		3	15.0
Less than annually	2		1	1	4	20.0
Total	4	2	8	6	20	100

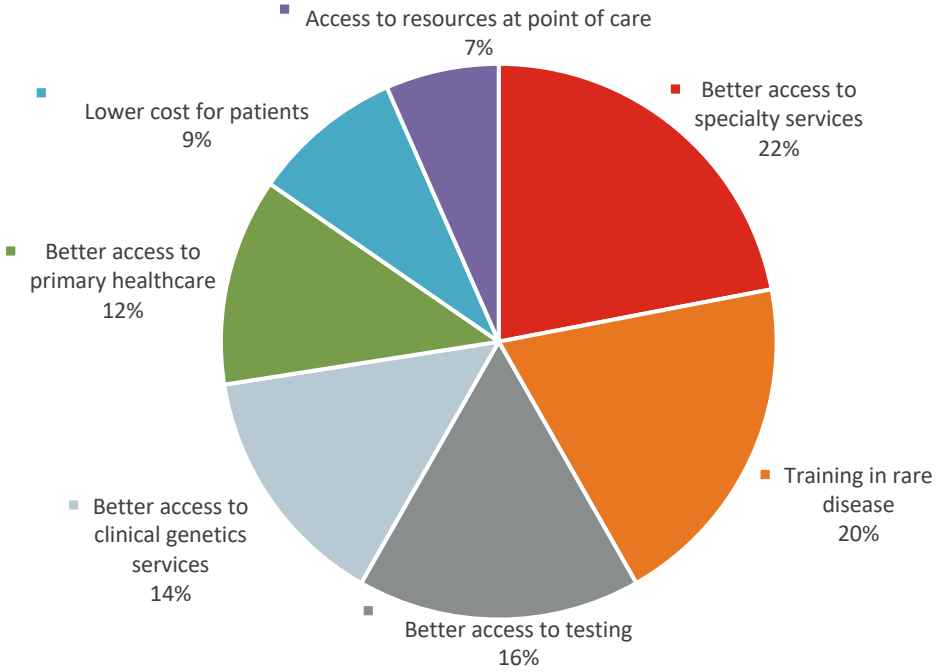
The majority of health professional respondents indicated that changes are required for rare disease diagnosis in Tasmania (n=27 out of 31, 90%) and listed opportunities for improvements to diagnostic care in Tasmania. The top three opportunities for improvement identified include:

- Better access to specialty services
- Training / education in rare disease
- Improved access to testing.

Other opportunities include improved access to clinical genetics services and primary care, reduced financial burden for patients, and improved access to resources and information for patients and families.

This is illustrated in figure 14 below.

Figure 14: Opportunities for improvement for rare disease diagnosis in Tasmania identified by health professional respondents (n=31)



Appendix 4: Survey tools

Patient, Carer or Family Survey

Q1 How are you connected to rare disease?

- ○ I have a rare disease
- ○ I support/am related to someone with rare disease

Q2 What is the gender of the person affected with or investigated for a rare disease?

- ○ Woman
- ○ Man
- ○ Non-binary (Gender Fluid, Genderqueer, Third Gender, etc.)
- ○ Unsure
- ○ Prefer not to answer
- ○ Identity not listed (please specify)

Q3 What is the age of the person affected with or investigated for a rare disease?

- ○ Months (if less than 1 year old)
- ○ Years (if 1 year old and above)

Q4 In what area of Tasmania do you live?

- ○ Greater Hobart
- ○ North West Tasmania
- ○ North East Tasmania
- ○ Greater Launceston
- ○ West Coast Tasmania
- ○ South East Tasmania

Q5 Do you, or the person you support, have a diagnosis for your/their rare disease?

- ○ Yes
- ○ No - I am waiting for a diagnosis

Display This Question:

If 5. Do you, or the person you support, have a diagnosis for your/their rare disease? = Yes

5a What was the approximate time (in months or years) between first symptoms and diagnosis for you or the person you support?

- ○ Months (if less than 1 year old)
- ○ Years (if 1 year old or more)

Display This Question:

If 5. Do you, or the person you support, have a diagnosis for your/their rare disease? = No - I am waiting for a diagnosis

Q5b What is the approximate time (in months or years) between first symptoms and now?

- ○ Months (if less than 1 year old)
- ○ Years (if 1 year old or more)

Q6 What was the age of the person when symptoms were first noticed?

- ○ Months (if less than 1 year old)
- ○ Years (if 1 year old or more)

Display This Question:

If 5. Do you, or the person you support, have a diagnosis for your/their rare disease? = Yes

Q7 What age were you or the person you support when you got a diagnosis?

- ○ Age in months at diagnosis (if less than 1 year old)
- ○ Age in years at diagnosis (if 1 year old or more)

Q8 Where did you go when symptoms were first noticed? (mark all that apply)

- ☐ Local doctor's clinic
- ☐ Hospital emergency
- ☐ Neonatal unit
- ☐ Child specialist (paediatrician)
- ☐ Other (please comment below)

Q9 Please list the type of medical specialist(s) you or the person you support have seen to get a diagnosis (e.g. cardiologist; neurologist; paediatrician etc.). Please list all that you remember below.

- ○ Type of specialist 1 _____
- ○ Type of specialist 2 _____
- ○ Type of specialist 3 _____
- ○ Type of specialist 4 _____
- ○ Type of specialist 5 _____
- ○ Other (list all other specialists that are not listed above)

Display This Question:

If 5. Do you, or the person you support, have a diagnosis for your/their rare disease? = Yes

Q10 Who gave you or the person you support the diagnosis? (mark all that apply)

- ☐ General Practitioner
- ☐ Medical Specialist
- ☐ Clinical Genetics Service
- ☐ Other (please comment below)

Q11 Have you or the person you support had to travel more than 50 kilometres (in one trip) to see health professional(s) for a diagnosis?

- ○ No - but no diagnosis yet
- ○ No
- ○ Yes

Display This Question:

If 11. Have you or the person you support had to travel more than 50 kilometres (in one trip) to see... = Yes

Q11a If yes, where to and how many times? (please include approximate number of visits)

- ☐ Within Tasmania _____
- ☐ Interstate _____
- ☐ International _____

Display This Question:

If 11a. If yes, where to and how many times? (please include approximate number of visits) = International

Q11b If international, list the countries you visited for a diagnosis?

- ○ Country 1 _____
- ○ Country 2 _____
- ○ Country 3 _____

Q12 Have you or the person you support had to pay out-of-pockets costs in seeking a diagnosis from health professionals?

- ○ No
- ○ Yes (please provide total approximate expense over time)

_____

Q13 Have up-front or out-of-pocket costs been a barrier to you or the person you support in seeking a diagnosis?

- ○ No
- ○ Yes

Q14 Did you (or the person you support) experience (or still experiencing) other difficulties in seeking a diagnosis?

- ○ No
- ○ Yes (please specify kind of difficulties experienced)

_____

Q15 Did you change or do you plan to change your residential address in relation to make it easier for you to get a diagnosis?

- ○ Yes (please provide more detail)
- ○ No

Q16 Your experience is valuable for this project. Information you provide will help us in understanding your experience in seeking a diagnosis for yourself or the person you support. This will help us design better diagnosis pathways. Are you willing to speak with our researchers in more detail about your experience with rare disease diagnosis? If yes, please provide your contacts details below.

- ○ Name: _____
- ○ Phone _____
- ○ Email _____

Thank you.

If you have questions about the actual survey, feel free to contact Dr Sanjoti Parekh (07) 3891 4136, or email Sanjoti.Parekh@abtassoc.com.au OR Andrew Alderdice at Andrew.alderdice@abtassoc.com.au.

Clinician, Researcher, Advocacy Organisation Survey

Q1 How are you connected to rare disease?

- ○ I am a health worker/ clinician
- ○ I am a researcher
- ○ I work in rare disease advocacy

Display This Question:

If How are you connected to rare disease? = I am a health worker/ clinician

Q2 What is the area you work in?

- ☐ General Practitioner
- ☐ Medical Specialist
- ☐ Allied Health
- ☐ Nurse
- ☐ Community Health Worker
- ☐ Researcher (6)
- ☐ Other (please specify below) (7) _____

Display This Question:

If How are you connected to rare disease? = I am a health worker/ clinician

Q3 In what region/s of Tasmania do you practice? (Please tick all that apply)

- ☐ Greater Hobart
- ☐ North West Tasmania
- ☐ North East Tasmania
- ☐ Greater Launceston
- ☐ West Coast Tasmania
- ☐ South East Tasmania

Display This Question:

If How are you connected to rare disease? = I am a health worker/ clinician

Q4 How confident are you in helping someone with a suspected rare disease get a diagnosis?

	Not at all confident (1)	Not very confident (2)	Somewhat confident (3)	Confident (4)	Very confident (5)	Not applicable (6)
Choose as applicable (1)	○	○	○	○	○	○

Display This Question:

If How are you connected to rare disease? = I am a health worker/ clinician

Q5 How many people with rare disease do you routinely see (e.g. are regularly seeing within a one year period)?

_____

Display This Question:

If How are you connected to rare disease? = I am a health worker/ clinician

Q6 How often are you seeing someone in your practice who has a rare disease?

	Daily (1)	Weekly (2)	Monthly (3)	6-monthly (4)	6-12 monthly (5)	Less than annually (6)	Not applicable (7)
Choose as applicable (1)	○	○	○	○	○	○	○

Display This Question:

If How are you connected to rare disease? = I am a health worker/ clinician

Q7 On average, how often do you review the person with a rare disease?

	Weekly (1)	Monthly (2)	6-monthly (3)	6-12 monthly (4)	Less than annually (5)	Not applicable (6)
Choose as applicable (1)	○	○	○	○	○	○

Display This Question:

If How are you connected to rare disease? = I am a health worker/ clinician

Q8 Are changes needed for rare disease diagnosis in Tasmania?

- ○ No (1)
- ○ Yes (2)

Display This Question:

If Are changes needed for rare disease diagnosis in Tasmania? = Yes

Q8a What changes are needed for rare disease diagnosis in Tasmania? (Please tick all that apply)

- ☐ Better access to testing (1)
- ☐ Better access to primary healthcare (7)
- ☐ Better access to specialty services (8)
- ☐ Better access to clinical genetics services (2)
- ☐ Training in rare disease (3)
- ☐ Access to resources at point of care (4)
- ☐ Lower cost for patients (5)
- ☐ Other (please specify below) (6) _____

Display This Question:

If How are you connected to rare disease? = I am a researcher

Q10 What is your research focused on? (e.g. rare disease diagnosis; rare disease care; cost of rare disease diagnosis etc.)

_____

Display This Question:

If How are you connected to rare disease? = I am a researcher

Q11 What difficulties (if any) do you experience in undertaking research for rare disease in Tasmania?

_____

Display This Question:

If How are you connected to rare disease? = I am a researcher

Q12 Do you have any non-research involvement with rare disease?

- ○ No (1)
- ○ Yes (Please describe) (2) _____

Display This Question:

If How are you connected to rare disease? = I work in rare disease advocacy

Q13 Which patient organisation/peer support group/advocacy group do you work or volunteer for?

_____

Display This Question:

If How are you connected to rare disease? = I work in rare disease advocacy

Q14 What condition/s do you advocate for?

_____

Display This Question:

If How are you connected to rare disease? = I work in rare disease advocacy

Q15 How does your organisation advocate for and/or support patients, family and carers? (Please tick all that apply)

- ☐ Direct patient / carer support (1)
- ☐ Policy advocacy (2)
- ☐ Other (please specify below) (3) _____

Display This Question:

If How are you connected to rare disease? = I work in rare disease advocacy

Q16 Do you think there are barriers to rare disease diagnosis in Tasmania?

- ○ No (1)
- ○ Yes (2)

Display This Question:

If Do you think there are barriers to rare disease diagnosis in Tasmania? = Yes

Q16a What do you think are the barriers to rare disease diagnosis in Tasmania (Please tick all that apply)

- ☐ Access to testing (1)
- ☐ Access to primary healthcare (9)
- ☐ Access to specialty services (10)
- ☐ Access to clinical genetics services (2)
- ☐ GP understanding of rare disease (3)
- ☐ Access to resources at point of care (4)
- ☐ Hight cost of services for patients (5)
- ☐ Need to travel to access service (6)
- ☐ Cultural appropriateness (7)
- ☐ Other (please specify below) (8) _____

Display This Question:

If How are you connected to rare disease? = I work in rare disease advocacy

Q17 Do you support patients in/advocate within the Tasmanian health system?

- ○ No (1)
- ○ Yes (2)

Q18 Your experience is valuable for this project. Information you provide will help in understanding diagnostic pathways and therefore help to improve better patient access to diagnosis and care. Are you willing to speak with our researchers in more detail about your experience with rare disease diagnosis? If yes, please provide your contacts details below.

- ○ Name: (1) _____
- ○ Preferred contact (e.g. phone number / email address) (2) _____

Appendix 5: Interview and Focus Group questions

Interviews with Specialists

1. Please tell us a little about the rare/undiagnosed disease(s) that you predominantly see
2. Do you have a sense of the number of people with this rare/undiagnosed disease? Is there a mechanism to be definitive about this? E.g. coding or does a registry exist, would this be beneficial?
3. Could you outline for us the care pathway that you/your patients follow?
4. What do you consider is working really well for your clients/you?
5. Where do you see the barriers, gaps, issues on that pathway?
6. Are you aware of any literature/documentation of a best evidence care pathway for the disease? Do you use the website resources such as Orphane, OMIM, GeneReviews at all?
7. Brief discussion of optimal pathway based on the Ireland model below; Then ask, "If applicable, What do you consider to be the major variance from that optimal pathway for the Tasmanian context?"
8. Who else do you think is critical to speak with for this mapping and problem identification phase?

Prioritising the issues:

9. If you could address the barriers/issues of the care pathway, what would be your three priorities, ideally in order?
10. Do you consider that patients might have a different priority issue, and if so what would be the issue(s) they would raise?

Issues to raise and explore if not addressed in responses above:

- Diagnostic delays?
- Is overarching care coordination an issue?
- Are there issues relating to the transition from child to adult services (disease dependent)?
- Are GPs sufficiently educated in rare/undiagnosed disease?
- Barriers to genetic testing?
- Barriers to clinical trials?
- Are there particular geographic barriers to access?
- Are there particular cultural barriers to access?
- Are there systemic/structural barriers to access?

Optimal Pathway from Ireland

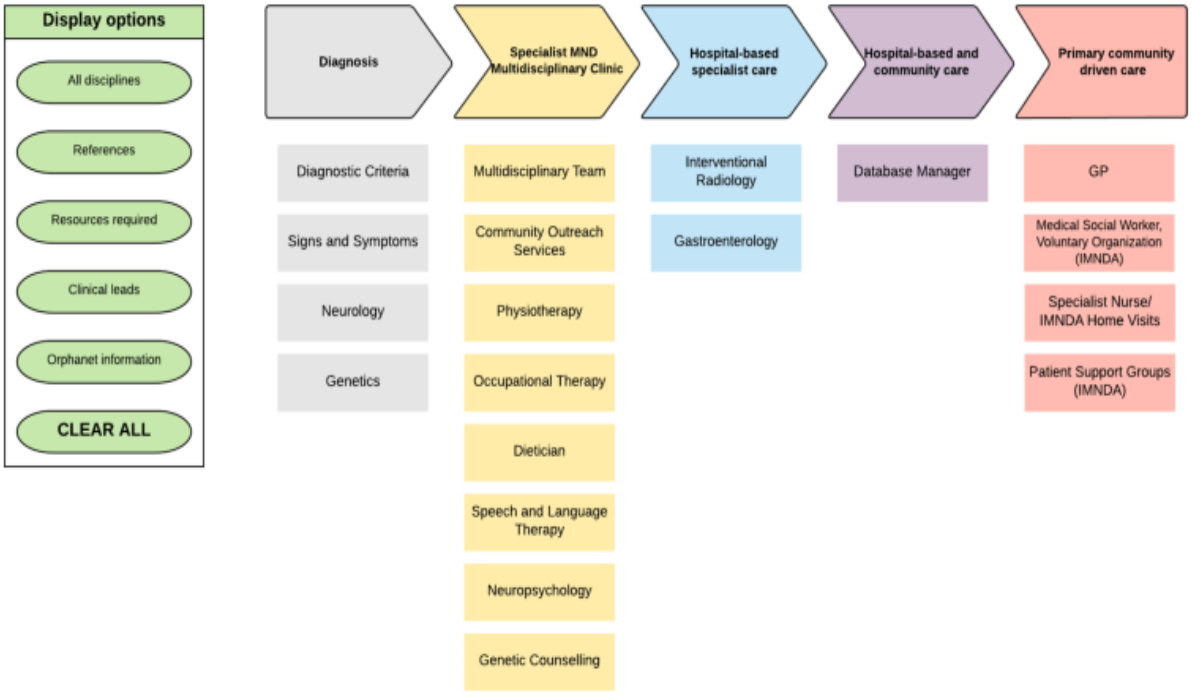


Fig. 4 Lucid Chart care pathway model of the Amyotrophic Lateral Sclerosis Care Pathway ref [49] illustrating the required medical disciplines, HSCPs, steps of diagnosis and care. The display options section shows the core information—references, clinical leads, centres of expertise, Orphanet information and resources

Interviews with Researchers

1. Please tell us a little about the rare/undiagnosed disease(s) that you/your research predominantly focus on?
2. Could you outline for us the care pathway that patients follow?
3. What do you consider is working really well for the clients?
4. Where do you see the barriers, gaps, issues on the diagnostic pathway?
5. Brief discussion of optimal pathway based on the Ireland model below; Then ask, "If applicable, What do you consider to be the major variance from that optimal pathway for the Tasmanian context?"
6. Who else do you think is critical to speak with for this mapping and problem identification phase?

Prioritising the issues:

7. If you could address the barriers/issues of the diagnostic and care pathway, what would be your three priorities, ideally in order?
8. Do you consider that patients might have a different priority issue, and if so what would be the issue(s) they would raise?

Issues to raise if not addressed in responses above:

- Diagnostic delays?
- Is overarching care coordination an issue?
- Are there issues relating to the transition from child to adult services (disease dependent)?
- Are GPs sufficiently educated in rare/undiagnosed disease?
- Barriers to genetic testing?
- Barriers to clinical trials?
- Are there particular geographic barriers to access?
- Are there particular cultural barriers to access?
- Are there systemic/structural barriers to access?

Focus group/Interviews with Patients, Family or Carers

The phrasing and delivery of these questions will be amended depending on whether it is 1:1 interview with a patient, a group of patients, or a patient peak organisation.

1. Please tell us a little about the health condition that you experience
2. How long have you been diagnosed with this condition?
    a. how long did it take you to get a diagnosis?
    b. are you still waiting to get a specific diagnosis (undiagnosed)?
3. Do you obtain support through a patient organisation?
4. Have you had to travel to see health professionals for a diagnosis?
5. Have you had to pay out-of-pockets costs in seeking a diagnosis from health professionals? Has this been a barrier to you in seeking a diagnosis?
6. Could you outline for us the journey you have taken/are on from before being diagnosed to the point of ongoing management
7. What are the things that helped you on that journey?
8. What do you think are the major problems on that journey?
9. If you do not have a diagnosis, what impact has this had on you? (suggestions – access to treatment, trials, welfare, employment)

Prioritising the issues:

10. If you could address the barriers/issues of the care pathway, what would be your three priorities, ideally in order?

Issues to raise if not addressed in responses above:

- Do you have a health professional who is coordinating your care?
- If not, who could that person be?
- If you do have a health coordinator, are there any issues with that coordination?
- What aspects of your care need coordination?
- Are there issues relating to the transition from child to adult services (disease dependent)?
- Do you think GPs (other providers) are sufficiently educated in your rare/undiagnosed disease?
- Are there barriers to genetic testing (cost, access, cultural)?
- Are there barriers to access because of where you live in Tasmania? (e.g. diagnostic delays, lack of available treatments, difficulty in finding the appropriate health services, families feeling isolated, under-supported, facing economic hardship.
- Are there barriers to access or other issues you face because of your cultural background?
- Are there barriers for you because of your ability to read and write or understand the information health professionals give you?
- Are there health system barriers for you?

Focus group/Interviews with Patient advocacy groups

1. Please tell us a little about the health condition that you advocate for?
2. Could you outline for us the journey that patients have taken/are on from before being diagnosed to the point of ongoing management
    a. What are the things that helped them work really well on that journey?
    b. What do you think are the major problems barriers, gaps, issues on that journey?
3. If patients do not have a diagnosis, what impact has this had on them? (e.g. – access to treatment, trials, welfare, employment)
4. Are you aware of any literature/documentation of a best evidence care pathway for the disease you advocate for?
5. Who else do you think it is critical to speak with for mapping and problem identification?

Prioritising the issues:

6. If you could address the barriers/issues of the diagnostic and care pathway, what would be your three priorities, ideally in order?
7. Do you consider that patients might have a different priority issue, and if so what would be the issue(s) they would raise?

Issues to raise if not addressed in responses above:

- Need to travel to see health professionals for a diagnosis?
- Paying out-of-pockets costs in seeking a diagnosis from health professionals? A barrier to patients seeking a diagnosis?
- Diagnostic delays?
- Is overarching care coordination an issue?
- Are there issues relating to the transition from child to adult services (disease dependent)?
- Are GPs sufficiently educated in rare/undiagnosed disease?
- Barriers to genetic testing?
- Barriers to clinical trials?
- Are there particular geographic barriers to access?
- Are there particular cultural barriers to access?
- Are there systemic/structural barriers to access?

Interviews with 'Generalist' providers
(not specialists in one disease group)

1. Can you tell us about the patients you have seen with rare or rare undiagnosed diseases?
2. Could you outline for us where you sit on the care pathway, who refers to you and who do you refer on to?
3. What do you think is generally working well for people with rare/undiagnosed diseases and for your profession/role?
4. Where do you see the barriers, gaps, issues on that pathway for people with rare/undiagnosed diseases and for your profession/role?
5. Are you aware of other points along the pathway where there are barriers for patients, carers or providers?
6. Are you aware of any literature/documentation of a best evidence care pathway for any of the rare/undiagnosed disease groups?
7. If applicable, What do you consider to be the major variance from that optimal pathway?
8. Who else do you think is critical to speak with for this mapping and problem identification phase?
9. Do you have a sense of the number of people with these rare/undiagnosed disease? Is there a mechanism to be definitive about this? E.g. does a registry exist, would this be beneficial?

Prioritising the issues:

10. If you could address the barriers/issues of the care pathway, what would be your three priorities, ideally in order?
11. Do you consider that patients might have a different priority issue?

Issues to raise if not addressed in responses above:

- Is overarching care coordination an issue?
- Are there issues relating to the transition from child to adult services (disease dependent)?
- Are GPs sufficiently educated in rare/undiagnosed disease?
- Barriers to genetic testing?
- Barriers to clinical trials?
- Are there particular geographic barriers to access?
- Are there particular cultural barriers to access?
- Are there systemic/structural barriers to access?

Appendix 6: Health professional seen when symptoms first noticed

Table 1: Full list of health professionals seen when symptoms first noticed (patient/carer survey) – n=680 with total 866 health professionals identified

Type of Health Professional	Total	% (total)
Local Doctor / GP	493	57%
Hospital emergency	150	17%
Paediatrician	77	9%
Neonatal unit	22	3%
Specialist (not specified)	12	1%
Other/not specified	11	1%
Rheumatologist	10	1%
Neurologist	10	1%
Hospital	7	1%
Optometrist / Optician	6	1%
Physiotherapist	5	1%
Pulmonary / lung / resp specialist	5	1%
Maternity ward	4	0%
No-one	4	0%
Blood test / cancer screening	3	0%
Child health clinic	4	0%
Geneticist	4	0%
Imaging / radiology	3	0%
Cardiologist	3	0%
Gastroenterologist	4	0%
Gynaecologist	1	0%
Oncologist	1	0%
Haematologist	2	0%
ICU	1	0%
Neurosurgeon	2	0%
Physician	1	0%
Podiatrist	1	0%
Dentist	2	0%
Ophthalmologist	3	0%
Dermatology / skin clinic	4	0%
ENT	3	0%

Type of Health Professional	Total	% (total)
Liver specialist	1	0%
Vascular surgeon	1	0%
Surgeon	2	0%
Pain specialist	2	0%
Hearing / audiology	1	0%
Immunologist	1	0%
Total	866	100%

Table 2: Full list of health professionals seen to get a diagnosis (patient/carer survey) – n=652 with total 1080 health professionals identified

Type of health professional	Total	% (total)
Allied health	121	11%
Neurologist	118	11%
Surgeon	76	7%
Cardiologist	74	7%
Rheumatologist	66	6%
Gastroenterologist	60	6%
Geneticist	56	5%
Other	45	4%
Paediatrician	42	4%
Haematologist	30	3%
Immunologist	30	3%
GP	29	3%
Endocrinologist	29	3%
Ophthalmologist	28	3%
Respiratory specialist	27	3%
ENT	25	2%
Oncologist	23	2%
Gynaecologist	23	2%
Neurosurgeon	20	2%
Pain specialist	20	2%
Dermatologist	18	2%
Psychiatrist	16	1%
Radiologist	15	1%

Type of health professional	Total	% (total)
Physician	15	1%
General Medicine physician	12	1%
Infectious disease specialist	10	1%
Urologist	9	1%
Sports Physician	6	1%
Liver specialist	6	1%
Nephrologist	6	1%
Metabolic Specialist	5	0%
Dentist	3	0%
Colorectal specialist	3	0%
Pathologist	3	0%
Genetic counsellor	3	0%
Integrative physician	3	0%
Vascular specialist	2	0%
ME/CFS specialist	2	0%
Other	1	0%
Total	1,080	

9 References

1. Australian Government Department of Health and Aged Care, 2023, *What we're doing about rare diseases,* < https://www.health.gov.au/topics/chronic-conditions/what-were-doing-about-chronic-conditions/what-were-doing-about-rare-diseases>.
2. Australian Government Department of Health, 2020, *National Strategic Action Plan for Rare Diseases*, <https://www.health.gov.au/resources/publications/national-strategic-action-plan-for-rare-diseases>.
3. Chung, C.C.Y., A.T.W. Chu, and B.H.Y. Chung, 2022, 'Rare disease emerging as a global public health priority', *Front Public Health*, 2022, 10, p. 1028545.
4. Nguengang Wakap, S., et al., 2020, 'Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database', *European Journal of Human Genetics*, 28(2), pp. 165-173.
5. Elliott, E.J. and Y.A. Zurynski, 'Rare diseases are a 'common' problem for clinicians', in *Australian Family Physician*, 2015, 44, pp. 630-633.
6. NORD®, 2020, *Barriers To Rare Disease Diagnosis, Care And Treatment In The US: A 30-Year Comparative Analysis*. National Organisation for Rare Diseases, US, < https://rarediseases.org/wp-content/uploads/2020/11/NRD-2088-Barriers-30-Yr-Survey-Report_FNL-2.pdf>.
7. Vanhaecht, K., 2007, *The impact of Clinical Pathways on the organisation of care processes,* PhD thesis, Kathlieke Universiteit Leuven, Belgium.
8. Australian Genomics Health Alliance, 2019, *Clinical Genomics: Investigating perceptions of unmet need in Australia. An Initial Study*, National Diagnostic and Research Network Program, Australian Genomics.
9. Government of Western Australia Child and Adolescent Health Services, 'New Rare Care Centre will coordinate rare disease care at Perth Children's Hospital', 2022, <https://pch.health.wa.gov.au/About-us/News/New-Rare-Care-Centre-at-PCH#:~:text=New%20Rare%20Care%20Centre%20will%20coordinate%20rare%20disease%20care%20at%20Perth%20Children's%20Hospital,-Rare%20Care%20Centre&text=Western%20Australia%20is%20taking%20a,with%20rare%20and%20undiagnosed%20diseases>.
10. Rare Diseases International, *Operational Description of Rare Diseases*, accessed 21 June 2024, < https://www.rarediseasesinternational.org/description-for-rd/>.
11. United States Census Bureau, 2020, 'U.S. Census Bureau Today Delivers State Population Totals for Congressional Apportionment', <https://www.census.gov/library/stories/2021/04/2020-census-data-release.html>.
12. Australian Therapeutic Goods Administration, 'Orphan drug designation eligibility criteria', accessed 21 June 2024, <https://www.tga.gov.au/resources/resource/guidance/orphan-drug-designation-eligibility-criteria>.
13. National Clinical Programme for Rare Diseases, 2020, *Model of Care for Rare Diseases*, Health Service Executive, Ireland.
14. Walker, C.E., et al., 2017, 'The collective impact of rare diseases in Western Australia: an estimate using a population-based cohort', *Genet Med*, 19 (5), pp. 546-552.
15. Lacaze, P., et al., 2017, 'Rare disease registries: a call to action', *Internal Medicine Journal*, 47 (9), pp. 1075-1079.
16. Ruseckaite, R., et al., 2023, 'Current state of rare disease registries and databases in Australia: a scoping review', *Orphanet Journal of Rare Diseases*, 18 (1), p. 216.
17. Ward, M.M., 2005, 'Estimating rare disease prevalence from administrative hospitalization databases', *Epidemiology*, 16 (2), pp. 270-1.
18. Aymé, S., B. Bellet, and A. Rath, 2015, 'Rare diseases in ICD11: making rare diseases visible in health information systems through appropriate coding', *Orphanet Journal of Rare Diseases*, 10 (1): p. 35.
19. Finnish Institute for Health and Welfare, 2023, *ORPHAcodes for rare diseases*, <https://thl.fi/en/research-and-development/research-and-projects/national-coordination-of-rare-diseases/orphacodes-for-rare-diseases>.

20. Kole, A., Hedley V., et al., 2021, *Recommendations from the Rare 2030 Foresight Study: The future of rare diseases starts today.* Eurordis, < https://www.rare2030.eu/recommendations/>.
21. eClinicalMedicine, 2023, 'Raising the voice for rare diseases: under the spotlight for equity', *EClinicalMedicine*, 57, p. 101941.
22. Adachi, T., et al., 2023, 'Enhancing Equitable Access to Rare Disease Diagnosis and Treatment around the World: A Review of Evidence, Policies, and Challenges', *Int J Environ Res Public Health*, 20(6).
23. Australian Genomics, 2023, *Australian Genomics Health Alliance*, viewed 10 May 2023, <https://www.australiangenomics.org.au/>.
24. Australian Government Department of Health and Aged Care, 2023, *Medical Research Future Fund*. 2023, viewed 10 May 2023, <https://www.health.gov.au/our-work/medical-research-future-fund>.
25. Rare Voices Australia, 2023, *Rare Voices Australia*, viewed 1 May 2023, <https://rarevoices.org.au/>.
26. Australian Government Department of Health, 2017, *National Health Genomics Policy Framework*, accessed 21 June 2024, < https://www.health.gov.au/resources/collections/national-health-genomics-policy-framework>.
27. Queensland Genomics, 2024, *Rare diseases in children*, viewed 21 June 2024, https://queenslandgenomics.org/clinical-projects-3/rare-diseases/.
28. Genetic Support Network Victoria, 2023, *Genetic Support Network of Victoria*, viewed 10 May 2023, <https://www.gsnv.org.au/>.
29. Genetic Support Network Victoria, 2023, *Patient Pathways Program* 2023, viewed 20 May 2023, <https://www.gsnv.org.au/patient-pathways-program/.
30. QIMR Berghofer Medical Research Institute, 2023, *Genetic Health Pathways*, viewed 1 May 2023, <https://www.qimrberghofer.edu.au/our-research/aboriginal-torres-strait-islander-health/genetic-health-pathways/>.
31. Palmer, E.E., et al., 2023, 'Rare diseases: new approaches to diagnosis and care', *Medicine Today*, 24 (1-2), pp. 69-75.
32. Field, M., Boat, T., (eds), 2015, *Rare Diseases and Orphan Products Accelerating Research and Development*, Institute of Medicine Committee on Accelerating Rare Diseases Research, National Academies Press, Washington DC.
33. Wright, C.F., D.R. FitzPatrick, and H.V. Firth, 2018, 'Paediatric genomics: diagnosing rare disease in children', *Nat Rev Genet*, 19 (5), pp. 253-268.
34. Primary Health Tasmania, 2021, *Health in Tasmania: Primary Health Tasmania Health Needs Assessment 2022–23 to 2024–25*, <https://www.primaryhealthtas.com.au/wp-content/uploads/2022/04/Primary-Health-Tasmania-Needs-Assessment-2022-25.pdf>.
35. Australian Bureau of Statistics, 2021, *2021 Hobart, Census All persons QuickStats*, <https://abs.gov.au/census/find-census-data/quickstats/2021/601051027>.
36. Australian Bureau of Statistics, 2023, *Remoteness Areas: Australian Statistical Geography Standard (ASGS) Edition 3*. 2023, <https://www.abs.gov.au/statistics/standards/australian-statistical-geography-standard-asgs-edition-3/jul2021-jun2026/remoteness-structure/remoteness-areas>.
37. Tasmanian Department of Health, 2019, *Health Workforce 2040: Strategy*, <*https://www.health.tas.gov.au/sites/default/files/2021-12/Health_workforce2040_strategy_DoHTasmania2019.pdf*>.
38. Primary Health Tasmania, 2022, *Health in Tasmania: Primary Health Tasmania Health Needs Assessment 2022-23 to 2024-25 - November 2022 (Update)*, <https://www.primaryhealthtas.com.au/wp-content/uploads/2023/05/Health-in-Tasmania-Comprehensive-Needs-Assessment-2022-25.pdf>.
39. Tasmanian Department of State Growth, *Transport Access Strategy*, viewed 21 June 2024, <https://www.stategrowth.tas.gov.au/__data/assets/pdf_file/0007/174076/Transport_Access_Strategy.PDF>.
40. Australan Bureau of Statistics, 2021, *Socio-Economic Indexes for Australia (SEIFA), 2021*, <https://www.abs.gov.au/statistics/people/people-and-communities/socio-economic-indexes-areas-seifa-australia/latest-release>.

41. Tasmanian Department of Health, 2019, *Health Literacy Action Plan 2019-2024*, accessed 21 June 2024, <https://www.health.tas.gov.au/sites/default/files/2021-11/Health_Literacy_Action_Plan_DoHTasmania2019.pdf>.
42. Elmer, S., Cheng, C., & Osborne, R., 2022, *Optimising Health Care for Tasmanians,* Swinburne University of Technology, <https://doi.org/10.26185/A52N-GQ19>.
43. Choukair, D., et al., 2021, 'An Integrated clinical pathway for diagnosis, treatment and care of rare diseases: model, operating procedures, and results of the project TRANSLATE-NAMSE funded by the German Federal Joint Committee*', Orphanet Journal of Rare Diseases*, 16 (1), p. 474.
44. Department of Health Tasmanian Government, 2020, *Our Healthcare Future: Immediate actions and consultation paper*, accessed 21 June 2024, <https://www.health.tas.gov.au/sites/default/files/2021-12/Our_Healthcare_Future_Immediate_actions_and_consultations_paper_DoHTasmania2021.pdf>.
45. Wells, C.E. and S.J. Smith, 2017, 'Diagnostic Care Pathways in Dementia', *J Prim Care Community Health*, 8 (2), pp. 103-111.
46. Zanello, G., C.H. Chan, and D.A. Pearce, 2022, 'Recommendations from the IRDiRC Working Group on methodologies to assess the impact of diagnoses and therapies on rare disease patients', *Orphanet Journal of Rare Diseases*, 17 (1), p. 181.
47. Allen, D., E. Gillen, and L. Rixson, 2009, 'The Effectiveness of Integrated Care Pathways for Adults and Children in Health Care Settings: A Systematic Review', *JBI Libr Syst Rev*, 7 (3), pp. 80-129.
48. Schrijvers, G., A. van Hoorn, and N. Huiskes, 2012, 'The care pathway: concepts and theories: an introduction', *Int J Integr Care*, 12 (Spec Ed Integrated Care Pathways), p. e192.
49. Tumiene, B. and H. Graessner, 2021, 'Rare disease care pathways in the EU: from odysseys and labyrinths towards highways', *J Community Genet*, 12 (2), pp. 231-239.
50. Blashki, G., S. Metcalfe, and J. Emery, 2014, 'Genetics in general practice', *Aust Fam Physician*, 43 (7), pp. 428-31.
51. Mattick, J.S., et al., 2018, 'Whole genome sequencing provides better diagnostic yield and future value than whole exome sequencing', *The Medical Journal of Australia*, 209 (5), pp. 197-199.
52. Adachi, T., et al., 2023, 'Enhancing Equitable Access to Rare Disease Diagnosis and Treatment around the World: A Review of Evidence, Policies, and Challenges', *Int J Enivron Res Public Health*, 20 (6), p. 4732.
53. Iriart, J.A.B., et al., 2019, 'From the search for diagnosis to treatment uncertainties: challenges of care for rare genetic diseases in Brazil', *Cien Saude Colet*, 24 (10), pp. 3637-3650.
54. Benito-Lozano, J., et al., 2022, 'Diagnostic Process in Rare Diseases: Determinants Associated with Diagnostic Delay', *Int J Environ Res Public Health*, 19 (11).
55. Isono, M., M. Kokado, and K. Kato, 2022, 'Why does it take so long for rare disease patients to get an accurate diagnosis?—A qualitative investigation of patient experiences of hereditary angioedema', *PLoS ONE*, 17 (3), p. e0265847.
56. Shire, 2013, *Rare disease impact report: insights from patients and the medical community*, <https://globalgenes.org/wp-content/uploads/2013/04/ShireReport-1.pdf>.
57. Blöß, S., et al., 2017, 'Diagnostic needs for rare diseases and shared prediagnostic phenomena: Results of a German-wide expert Delphi survey', *PLoS One*, 12 (2), p. e0172532.
58. Zhu, Q., et al., 2021, 'Scientific evidence based rare disease research discovery with research funding data in knowledge graph', *Orphanet Journal of Rare Diseases*, 16 (1), p. 483.
59. Gordon, J., et al., 2022, General Practice Statistics in Australia: Pushing a Round Peg into a Square Hole, *Int J Environ Res Public Health*, 19 (4).
60. Knight, A.W. and T.P. Senior, 2006, 'The common problem of rare disease in general practice', *Med J Aust*, 185 (2), pp. 82-3.
61. McMullan, J., et al., 2021, 'Perceptions and experiences of rare diseases among General Practitioners: an exploratory study', *medRxiv*, p. 2021.09.07.21263025.
62. Best, S., et al., 2023, 'Supporting healthcare professionals to offer reproductive genetic carrier screening: a behaviour change theory approach', *Aust J Prim Health*, 29 (5), pp. 480-489.
63. United Kingdom Department of Health, 2013, *The UK Strategy for Rare Disease*, <https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/260562/UK_Strategy_for_Rare_Diseases.pdf>.

64. Elsum, I., et al., 2020, 'A community-based co-designed genetic health service model for Aboriginal Australians', *PLoS ONE*, 15 (10), p. e0239765.
65. D'Angelo, C.S., et al., 2020, 'Barriers and Considerations for Diagnosing Rare Diseases in Indigenous Populations', *Front Pediatr*, 8, p. 579924.
66. Sandquist, M., et al., 2022, 'The Transition to Adulthood for Youth Living with Rare Diseases', *Children* (Basel), **9** (5).
67. Berens, J.C., et al., 2017, 'Young adults with chronic illness: how can we improve transitions to adult care?', *Pediatrics*, 139 (5).
68. Teutsch, S., et al., 2023, 'Australian children living with rare diseases: health service use and barriers to accessing care', *World J Pediatr*, 19 (7), pp. 701-709.
69. Australian Genomics, 2019, *Clinical Genomics: Investigating perceptions of unmet need in Australia. An Initial Study,* National Diagnostic And Research Network Program, Australian Genomics, <https://www.australiangenomics.org.au/wp-content/uploads/2021/09/Unmet-Needs-Report.pdf>.
70. The McKell Institute, 2021, *2021 Progress Update. Funding Rare Disease Therapies in Australia: Ensuring equitable access to health care for all Australians*, Sydney, Australia.
71. Zurynski, Y., et al., 2017, 'Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays', *Orphanet Journal of Rare Diseases*, 12 (1), p. 68.
72. Jaeger, G., et al., 2022, 'Consequences of rare diagnoses for education and daily life: development of an observation instrument', *Orphanet Journal of Rare Diseases*, 17 (1), p. 165.
73. Navarrete-Opazo, A.A., et al., 2021, 'Can you hear us now? The impact of health-care utilization by rare disease patients in the United States', *Genet Med*, 23 (11), pp. 2194-2201.
74. Zurynski, Y.A., et al., 2007, 'Impacts of national surveillance for uncommon conditions in childhood', *J Paediatr Child Health*, 43 (11), pp. 724-31.
75. Castro, R., et al., 2017, 'Bridging the Gap between Health and Social Care for Rare Diseases: Key Issues and Innovative Solutions', in *Rare Diseases Epidemiology: Update and Overview*, M. Posada de la Paz, D. Taruscio, and S.C. Groft (eds), Springer International Publishing: Cham, pp. 605-627.
76. Nuffield Department of Primary Care Health Sciences, 2023, *Suspected CANcer (SCAN) Diagnostic Pathway*, accessed 1 May 2023, <https://www.phc.ox.ac.uk/research/cancer-research-group/suspected-cancer-scan-diangostic-pathway>.
77. Kitchiner, D., C. Davidson, and P. Bundred, 1996, 'Integrated care pathways: effective tools for continuous evaluation of clinical practice', *J Eval Clin Pract*, 2 (1), pp. 65-9.
78. Allen, D., E. Gillen, and L. Rixson, 2009, 'Systematic review of the effectiveness of integrated care pathways: what works, for whom, in which circumstances?', *Int J Evid Based Health*, 7 (2), pp. 61-74.
79. Department of Health Victorian Government, 2023, *Optimal care pathways*, accessed 2 May 2023, <https://www.health.vic.gov.au/health-strategies/optimal-care-pathways>.
80. Cancer Council Victoria and Commonwealth Government, 2022, *Optimal care pathway for people with neuroendocrine tumours 1st Edition,* Cancer Council Victoria, Melbourne
81. Ward, A.J., et al., 2022, 'Designing rare disease care pathways in the Republic of Ireland: a co-operative model', *Orphanet Journal of Rare Diseases*, 17 (1), p. 162.
82. NSW Government Agency for Clinical Innovation, 2021, *Clinical genomics Model of Care: Organisational Model of Care*, ACI, Sydney.
83. Baynam, G., et al., 2017, 'Initiating an undiagnosed diseases program in the Western Australian public health system', *Orphanet Journal of Rare Diseases*, 12 (1), p. 83.
84. Australian Bureau of Statistics, 2022, *Patient Experiences*, <https://www.abs.gov.au/statistics/health/health-services/patient-experiences/2021-22>.
85. Howes F, A.S., Lin L, Kitsos A, Shaw K, 2020, *General Practice in Tasmania 2019*, Primary Health Tasmania.
86. Medical Board of Australia, 2023, *Registrant Data June 2023*, <https://www.medicalboard.gov.au/News/Statistics.aspx>.
87. Human Genetics Society of Australasia, 2022, *Core Capabilities in Genetics & Genomics for Medical Graduates*, <https://hgsa.org.au/common/Uploaded%20files/pdfs/policies,%20position%20statements%20and

%20guidelines/eesic/Core%20Capabilities%20in%20Genetics%20and%20Genomics%20for%20Medical%20Graduates.pdf>.
88. Augustine, E.F., E.R. Dorsey, and P.L. Saltonstall, 2017, The Care Continuum: An Evolving Model for Care and Research in Rare Diseases', *Pediatrics*, 140 (3).