Australian Genomics Health Alliance

Australian Genomics: Genomics in the Community Project

Landscape Analysis Report

Authors:

Miranda Lewit-Mendes[1,2], Keri Pereira[1,2], Kate Dunlop[1,3], Bronwyn Terrill[1,4,5] and Tiffany Boughtwood[1,2].

Contributors:

Genomics in the Community working group: Zornitza Stark[1,2], Richard Vines[6], John Cannings[6], Shane Porter[7], Marie Ludlow[8], Heather Renton[9].

Affiliations:

1. Australian Genomics
2. Murdoch Children's Research Institute
3. Centre for Genetics Education NSW Health
4. UNSW, Sydney
5. Genome.One
6. Rare Cancers Australia
7. Department of Health
8. Kidney Health Australia
9. Syndromes Without A Name

MAY 2018

Overview:

The Australian Genomics Health Alliance ("Australian Genomics") is a national collaborative initiative working toward the optimal approach to integrating genomic medicine into healthcare across Australia. Funded principally by an NHMRC Targeted Call for Research grant of $25 million over a five-year period, Australian Genomics is working with State, Territory and Federal Governments, Patient Advocates and the research community. Our purpose is to demonstrate an equitable, efficient and ethical approach to clinical genomics to the benefit of all Australians.

The Genomics in the Community Project is an initiative to develop public and patient-orientated information materials about genomics, in order to ensure reliable and current information is provided to the community. Australian Genomics and Patient Advocacy Groups are working together to develop accessible materials about genomics for their patients and the wider community. Project workshops have established content themes, as well as key issues and concerns of the general public, which will be a focus of the materials provided.

Aims:

We undertook a landscape analysis to establish the quality and content of publically available information materials for patients relating to genomic medicine. From this, we can determine what can be used or adapted for the Australian context, and where community information needs are unmet. This will inform our targeted development of resources for patients, initially, and ultimately for the broader Australian community.

The landscape analysis also aims to engage and involve Patient Advocacy Groups to determine which of the available existing materials are most appropriate and useful for the target audiences.

Methods:

Literature search

A PubMed literature search was used to establish whether any previous analyses had been conducted on the landscape of information materials relating to genomics. Past literature related primarily to evaluation of the genomic education of healthcare professionals (HCPs) or genomic materials targeted at HCPs (Demmer & Waggoner, 2014; Slade, Subramanian, & Burton, 2016; Talwar, Tseng, Foster, Xu, & Chen, 2017; Weitzel, Aquilante, Johnson, Kisor, & Empey, 2016; Zhang, Zhang, Ling, & Jia, 2015). Past literature also indicated a need for patient education regarding genomic testing but did not evaluate any patient-targeted information materials (Blanchette et al., 2014; Cuffe et al., 2014). We were not able to locate any previously published analyses on genomic information materials targeted to a patient audience.

Establishing major topics / themes

Initial project workshops with collaborating Patient Advocates established the main content themes and issues of interest for patients when seeking information on genomic testing. These one-day workshops were conducted in Sydney (16 August 2017) and Melbourne (18 August 2017). The representation at the workshops included cancer and rare disease advocacy groups, Australian Digital Health Agency, Commonwealth

Department of Health, ethicists, genetics/genomics educators, flagship clinicians, genetic counsellors and Australian Genomics.

The main content themes were:

(a) Basic genomics;
(b) Relevance of genomics;
(c) Reasons for undertaking genetic/genomic testing; and
(d) The benefits/risks of undertaking genetic/genomic testing.

Key issues and concerns of the community were identified as:

(1) Insurance;
(2) Family communication regarding testing, results and implications;
(3) Cost;
(4) Data security; and
(5) Mental health.

These themes and issues became the search terms to identify web-accessible genomic information materials.

Locating existing materials

The Australian Genomics partners list of over 80 state, national, international and global institutions was used to identify key websites that could contain patient materials relating to genomics. After exhausting this list, desktop research using Google was conducted using key terms, such as "genomics", "genomic testing" and "patient materials".

Information materials were identified from 38 different sources, including companies offering genomic testing, research centres, Patient Advocacy Groups, government bodies, hospitals, genetic counselling services and educational websites. Written, graphic and video materials were all identified.

Framework for critical analysis of materials

A framework for the critical analysis of existing materials was created for this study, based on established tools for assessing patient educational materials and with input from experts in the field (Charnock, Shepperd, Needham, & Gann, 1999; MaineHealth, 2010; Shoemaker SJ). Separate frameworks were created for written materials (with or without accompanying graphics), stand-alone graphics and videos.

The frameworks include criteria for evaluating the materials in five areas:

(1) Content;
(2) Language and readability;
(3) Structure and organisation;
(4) Design and;
(5) Neutrality and balance.

Each material was scored, and percentages were used to compare written, graphic and video materials. Materials were categorised by topic, and were also assigned a letter, based on their relevance for patients (A) before, (B) during or (C) after genomic testing. The evaluation was completed by one person to ensure consistency across all materials.

Critical analysis using framework

Each information material was evaluated using the appropriate framework (written, graphical, video) and the percentage of criteria satisfied was calculated.

This percentage 'score' was used to rank the materials into three tiers:

a) 75-100%: meets most criteria
b) 50-74%: meets some criteria
c) < 50%: did not meet criteria satisfactorily

Survey for collaborator consultation

On completion of the critical analysis, the perspectives of collaborating Patient Advocacy Groups were sought via an online survey. The Australian Genomics community advisory group members and operational personnel were also given the opportunity to participate in this survey and input was sought from experts in the field.

The survey was produced using Research Electronic Data Capture (REDCap) tools hosted at MCRI (Harris et al., 2009). REDCap is a web application, used for building online surveys, and securely storing and exporting data. Due to the large number of materials in the landscape analysis, only materials that satisfied at least 70% of criteria were selected for inclusion in the survey. To minimise survey fatigue, three separate surveys were designed containing links to approximately one third of the top materials.

The survey initially presented the framework for evaluation as a reference for why these materials were selected as being in the top 30%. Following this, links to each material or embedded videos were provided for the groups to view. They were then asked to indicate whether they found the material to be useful, not useful or neutral. A free text comment box was provided for each material.

The surveys were distributed via email to the Patient Advocacy Group representatives, Community Advisory Group members and operational personnel. Each collaborator was randomly allocated to one of the three surveys, which was available to them via email from 24 January 2018 until 16 February 2018.

Survey analysis

The quantitative responses from the survey regarding usefulness of patient materials were evaluated by ranking each material, based on the percentage of yes, neutral and no responses that they received.

A thematic analysis was conducted on the comments received regarding the materials, in order to identify key themes that collaborators considered important to the usefulness of patient materials.

Results:

Critical analysis

The desktop research identified 138 relevant information materials, including 124 written, three stand-alone graphic and 11 video materials. The mean percentage score for the materials was 60.0% (with 75-100% indicating materials meet most criteria of the patient information framework developed for this study), with 108/138 (78.3%)

materials scoring at least 50%, 61/138 (44.2%) materials scoring at least 66.7% and 20/138 (14.5%) materials scoring over 75%.

Many materials satisfied some aspects of the criteria well, while few materials managed to satisfy all five aspects of the criteria satisfactorily. A large number of materials focused on basic information relating to genetics and genetic testing. There were few materials relating to genomics and genomic testing.

Survey

The survey was circulated to representatives from Patient Advocacy Groups (25), who were involved in the workshops or expressed interest in this project, Community Advisory Group members (8) and Australian Genomics operational personnel (34). A total of 44 responses were received across the three surveys; 19 full responses, 5 partial, 9 accidental duplicates and 11 blank surveys. The three surveys respectively had response rates of 42.1%, 20.8% and 45.8%, with an average response rate overall of 36.3%.

The results were mostly positive towards the materials, with 6 materials receiving a unanimous 'yes' for usefulness, 30 materials receiving over 50% 'yes' responses, and 10 scoring 50% or less 'yes' responses. The response provided for each material is shown in **Table 1,** ranked based on the percentage of 'yes' responses they received.

Table 1: Collaborator opinions on the usefulness of materials.

Material title/link	Source	Category	Survey responses	Usefulness		
				YES	Neutral	NO
Inheriting genetic conditions (chapter)	**Genomics Home Reference**	Basic genetics/ genomics	4	4 100%	0	0
Understanding cancer genomics	**Genomics England**	Basic genetics/ genomics	8	8 100%	0	0
What is genomics	**Genomics British Columbia**	Basic genetics/ genomics	4	4 100%	0	0
What is genomics?	**Melbourne Genomic Health Alliance**	Basic genetics/ genomics	11	11 100%	0	0
Data in the 100,000 Genomes Project	**Genomics England**	Data sharing	8	8 100%	0	0
Ethical issues in human genetics and genomics	**NSW Health Centre for Genetics Education**	Ethics	4	4 100%	0	0
What is a genetic test?	**Syndromes Without A Name**	Genetic/ genomic testing	9	8 88.9%	1 11.1%	0
Getting your genetic test results	**Cancer Research UK**	Outcomes of genetic/ genomic testing	9	8 88.9%	1 11.1%	0
Talking to your family about a genetic diagnosis or test result	**NSW Health Centre for Genetics Education**	Talking to your family	9	8 88.9%	1 11.1%	0
Genes, DNA and cancer	**Cancer Research UK**	Basic genetics/ genomics	8	7 87.5%	1 12.5%	0
Genetic services in Victoria	**Better Health Channel**	Genetic counselling/ genetics services	8	7 87.5%	1 12.5%	0
Frequently Asked Questions about genetic testing	**National Human Genome Research Institute**	Genetic/ genomic testing	8	7 87.5%	1 12.5%	0
Genes and genetics	**Better Health Channel**	Basic genetics/ genomics	11	9 81.8%	2 18.2%	0
Genetic testing for mitochondrial disease	**Australian Mitochondrial Disease Foundation**	Basic genetics/ genomics	5	4 80%	1 20%	0
Introduction to genetics	**Learning Genetics**	Basic genetics/ genomics	9	7 77.8%	1 22.8%	1
Information about your genetics appointment	**Syndromes Without A Name**	Genetic counselling/ genetics services	9	7 77.8%	1 22.8%	1

Talking about BRCA in your family tree	**Facing Our Risk**	Talking to your family	9	7 77.8%	1 22.8%	1
Gene mutations	**NSW Health Centre for Genetics Education**	Basic genetics/ genomics	8	6 75%	2 25%	0
Frequently Asked Questions about genetic counseling	**National Human Genome Research Institute**	Genetic counselling/ genetics services	8	6 75%	2 25%	0
Genetic testing	**Healthdirect**	Genetic/ genomic testing	4	3 75%	1 25%	0
Variations in the genetic code	**NSW Health Centre for Genetics Education**	Basic genetics/ genomics	8	6 75%	1 12.5%	1 12.5%
Insurance	**Genomics England**	Insurance	8	6 75%	1 12.5%	1 12.5%
What is pharmacogenomics?	**PharmaKB**	Pharmacogenetics/ Pharmacogenomics	8	6 75%	2 25%	0
Talking to children about genetic conditions	**Nottingham Regional Clinical Genetics Service**	Talking to your family	8	6 75%	2 25%	0
Family health history: the basics	**Centre for Disease Control and Prevention**	Talking to your family	8	6 75%	2 25%	0
Shared risk: talking to family members about genetic test results	**Facing Our Risk**	Talking to your family	4	3 75%	0	1 25%
Benefits and risks of genetic testing	**Genetic Alliance UK**	Genetic/ genomic testing	9	6 66.7%	3 33.3%	0
Taking part in the 100,000 Genomes Project	**Genomics England**	Genetic/ genomic testing	9	6 66.7%	3 33.3%	0
Frequently Asked Questions about rare diseases	**National Human Genome Research Institute**	Genetic disorders	8	5 62.5%	3 37.5%	0
An introduction to DNA, genes and chromosomes	**NSW Health Centre for Genetics Education**	Basic genetics/ genomics	8	5 62.5%	2 25%	1 12.5%
Whole exome sequencing	**Iowa Institute of Human Genetics**	Genetic/ genomic testing	4	2 50%	2 50%	0
What you need to know about pharmacogenomic testing	**Mayo Clinic**	Pharmacogenetics/ Pharmacogenomics	4	2 50%	2 50%	0
Whole genome sequencing and you	**Icahn School of Medicine**	Basic genetics/ genomics	10	5 50%	3 30%	2 20%
My genome sequence	**Great Ormond Street Hospital and Charity**	Basic genetics/ genomics	4	2 50%	1 25%	1 25%
Medical testing: health information for you and your family	**National Health and Medical Research Council**	Basic genetics/ genomics	4	2 50%	1 25%	1 25%
Genetic counselling for mitochondrial disease	**Australian Mitochondrial Disease Foundation**	Genetic counselling/ genetics services	4	2 50%	1 25%	1 25%
Cells and DNA (chapter)	**Genomics Home Reference**	Basic genetics/ genomics	11	5 45.5%	5 45.5%	1 9%
A guide to genetic tests that are used to examine many genes at the same time	**The European Society of Human Genetics**	Genetic/ genomic testing	8	3 37.5%	5 62.5%	0
Whole exome sequencing	**Sickkids**	Genetic/ genomic testing	4	1 25%	2 50%	1 25%
Life insurance products and genetic testing in Australia	**NSW Health Centre for Genetics Education**	Insurance	4	0	3 75%	1 25%

Thematic analysis

There were four predominant themes that emerged during thematic analysis of comments on the usefulness of patient materials:

(a) Complexity of content and language;
(b) Amount of content;
(c) Graphics/videos/visual learning and;
(d) Relevance to target population.

Comments relating to the complexity of content and language mostly centred around the content being too detailed or complex for a patient audience, or that the content was clear and easy to understand. The comments regarding complexity of content were not always consistent for each material between reviewers. The amount of content was often mentioned, with most comments indicating a material had too much text or an overwhelming amount of content. Visual learning was a prominent theme; comments praised the use of video content over text and a large number comments suggested the use of graphics in materials without them. The theme of relevance to the target population was less commonly seen but appeared consistently in relation to materials containing information that may differ between countries or population groups.

Discussion:

Very few materials were identified that focus primarily on genomics and genomic testing; however, a large number of materials about genetics and genetic testing are available for patients that may have the potential to be adapted for a genomic context. Materials with information pertaining to insurance and data privacy were also lacking and may differ between countries. Therefore, it may be necessary to adapt or produce new materials that contain information relevant to patients in Australia.

The critical analysis revealed that few existing materials were able to satisfy all aspects of the defined criteria. This indicates it may be necessary to produce new materials, in order to ensure information provided to patients is of the highest standard and satisfies all evaluation criteria. If existing materials are to be used, it may be necessary to identify which parts of the criteria are the most important for the specific target population, in order to ensure materials provided are of adequate standard.

The survey revealed that collaborators found majority of the materials evaluated in the survey to be useful overall, however there were specific concerns highlighted in their comments. The most predominant themes arising from the comments were content complexity, the amount of content, use of visual content and relevance to target populations. Despite most materials being selected as useful, there were a large number of comments indicating the content was too comprehensive or dense. Most materials received relatively consistent comments regarding their complexity, however there were some inconsistencies. These inconsistencies could be due to the role of the collaborator, their knowledge of the area and who their target audience would be. Therefore, when choosing appropriate and useful materials, it will be vital to consider the needs of target audience and the range of needs even within that target audience. Visual content was consistently indicated to be preferred over non-visual content, so there may be a need to develop more videos/graphics to accompany or substitute written materials.

Limitations:

The identification of materials is inherently limited by the search engines used and continual release of new materials, so it is not possible to ensure every material was captured. Due to continual release and updating of materials, it will be necessary to periodically review materials provided to the target audience.

The evaluation of existing materials is limited by human error, as some criteria is objective and complete consistency between materials cannot be guaranteed. A

limitation of the survey was that the information provided at the start of the survey was not always read or accurately understood. A number of collaborators indicated in the comments that they believed that they had received all the existing materials for each topic, when they had actually only been provided with a selection of the top scoring materials. Therefore, given that they did not read the introductory information carefully, there is also a risk that they may have not reviewed the patient materials in detail.

Acknowledgements:

We would like to acknowledge the Patient Advocacy Groups who contributed their time to this project:

Australasian Gastro-Intestinal Trials Group,

Australian Mitochondrial Disease Foundation,

Breast Cancer Network Australia,

CanTeen,

Cystic Fibrosis Community Care,

Cystic Fibrosis NSW,

Genetic and Rare Disease Network,

Kidney Health Australia,

Leukaemia Foundation,

Leukodystrophy Australia,

Myeloma Australia,

Rare Voices Australia,

Rare Cancers Australia, and

Syndromes Without A Name Australia.

We would also like to acknowledge all the individuals who took part in the workshops, as well as the survey participants.

Australian Genomics Health Alliance is funded by the NHMRC (GNT1113531)

References:

Blanchette, P. S., Spreafico, A., Miller, F. A., Chan, K., Bytautas, J., Kang, S., . . . Siu, L. L. (2014). Genomic testing in cancer: patient knowledge, attitudes, and expectations. *Cancer, 120*(19), 3066-3073. doi:10.1002/cncr.28807

Charnock, D., Shepperd, S., Needham, G., & Gann, R. (1999). DISCERN: an instrument for judging the quality of written consumer health information on treatment choices. *Journal of Epidemiology and Community Health, 53*(2), 105-111.

Cuffe, S., Hon, H., Qiu, X., Tobros, K., Wong, C. K., De Souza, B., . . . Liu, G. (2014). Cancer patients acceptance, understanding, and willingness-to-pay for pharmacogenomic testing. *Pharmacogenetics and Genomics, 24*(7), 348-355. doi:10.1097/fpc.0000000000000061

Demmer, L. A., & Waggoner, D. J. (2014). Professional medical education and genomics. *Annual Review of Genomics and Human Genetics, 15*, 507-516. doi:10.1146/annurev-genom-090413-025522

Harris, P. A., Taylor, R., Thielke, R., Payne, J., Gonzalez, N., & Conde, J. G. (2009). Research electronic data capture (REDCap)--a metadata-driven methodology and workflow process for providing translational research informatics support. *Journal of Biomedical Informatics, 42*(2), 377-381. doi:10.1016/j.jbi.2008.08.010

MaineHealth. (2010). A Guide to Creating and Evaluating Patient Materials: Guidelines for Effective Print Communication. Retrieved from https://mainehealth.org/healthcare-professionals/community-education-program/health-literacy/tools-for-health-literacy:

Shoemaker SJ, W. M., Brach C. The Patient Education Materials Assessment Tool (PEMAT) and User's Guide. (Prepared by Abt Associates, Inc. under Contract No. HHSA290200900012I, TO 4). Retrieved from Rockville, MD: Agency for Healthcare Research and Quality; November 2013.

Slade, I., Subramanian, D. N., & Burton, H. (2016). Genomics education for medical professionals - the current UK landscape. *Clinical Medicine, 16*(4), 347-352. doi:10.7861/clinmedicine.16-4-347

Talwar, D., Tseng, T. S., Foster, M., Xu, L., & Chen, L. S. (2017). Genetics/genomics education for nongenetic health professionals: a systematic literature review. *Genetics in Medicine, 19*(7), 725-732. doi:10.1038/gim.2016.156

Weitzel, K. W., Aquilante, C. L., Johnson, S., Kisor, D. F., & Empey, P. E. (2016). Educational strategies to enable expansion of pharmacogenomics-based care. *American Journal of Health-System Pharmacy, 73*(23), 1986-1998. doi:10.2146/ajhp160104

Zhang, G., Zhang, Y., Ling, Y., & Jia, J. (2015). Web Resources for Pharmacogenomics. *Genomics, Proteomics & Bioinformatics, 13*(1), 51-54. doi:10.1016/j.gpb.2015.01.002