Clinical Dataset

Study number:	
Recruitment site:	
Site:	☐ NICU Gestational age (weeks): _____ Birth weight (grams): _____ ☐ PICU ☐ Other: Specify reason for urgency: _____ _____
Date of hospital admission:	
Date of ICU admission (if different):	
Date of clinical genetics referral:	
Date of clinical genetics consult:	
Referring Clinician Details:	
First name:	
Surname:	
Email:	
Phone number:	
Clinical geneticist (if different):	
Additional Clinicians e.g. NICU/PICU consultant, sub-specialists. These clinicians will receive: • Updates on the progress of the case • A copy of the report • A survey to collect the clinical outcomes one-month post-result The genetic counsellor completing the consent will automatically be included.	
Clinician 1:	Name: _____ Role: _____ Email: _____
Clinician 2:	Name: _____ Role: _____ Email: _____

Clinician 3:	Name: _____ Role: _____ Email: _____
Clinician 4:	Name: _____ Role: _____ Email: _____
Patient details	
Unit Record (UR) number:	
Genetic File number (if available):	
First name:	
Surname:	
Sex	☐ Male ☐ Female ☐ Other
DOB:	
Address:	Street name: _____ Suburb: _____ State: _____ Postcode: _____
Biological Mother details	
First name:	
Surname:	
DOB:	
Is the Biological Mother's address the same as the patient?	☐ Yes ☐ No If no: Street name: _____ Suburb: _____ State: _____ Postcode: _____
Ethnicity: ** uses HANCESTRO ontology*	
Biological Father details	
First name:	
Surname:	
DOB:	
Is the Biological Father's address the same as the patient?	☐ Yes ☐ No If no: Street name: _____ Suburb: _____ State: _____ Postcode: _____

Ethnicity: ** uses HANCESTRO ontology*	
Clinical Information Start with the most prominent feature. Aim for 5-10 HPO terms.	
Clinical features: Aim for 5-10 positive/negative (e.g. microcephaly, seizures, not dysmorphic) ** HPO Capture Field - Capacity to capture multiple entries*	
Is the onset of the condition congenital?	☐ Yes ☐ No If no, age of onset: _____
Pedigree and Family History Use the tool to draw the core family unit including the proband and first degree relatives, in particular noting consanguinity and any first degree relatives who are similarly affected as this will assist us in genomic analysis. Only include extended family members if there is a significant family history of a genetic condition relevant to the analysis.	
Pedigree **online pedigree drawing tool plugin*	
Investigations Provide details of relevant prior investigations that may assist in the analysis, in particular microarray result, but also imaging, complex biochemistry or biopsy results.	
Microarray result:	☐ Pending ☐ Normal ☐ Abnormal If abnormal, provide details: _____ _____

Other relevant investigations or clinical information (not entered above):	
Virtual Panels for this Analysis Please select relevant virtual panels to guide the analysis. All patients will have Mendeliome analysis, including analysis for copy number variants and variants in the mitochondrial genome. For details on the gene content of panels, please go to PanelApp Australia.	
Do you strongly suspect a specific clinical diagnosis?	☐ Yes ☐ No If yes, specify gene name(s): _____ _____
Any other additional information not provided above:	

Clinical Impact of Result

Result:	☐ A diagnosis was made ☐ A partial diagnosis was made ☐ More than 1 diagnosis was made ☐ A diagnosis was not made
Date patient discharged from hospital:	
Date of death (if relevant):	
Date of discharge from ICU (if relevant):	
Changes in patient management arising from result	
Medication started	☐ Yes ☐ No Details: _____ _____
Medication stopped	☐ Yes ☐ No Details: _____ _____
Medication adjusted	☐ Yes ☐ No Details: _____ _____
Investigation cancelled	☐ Yes ☐ No Details: _____ _____
Additional investigation ordered	☐ Yes ☐ No Details: _____ _____
Subspecialist referral initiated	☐ Yes ☐ No Details: _____ _____
Prior subspecialist service no longer required	☐ Yes ☐ No Details: _____ _____
Surgical procedure initiated (incl biopsy)	☐ Yes ☐ No Details: _____ _____

Surgical procedure cancelled	☐ Yes ☐ No Details: _____ _____
Surgical procedure changed	☐ Yes ☐ No Details: _____ _____
Management redirected towards palliation	☐ Yes ☐ No Details: _____ _____
Decision to palliate reversed	☐ Yes ☐ No Details: _____ _____
Patient eligibility for a new research study affected	☐ Yes ☐ No Details: _____ _____
Was there a change in management as a result of the genomic testing in this patient?	☐ Yes ☐ No Details: _____ _____
Additional family members tested (e.g. sibs)	☐ Yes ☐ No If yes, outcome: _____ _____
Reproductive risk established for parents?	☐ Yes ☐ No If yes: ☐ <1% ☐ 25% ☐ 50% ☐ Other
How do you rate the clinical utility of genomic testing for this patient?	☐ Neutral ☐ Useful ☐ Very useful ☐ Not useful at all ☐ Not very useful
Do you think the length of ICU stay was shortened by genomic testing?	☐ Yes ☐ No If so, by how many days? _____ Explain: _____ _____
Do you think the length of ICU stay was extended by genomic testing?	☐ Yes ☐ No If so, by how many days? _____ Explain: _____ _____

Genomic results (tick all that apply)	☐ Enabled cessation of additional testing ☐ Required additional testing to confirm diagnosis ☐ Allowed avoidance of complications ☐ Required additional testing to screen for complications ☐ Enabled targeted treatment that may improve long-term outcomes ☐ Enabled improved communication of outcomes/expectations/prognosis with the family ☐ Decreased stress and confusion for the family ☐ Increased stress and confusion for the family ☐ Decreased confusion among medical staff ☐ Increased confusion among medical staff ☐ Resulted in a diagnosis not fully understood at this time Comment: _____ _____ _____ _____ _____