Australian Genomics

Clinical Dataset

1. Study number:	
2. Site:	☐ NICU ☐ PICU ☐ Other: If other, specify reason for urgency: _____ _____
3. Name of referring clinician:	
4. Name of clinical geneticist (if different):	
5. Name:	
6. Hospital UR No:	
7. DOB:	
8. Sex:	
9. Age of patient (years, months):	
10. Gestational age (weeks):	
11. Birth weight (grams):	
12. Relevant pregnancy information free text (e.g. increased NT, gestational diabetes, possible HIE): ** HPO Capture Field - Capacity to capture multiple entries*	
13. Parental consanguinity	☐ Yes ☐ No
14. Maternal ethnicity:	
15. Paternal ethnicity	
16. Affected family members?	☐ Yes ☐ No

If Yes: Mother   ☐ No ☐ Yes, number:
         Father   ☐ No ☐ Yes, number:
         Siblings ☐ No ☐ Yes, number:	

17. Microarray result:	☐ Pending ☐ Normal ☐ Abnormal If abnormal, provide details: _____ _____
18. Principal phenotypic features: Aim for 5-10 positive/negative (e.g. microcephaly, seizures, not dysmorphic) ** HPO Capture Field - Capacity to capture multiple entries*	

Genes to be prioritized for analysis based on phenotype:
1. For some cases, there will be a <u>shortlist</u> of 1-5 genes with high clinical index of suspicion: please highlight these for the laboratory team
2. All cases will have one or more <u>virtual gene panels</u> prioritized for analysis (choose from laboratory's catalogue)

All cases will have <u>Mendeliome</u> analysis, and if negative, outside of Mendeliome

If rapid WES/WGS was not available, would you order a single gene or panel test?
☐ Yes ☐ No
If Yes, which one: _____
_____
If rapid WES/WGS was not available, what other investigations would you order in the next 2 weeks?
_____
_____
_____

Clinical Outcomes

1. Date of birth:	
2. Date of hospital admission:	
3. Date of ICU admission:	
4. Date of clinical genetics referral:	
5. Date of clinical genetics consult:	
6. Date proposed for Acute Care WES/WGS:	
7. Date accepted for Acute Care WES/WGS:	
8. Date consent obtained:	
9. Date result disclosed:	
10. Total number of clinical genetics inpatient consultations:	☐ 1 ☐ 2 ☐ 3 ☐ 4 ☐ >5 Dates and durations of consultations: Consult 1: Consult 2: Consult 3: Consult 4: Consult 5:
11. Date of discharge from ICU:	
12. Date of discharge from hospital:	
13. Date of death:	
14. Comments/Notes: 15. e.g. sources of delay in recruitment (for example patient initially stable, deteriorated unexpectedly on D20 of admission, transferred to ICU, WES initiated then)	
Laboratory Timeline (to be collected by labs)	
1. Date/time samples and consent received:	
2. Date/time library prep initiated:	
3. Date/time library prep completed:	
4. Date/time sequencing initiated:	
5. Date/time sequencing completed:	
6. Date/time bioinformatics analysis initiated:	
7. Date/time bioinformatics analysis complete:	
8. Date/time variant analysis initiated:	
9. Date/time variant analysis complete:	
10. Date/time reviewed by MDT:	
11. Date/time report issued:	
Clinical impact of result	
1. Molecular diagnosis made?	☐ Yes ☐ No

2.	How many genes were implicated	

Number of genes implicated: ☐ 1 ☐ 2 ☐ 3

Gene 1:	Condition (OMIM number) 1:
Gene 2:	Condition (OMIM number) 2:
Gene 3:	Condition (OMIM number) 3:

Report upload in RedCap

3.	**Changes in patient management arising from result**	
i.	Medication started	☐ Yes ☐ No
	Details: _____	
	_____	
ii.	Medication stopped	☐ Yes ☐ No
	Details: _____	
	_____	
iii.	Medication adjusted	☐ Yes ☐ No
	Details: _____	
	_____	
iv.	Investigation cancelled	☐ Yes ☐ No
	Details: _____	
	_____	
v.	Additional investigation ordered	☐ Yes ☐ No
	Details: _____	
	_____	
vi.	Subspecialist referral initiated	☐ Yes ☐ No
	Details: _____	
	_____	
vii.	Prior subspecialist service no longer required	☐ Yes ☐ No
	Details: _____	
	_____	
viii.	Surgical procedure initiated (incl biopsy)	☐ Yes ☐ No
	Details: _____	
	_____	
ix.	Surgical procedure cancelled	☐ Yes ☐ No
	Details: _____	
	_____	
x.	Surgical procedure changed	☐ Yes ☐ No
	Details: _____	
	_____	
xi.	Management redirected towards palliation	☐ Yes ☐ No
	Details: _____	
	_____	
xii.	Decision to palliate reversed	☐ Yes ☐ No
	Details: _____	
	_____	
xiii.	Patient eligibility for a new research study affected ☐ Yes ☐ No	
	Details: _____	
	_____	

4. Additional family members tested (e.g. sibs)	☐ Yes ☐ No If yes, outcome: _____ _____
5. Reproductive risk established for parents?	☐ Yes ☐ No If yes: ☐ <1%　　☐ 25%　　☐ 50%　　☐ Other
6. How do you rate the clinical utility of genomic testing for this patient?	☐ Neutral ☐ Useful ☐ Very useful ☐ Not useful at all ☐ Not very useful
7. Do you think the length of ICU stay was shortened by genomic testing?	☐ Yes ☐ No If so, by how many days? _____ Explain: _____ _____
8. Do you think the length of ICU stay was extended by genomic testing?	☐ Yes ☐ No If so, by how many days? _____ Explain: _____ _____

9. 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: _____
_____
_____