Australian Genomics

Clinical Dataset

History	
a. Pregnancy	☐ Normal ☐ Abnormal Details: ________________
b. Delivery	☐ Normal ☐ Abnormal Details: ________________
c. Birth measurements	length ____________________cm weight ____________________kg head circumference ____________________cm
d. Presenting problem / reason for scan	
☐Abnormal prenatal ultrasound ☐Developmental delay ☐Epilepsy ☐Hemiparesis ☐Autism ☐Intellectual disability ☐Multiple congenital anomalies	☐Microcephaly ☐Macrocephaly ☐Disorder of vision ☐Disorder of hearing ☐Family history of malformation ☐Incidental finding ☐Other ____________________________________
e. Age at presentation	________ months
f. Developmental delay	
☐ Global ☐ Isolated – gross motor ☐ Isolated – fine motor	☐ Isolated – language ☐ Isolated – social
g. Epilepsy	
☐ Yes ☐ No	i. age of onset _____months
ii. seizure type(s) [*Check boxes (can be >1)]* ☐ Neonatal seizures ☐ Epileptic spasms ☐ Absence ☐ Myoclonic ☐ Tonic ☐ Atonic	☐ Hemiclonic ☐ Tonic clonic ☐ Focal – aware ☐ Focal – impaired awareness ☐ Focal – evolving to bilaterally convulsive ☐ Unclassified
iii. drug responsive ☐ Yes ☐ No	iv. best drug __________________________________
v. surgery performed ☐ Yes ☐ No	
vi. surgery type ☐ lesionectomy ☐ lobectomy ☐ TPO resection ☐ Hemispherectomy / otomy ☐ Callosotomy	
h. Vagal nerve stimulator	☐ Yes ☐ No
i. Learning disability	☐ Yes ☐ No

j. Autism	☐ Yes ☐ No
k. Intellectual disability	☐ Yes ☐ No VIQ ____PIQ ____ Full scale IQ____
l. Other	________________________________
Examination	
a. Age at last examination	_______ months/years
b. Length	_______cm
c. Weight	_______kg
d. Head circumference	_______cm
e. Dysmorphic features	☐ Yes ☐ No Details ____________________________
f. Spasticity	☐ Yes ☐ No
i. Spasticity type: ☐ Hemiparetic right ☐ Hemiparetic left ☐ Quadriparetic ☐ Paraplegic	
g. Ataxia	☐ Yes ☐ No Details ____________________________
h. Movement disorder	☐ Yes ☐ No Details ____________________________
i. Vision	☐ Normal ☐ Abnormal Details: _______________
j. Hearing	☐ Normal ☐ Abnormal Details: _______________
k. Neurocutaneous abnormalities	☐ Yes ☐ No Details ____________________________
l. Other physical abnormalities	☐ Yes ☐ No Details ____________________________
Investigations	
a. Brain MRI – (see separate coding sheet)	
b. Cranial US	☐ Normal ☐ Abnormal ☐ Not done Details (date, result, summary) __________________________
c. EEG	☐ Normal ☐ Abnormal ☐ Not done Details (date, result, summary) __________________________
d. Microarray / CGH	☐ Normal ☐ Abnormal ☐ Not done Details (date, result, summary) __________________________
e. Epilepsy surgery pathology	☐ Normal ☐ Abnormal ☐ Not done Details (date, result, summary) __________________________
f. CMV PCR (Guthrie)	☐ Normal ☐ Abnormal ☐ Not done Details (date, result, summary) __________________________

g. Audiology	☐ Normal ☐ Abnormal ☐ Not done Details (date, result, summary) ____________________________
h. Ophthalmology	☐ Normal ☐ Abnormal ☐ Not done Details (date, result, summary) ____________________________
i. CK	☐ Normal ☐ Abnormal ☐ Not done Details (date, result, summary) ____________________________
j. Blood lactate	☐ Normal ☐ Abnormal ☐ Not done Details (date, result, summary) ____________________________
k. CSF lactate	☐ Normal ☐ Abnormal ☐ Not done Details (date, result, summary) ____________________________
l. VLCFAs	☐ Normal ☐ Abnormal ☐ Not done Details (date, result, summary) ____________________________
m. MRI scans on other family members	☐ Normal ☐ Abnormal ☐ Not done Details (date, result, summary) ____________________________
n. Other	☐ Normal ☐ Abnormal ☐ Not done Details (date, result, summary) ____________________________

Imaging Dataset

a. Imaging Data	
i. Age at first scan _________ (months/years) ☐ 1.5T ☐ 3T	ii. Age at last scan_________ (months/years) ☐ 1.5T ☐ 3T
iii. Number of scans: _________	iv. 3T scan ever ☐ Yes ☐ No
v. Age at 3T scan_________ (months/years)	vi. CT scan ☐Yes ☐ No
vii. Best scan quality ☐ Good ☐ Adequate ☐ Poor	
b. Lesions	
i. Number of lesions: _________	ii. Lesion type: ☐ Unifocal – Clear Boundaries ☐ Unifocal – No Clear Boundaries ☐ Multifocal – Unilateral ☐ Multifocal – Bilateral ☐ Hemispheric
c. Localisation	

i. Left Hemisphere	ii. Right Hemisphere
☐ Frontal ☐ Parietal ☐ Temporal ☐ Occipital ☐ Insula	☐ Frontal ☐ Parietal ☐ Temporal ☐ Occipital ☐ Insula
iii. Depth of Lesion (1)	**iv. Depth of Lesion (2)**
☐ Cortical ☐ Subcortical ☐ Deep White Matter ☐ Periventricular	☐ Cortical ☐ Subcortical ☐ Deep White Matter ☐ Periventricular
d. Lesion Characteristics	
i. Gyral Width	☐ Normal ☐ Wide ☐ Narrow
ii. Cortical Thickness	☐ Normal ☐ Thick ☐ Thin ☐ Absent
iii. Grey-White Junction	☐ Normal ☐ Blurred ☐ Stippled
iv. Gyral Pattern	☐ Agyria / Pachygyria ☐ Polymicrogyriami ☐ Cobblestone ☐ Deep / Extended Gyri ☐ Dysgyria Nos ☐ Extended R Sylvian Fissure ☐ Extended L Sylvian Fissure

e. Associated Abnormalities

☐ Corpus Callosum ☐ Normal ☐ Abnormal

- ☐ Complete Agenesis
- ☐ Complete Hypoplasia
- ☐ Partial Agenesis
- ☐ Partial Hypoplasia
- ☐ Dysmorphic
- ☐ Thick

Corpus Callosum further details: ____

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☐ Cerebellum ☐ Normal ☐ Abnormal

☐ Vermis
☐ Left Hemisphere
☐ Right Hemisphere

Cerebellum further details: ____

____

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____

☐ Brainstem ☐ Normal ☐ Abnormal

☐ Hypoplasia
☐ Dysmorphism

Brainstem further details: ____

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☐ Hippocampi ☐ Normal ☐ Abnormal

Hippocampi further details: ____

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☐ White Matter ☐ Normal ☐ Abnormal

White Matter further details: ____

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☐ Lateral Ventricles ☐ Normal ☐ Abnormal

Lateral ventricles further details: ____

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☐ Third Ventricle ☐ Normal ☐ Abnormal

Third Ventricle further details: ____

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☐ Fourth Ventricle ☐ Normal ☐ Abnormal

Fourth Ventricle further details: ____

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☐ Blood Vessels ☐ Normal ☐ Abnormal

Blood Vessels further details: ____

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☐ Basal Ganglia ☐ Normal ☐ Abnormal

Basal Ganglia further details: ____

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☐ Other ☐ Normal ☐ Abnormal

Other further details: ____

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f. Diagnosis

- ☐ Holoprosencephaly
- ☐ Septo-Optic Dysplasia
- ☐ Pontocerebellar Hypoplasia
- ☐ Joubert Syndrome
- ☐ Lissencephaly
- ☐ Band Heterotopia
- ☐ Nodular Heterotopia
- ☐ Cobblestone
- ☐ Polymicrogyria
- ☐ Schizencephaly
- ☐ Cortical Dysplasia
- ☐ Hemimegalencephaly
- ☐ Other

Diagnosis Subtype ____

Pasted MRI Report ____

Likely Genes ____