Genomics Education Programme

100,000 Genomes Project Eligibility wheels A tool for clinicians

Rare Diseases:
Neurology and
neurodevelopmental
disorders Neurodevelopmental
disorders #2

(1) Major criteria

- Holoprosencephaly
- Single central incisor Minor criteria
- Unilateral or bilateral choanal stenosis/ atresia
- · Ptosis
- Small head circumference
- Learning difficulties
- Dysmorphic features
- Hypopituitarism

