Genomics Education Programme

100,000 Genomes Project Eligibility wheels A tool for clinicians

Rare Diseases: Skeletal disorders -Craniosynostosis & Choanal anomalies

(1)

- Dysmorphic features or ≥1major extracranial abnormality
- Significant learning disability
- Family history of craniosynostosis (FDR, SDR, consanguinity)

(2)

- <28 weeks' gestation
- Severe perinatal asphyxia
- Teratogenic exposure, (e.g. sodium valproate)
- Intrauterine growth restriction
- Rickets (genetic or acquired)

