

## Paediatrics

### Neurodevelopmental Disorders

#### Example: Intellectual Disability

- Moderate to Severe/ Profound ID disproportionate to parental IQ unless the family history is consistent with an X- linked disorder
- Congenital onset
- Developmental Delay
- - +/- clinical features suggestive of a specific syndrome

### Respiratory Ciliopathies

#### Example: Non-CF bronchiectasis

FEV1 < 30% predicted and age 4 lobes involved in bronchiectasis < aged 50 OR

- Extensive multilobar disease OR
- 2 < immediate family members affected OR
- High chloride in sweat test but CFTR mutation analysis negative OR
- Bronchiectasis with any suspected underlying immunodeficiency OR
- Bronchiectasis with any suspected underlying ciliopathy OR
- Young's Syndrome OR
- Mounier Kuhn syndrome

### Growth Hormone Disorders

#### Example: IUGR and IGF abnormalities

Height Standard Deviation Score (SDS) < -3 (very significant short stature - well below the 0.4th centile) AND at least one:

- being born small for gestational (birth weight and/or length < -2SDS), ± a history of intra-uterine growth restriction
- Body disproportion
- Dismorphic features
- Aberration in the GH-IGF axis
- Other pituitary hormonal deficiencies
- Family history
- Other features

### Congenital Heart Disease

#### Example: Syndromic Congenital Heart Disease

- Congenital heart disease
- One or more malformations outside the cardiovascular system

### Inherited Epilepsy Syndromes

#### Example: Familial Focal Epilepsies

- Patients with focal epilepsy not thought to be acquired in aetiology
- 2+ first degree affected family members across three generations who have focal epilepsy, not thought to be acquired in aetiology

### Other Examples Include:

- Skeletal Dysplasias
- Cardiomyopathies
- Cardiac Arrhythmias
- Disorders of renal function
- Ichthyosis
- Anaemias and red cell disorders
- Primary Immunodeficiency disorders
- Motor disorders of the CNS
- Specific metabolic abnormalities

There are over 180 eligible rare diseases please see link below for full list

### Rare Disease Eligibility

<https://www.genomicsengland.co.uk/information-for-gmc-staff/rare-disease-documents/>

For any questions please contact the South West Genomic Medicine Centre

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