

Endocrine

Adrenal

Extreme early-onset hypertension

- Age of onset < 30 years AND
- no known cause

Congenital adrenal hypoplasia

- Isolated primary glucocorticoid insufficiency OR
- Isolated primary mineralocorticoid insufficiency OR

Pituitary

Multiple endocrine tumours

- age at tumour diagnosis <60 years AND
- ≥1 endocrine tumour in pt or any family member age <60 years

Idiopathic hypogonadotropic hypogonadism

- Delayed or absent puberty AND
- Consanguinous parents OR affected relative OR other clinical features

Thyroid

Congenital hypothyroidism

- Primary or secondary

Resistance to thyroid hormone

- All patients with negative genetic test

Gonadal and sex development

Early onset familial premature ovarian insufficiency

- Primary or secondary AND
- age <30 AND
- Affected first or second degree family member

Parathyroid

Familial or syndromic hypoparathyroidism

- Hypoparathyroidism AND
- Affected First degree relative OR other clinical features

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 on

Phone: 01392 408565 **Email:** rde-tr.swgmc@nhs.net