

Diabetes

Isolated diabetes - MODY with no genetic diagnosis

- Age at diagnosis <30 years AND
- ≥1 year without insulin AND
- BMI<30

Syndromic diabetes - diabetes with additional phenotypes

- Developmental delay /renal/cardiac or other clinical features AND
- Age at diagnosis < 25 years AND
- ≥3 year without insulin AND

Diabetes with other autoimmune disease

- DM + ≥2 other autoimmune condition i.e. Graves', Coeliac AND
- any two with age of onset <10 years

Diabetes with Insulin resistance (including lipodystrophy)

- age of diagnosis < 25 years AND
- ≥3 year without insulin AND
- BMI<30 AND
- Acanthosis nigricans AND
- Biochemical Insulin resistance

Syndromic or non-syndromic severe obesity

- BMI > 3SD above the mean AND
- Age of onset <5 years

Other:

Hyperinsulinism

Neonatal diabetes

46,XY disorder of sex development

46,XX disorder of sex development

IUGR and IGF abnormalities

Parathyroid cancer

Lipid

Familial Hypercholesterolaemia

Simon Broome criteria

- TC >7.5 mmol/l or LDL > 4.9 mmol/l AND
- Tendon xanthomas in patient or family

Severe hypertriglyceridaemia

- Triglycerides >20mmol/L AND
- Asymptomatic, OR Acute pancreatitis event, OR Eruptive

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

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