Genetic Testing for Disorders of Sex Development (DSD) – Health Professionals

Genetic testing for Disorders of Sex Development (DSD) is available through the molecular genetics laboratory in the Laboratory Medicine Building at the Queen Elizabeth University Hospital Glasgow.

The laboratory offers targeted panel testing using next generation testing for more than 50 genes involved in XY DSD, XX DSD and hypogonadotrophic hypogonadism (HH). The 21 gene HH panel can be analysed independently if the diagnosis is suspected.

In addition, the laboratory offers array CGH for the detection of chromosome microdeletions and microduplications which can be implicated in DSD.

Clinicians requesting testing are asked to complete a proforma (either DSD or HH proforma) including the clinical details and the results of relevant investigations prior to testing. This will facilitate accurate interpretation of any genetic variants identified in the analysis.

Please send the relevant completed proforma with a standard genetic testing form (marked for DSD or hypogonadotrophic hypogonadism panel) and 5 – 10 ml EDTA sample to the molecular genetics laboratory (genetic.laboratories@ggc.scot.nhs.uk).

The DSD proforma, HH proforma and molecular testing form are available on the SDSD website

Referrals are accepted from within and out-with Scotland.

The proforma is discussed at the monthly DSD Diagnostic Board which meets between 1100 and 1230 on every second Monday of the month. The results of any genetic testing are also discussed at this meeting which has multidisciplinary input from members of the Glasgow DSD team listed below. Variants of uncertain significance will be discussed before being returned to the referring clinician. Teleconference facility is available upon request. If clinicians wish to be involved in discussion about their cases please contact Martin McMillan for details (martin.mcmillan@nhs.net).

Professor S Faisal Ahmed (paediatric endocrinology)
Dr Ruth McGowan (clinical genetics)
Dr Gabriella Gazdagh (clinical genetics)
Dr Angela Lucas-Herald (paediatric endocrinology)
Martin McMillan (child health)

Dr Jane McNeilly (clinical biochemistry)
Dr Louise Young (molecular genetics)
Professor Edward Tobias (clinical genetics)