



Genetic Testing for Disorders of Sex Development (DSD) – Families

Disorders of sex development (DSD) are a group of conditions that in boys range from hypospadias to atypical genitalia. As well as finding the cause of the condition, genetic testing may provide information about risk of recurrence in a future pregnancy, about prognosis and potentially influence management of the condition.

Genetic testing aims to identify alteration(s) in a gene(s) and in Scotland we are at present able to test more than 50 genes involved in DSD.

The request for genetic testing and the results are discussed at a multidisciplinary meeting with input from paediatric endocrinology, clinical biochemistry, clinical and molecular genetics and given to the referring doctor. In some instances, the laboratory may identify a gene alteration of uncertain clinical significance. Discussion at the diagnostic meeting and possibly further testing within the family will aim to clarify this.