

Scottish Differences of Sex Development **(SDSD)**
National Managed Clinical Network

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

Information sheet

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.

Further Information

This document has been prepared by Public Services Delivery Scotland (PSD Scotland) on behalf of the Scottish Differences of Sex Development (SDSD) network. Accountable to Scottish Government, PSD Scotland works at the heart of the health service providing national strategic services to the rest of NHS Scotland and other public sector organisations to help them deliver their services more efficiently and effectively. The SDSD network membership is made up of a multidisciplinary group of healthcare professionals who provide care to children and adults born with a difference in sex development or anomaly of the genitalia. The network is supported by a PSD Scotland Programme Team to drive improvement.

This leaflet will be available in other languages (for example, Punjabi, Urdu, Polish) or formats (for example, larger print) on request.

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