

Scottish Differences of Sex Development

SDSD

National Managed Clinical Network

Genetic testing

Information leaflet

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

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the genitalia. The network is supported by a PSD Scotland Programme Team to drive improvement.

What are DSDs?

Differences in sex development (DSDs) are conditions that may be caused by changes in the genes or hormones (or both) involved in the development of reproductive organs.

There are many types of DSD. They may be seen at birth if a child is born with atypical genitalia (differences in how the genitals look) or has hypospadias (when the opening of the tube that carries urine is not at the tip of the penis). These conditions may only be seen in adolescence if puberty is delayed or as an adult if there are concerns about fertility.

Why do DSDs happen?

A DSD can be caused by a change in the genes or the hormones that control the development of your genitals and your body during puberty. It can run in families, but there is often no clear reason why it happens. If doctors suspect you have a DSD, you will usually have some tests which may include genetic and hormone tests to try to find the cause. Sometimes we do not find a cause.

What are genes and chromosomes?

Our genes can be thought of as a library of instructions, unique to everyone, that are important for the development and function of our bodies. Multiple genes are grouped together to form larger structures called chromosomes found in each of the building blocks (cells) in our body.

In our cells there are usually 23 pairs of chromosomes and one of each pair comes from either parent. One of these 23 pairs is often different in males and females and is called the sex chromosome. In most, but not all, males this pair contains an X and a Y chromosome (XY). In most, but not all, females this pair has two X chromosomes (XX). In some people, the sex chromosome number and type can be different in other ways, such as 45,X, 47,XXY, or combinations such as 45,X/46,XY or 46,XX/46,XY. As these sex chromosomes play an important role in sex development, any differences in the number or type of chromosomes can lead to some concern about development of the reproductive organs.

What is genetic testing?

Genetic testing in DSD usually includes chromosome testing (called karyotyping or microarray) which looks at the number and type of your chromosomes.

Your medical team may suggest further tests. These include:

- gene panel testing looking at a specific set of genes known to be involved in DSD
- exome or genome sequencing looking at a larger number of genes at the same time

How do you do genetic testing?

We will often take a small sample of blood for genetic testing when you, or your child, are having other blood tests, for example to check your hormone levels. Occasionally, a sample of your saliva may be used for genetic testing. Sometimes it is useful to collect samples

from parents so that we can compare them. This is especially if we are doing more detailed genetic testing looking at a large number of genes, such as exome or genome sequencing.

Why do you do genetic testing?

A DSD may result from a change in a chromosome or gene involved in sex development. Test results will provide information for you and your family as they:

- will help guide your medical team on how to manage and treat your condition
- may provide information about the chance of the condition happening in another member of your family or in a future pregnancy.

You may be referred to clinical genetics to discuss genetic testing and what this would mean for you, your child, your family and any future pregnancy.

What are the possible outcomes of testing?

Results from genetic testing may show:

- a clear genetic cause for the DSD
- no clinically significant changes in the genes known to cause DSD
- a change that we are not sure is clinically significant. This means that we are not sure whether this is the cause of the DSD. Further investigations, longer-term monitoring or family testing may help to make the result clearer.

When and how will I get the results?

Genetic testing is complex, so results can take some time. Chromosome results may take one to two weeks, but gene panel results often take three to four months. You can discuss with your medical team how they will give you the results. You may be referred to clinical genetics so they can discuss the results with you. They can also answer any questions you may have and explain what the results mean for you and your family.

Will the genetic result have implications for my family?

A genetic result may have implications for other family members. In this case, you and your family may be referred to clinical genetics for genetic counselling.

Where can I get support?

If you have a DSD or you are a parent of a child with a DSD, the organisations listed below can give you more information and advice about living with a DSD. You can also read more about DSD and genetic testing on their websites.

Dsdfamilies: dsdfamilies.org

NHS Differences in sex development: nhs.uk/conditions/differences-in-sex-development/

NHS Genetic and genomic testing: nhs.uk/conditions/genetic-and-genomic-testing/

Genetic Alliance UK: geneticalliance.org.uk/support-and-information/genetic-services-and-testing/

Congenital Adrenal Hyperplasia Support Group: livingwithcah.com

Further Information

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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