

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.

- Scottish Differences of Sex Development Network – www.sdsd.scot.nhs.uk/
- dsdfamilies – www.dsdfamilies.org
- Differences in sex development NHS – www.nhs.uk/conditions/differences-in-sex-development/
- Genetic and genomic testing NHS – www.nhs.uk/conditions/genetic-and-genomic-testing/
- Genetic Alliance UK geneticalliance.org.uk/information/service-and-testing/benefits-and-risks-of-genetic-testing/
- Congenital Adrenal Hyperplasia Support Group – www.livingwithcah.com
- Mayer Rokitansky Kuster Hauser UK www.mrkh.org.uk

For more information, contact:

SDSD Managed Clinical Network
National Specialist and Screening Services
Directorate (NSD)
Procurement, Commissioning and Facilities
NHS National Services Scotland
Gyle Sq, 1 South Gyle Crescent
Edinburgh
H12 9EB

Phone: 0131 275 7378

Website: [//www.mcns.scot.nhs.uk/sdsd/](http://www.mcns.scot.nhs.uk/sdsd/)

This leaflet is available in other languages (for example, Punjabi, Urdu and Polish) or formats (for example, larger print). Please contact our office at the address above or visit our website www.sdsd.nhsscotland.com.

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Scottish Differences of Sex Development Network

Information about genetic testing



What are DSDs?

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

There are many types of DSD. They can be recognised at birth when a child is born with atypical genitalia (differences in how the genitals look) or hypospadias (when the opening of the tube that carries urine is not at the tip of the penis). However, these conditions may not show until adolescence (if puberty is delayed) or adulthood (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 be inherited, but there is often no clear reason why it happens. If doctors suspect you have a DSD, you will usually have some tests (which will often include genetic and hormone tests) to try to find the cause. However, 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 each individual, that are important for the development and function of our bodies. Multiple genes are grouped together to form larger structures called chromosomes, which are in each of our 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 mention further tests. These include:

- gene panel testing, which looks at a specific set of genes known to be involved in DSD; and
- exome or genome sequencing, which looks at a larger number of genes all 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 enough for genetic testing. Sometimes it is useful to collect samples from parents so that we can compare them, especially if we are doing more detailed genetic testing (looking at a large number of genetic codes), 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 – results will help guide your medical team on how to manage and treat your condition; and
- your parents and family – results may provide information about the chance of the condition occurring 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; or
- 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 other 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, 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 may be referred to clinical genetics for genetic counselling. Members of your family will also have this opportunity.