SWAN UK
syndromes without a name

HOW CAN I ACCESS GENETIC TESTING FOR MY CHILD?

To access genetic testing you must contact your GP or specialist clinician who will arrange a referral for you to an NHS Regional Genetics Service. Here we explain what will happen at that appointment, and how long you might have to wait for a result.

GENETIC ALLIANCE UK

Accessing genetic testing

Genetic testing on the NHS is available through your clinician who orders the tests that are appropriate for you from an NHS-certified laboratory. The clinician will request a test only if they know that the results will help them provide you with the most appropriate healthcare. NHS policies define who is most likely to benefit from specific genetic tests.

Your clinician will collect your sample for genetic analysis and send it to the laboratory. The laboratory will then analyse and interpret the results. Your clinician will then be available to talk you through what your results mean. Anyone having a genetic test on the NHS is also likely to see a genetic counsellor. If the test will be looking to determine whether you are affected by a serious genetic condition, counselling will be available both before and after you take the test.

What happens at a genetic testing appointment?

After being referred to an NHS Regional Genetics Service you will usually be sent a leaflet or letter explaining what will happen when you attend the clinic for your appointment. This letter often asks you to bring your child with you to the appointment. Before attending appointments, it is a good idea to write down any questions that you want to ask when you are there. You might also find it helpful to take a notebook along to the appointment. Some families record the appointment on their phone so they can listen again afterwards. You should always ask permission from the other people at the appointment before recording it.

If you are worried you won't remember some of the details at the appointment you can ask your geneticist to write them down for you to look at again when you get home. Usually they will write to you after the appointment summarising what was discussed so you have a written record of it.

If you are a single parent or your partner cannot attend the appointment you might want to take along another family member or trusted friend for support. They may also remember points about the appointment that you miss.

Often at the start of the first appointment the geneticist will ask parents what they are hoping to find out, for example:

- A diagnosis for their child
- How their child will be affected in the future
- If any future children could be affected
- Whether other children in the wider family could be affected

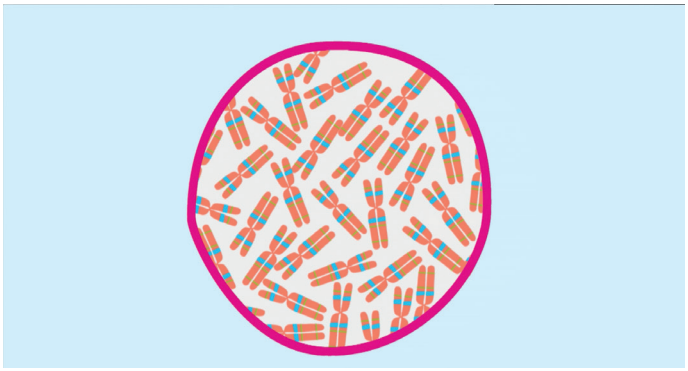
Most of the appointment will then be going through the 'genetic assessment process'. This is a diagnostic process where clinical geneticists work with laboratory staff and other medical professionals to explore the possible reasons for the different developmental patterns a child has.

Their aim is to identify a genetic syndrome or condition that explains the child's difficulties. The cause of the condition might be a single-gene disorder, or it might be that a particular chromosome has a piece missing (deletion) or an extra piece (duplication) or it may be that pieces of chromosomes have 'swapped around'

(translocation).

Identifying the genetic cause of a child's health problems occurs through a combination of investigations:

- Asking about the child's development in detail.
- Asking about the child's family's medical history.
- Physical examination including photographs.
- Laboratory investigations (if necessary blood samples are taken during the appointment or shortly afterwards).



Asking about the child's development in detail

The history of a child's development and the pattern of problems that they currently have or have had in the past is a key component of the assessment. It can be frustrating having to repeat your child's history to yet another professional, but this is a very important part of the assessment. This can sometimes feel upsetting as you have to focus on all the things that make your child different or that they can't do.

It is a good idea to have people you can talk to after your appointment. This might be a friend or family member or you might want to share your feelings with other SWAN UK families who understand how difficult these appointments can be. You can join SWAN UK by completing our online registration from, which you can access at: undiagnosed.org.uk/joinus.

Asking about the child's family's medical history

The geneticist will also ask about your family history to see if they can identify patterns that might give clues to the cause of your child's problem. Family history is also important because if the problem is genetic then it may be the result of changes in genes or chromosomes which were passed down from one or both of the parents.

Sometimes it may be possible to make a specific diagnosis based on these historical descriptions alone without the need for genetic testing. Sometimes families might find it difficult to describe their family histories because they may be adopted or have little contact with their family.

Physical examination including photographs

The aim of this is to identify any particular physical features that might give the geneticist a clue about a possible diagnosis. This can be an odd, and sometimes upsetting, process for both the child and parents.

'We sat in a room with a genetics team as they poked and prodded and listed all of my child's features that stood out to them. Things like unusual facial features, sparse hair and horizontal

creases on the hands along with many others. I came out of the meeting wondering what other people must see when they look at my child.'
SWAN UK member

Whilst procedure may seem odd, or even upsetting, it is important to remember that physical examinations are a vital part of genetic testing and can help identify possible genetic conditions.

Laboratory investigations (if necessary blood samples are taken during the appointment or shortly afterwards)

This is usually the final part of the diagnostic process. It takes place after careful examination of the child's history and the physical examination.

Samples of the child's DNA (and sometimes the parents') will be taken to test and look for changes in the DNA. The samples needed can vary and may include blood, saliva or skin. Once a geneticist has an idea about what might be a potential diagnosis for the child, they may order laboratory tests straight away to confirm the suspected diagnosis. There can often be a long wait for results.

It is helpful to have people you can talk to after your genetic testing appointment – a friend, family member or other SWAN UK families.

Will my child have to wait ages for a genetic test?

Sometimes it is not possible to undertake genetic testing straight away. This might be because your child is too young for their physical features to give any clear 'clue' about what the underlying condition may be.

In these situations, you will usually be invited back for follow-up appointments several years later when there may be more clues because your child has developed, because new medical knowledge has come to light, or because a new test has become available. This period is usually referred to by geneticists as 'watchful waiting'.

It is common for families to be surprised by the 'watchful waiting' period; most people don't realise how long genetic testing can take. It can be frustrating and worrying waiting for answers and many families wonder if they have been forgotten.

Whilst they are waiting many families start searching the internet to see if they can find answers about what is wrong with their child. Although this is understandable, internet searches for symptoms often produce photographs of children with a range of different conditions which can vary in the way they affect different children.

It is often the 'worst-case scenario' images that pop up and some of these images can

be very upsetting for families to see.

Genetic testing can take a long time and you may have to wait several years for any result

How is private genetic testing different to the type of genetic testing available on the NHS?

Private genetic testing involves the same genetic testing process we have already described, but you will have to pay for it instead of the NHS. It can often be delivered by clinicians who work both in the NHS and for a private practice, but not every clinician works for a private practice – there is a smaller pool of clinical expertise in private practice.

Many of the elements of the process are slow, and this is mostly not because of NHS resource problems, or waiting times, but because the process of making a genetic diagnosis takes time.

You will probably get to see the clinician faster with private genetic testing, but you might not get as good an outcome.

In NHS Regional Genetics Services, children with particular types of problems might be referred to experts in that kind of health problem, or your clinician might consult with them. If that expert does not provide private services, then you might lose out on that specific expertise by going privately.

There is also an issue with continuity. When it is working well, the NHS should function as a single complete system, where one clinician can refer your child to the most appropriate clinician to treat them. Before entering into private genetic testing, you should make sure you understand what the next steps for your child will be, and whether the NHS will continue their treatment.

What is direct-to-consumer genetic testing?

Direct-to-consumer genetic tests are genetic tests that are marketed directly to you as a consumer. You can order these online or by post, complete at home and then send away for analysis.

Direct-to-consumer genetic tests can provide you with some information about your genetics but they are not tailored to you. These tests are not designed to diagnose a medical condition and should not be used as a substitute for visiting your doctor if you are at all concerned about your current health or the risk that you may develop a condition in the future.

It is also important to note that they are not designed for use on children. If you have any concerns about your health, or the health of a loved one, and think that you or your loved one could benefit from having a genetic test, the best thing to do in the first instance is to speak to your doctor.

My child was seen by a geneticist years ago, should I ask to be seen again?

There have been significant advances in genetic testing over the last few years. Genomic microarray (array-CGH) is an advanced technique in genetic testing that

detects copy number changes in a person's chromosomes.

This means it looks for where there are deletions (bits missing) or duplications (extra bits) in your DNA that would not be identified using conventional microscopy-based chromosome analysis (karyotype). Such tests are now more widely available in the NHS.

Many children and young adults with developmental delay who had a 'normal' result from a microscopy-based chromosome analysis (karyotype) in the past have, after being re-tested using genomic microarray analysis, been found to have a microdeletion or microduplication that provided a diagnosis for the child's condition.

In the NHS at present, clinical genetics services are commissioned as specialised services meaning a referral has no cost implications for the referring clinician. Exome and genome sequencing are new genetic testing methods which test all the genes in the body. At present sequencing is not available in NHS Regional Genetics Services. It is only available by taking part in research studies.

Useful links

bit.ly/rrjoinswanuk

bit.ly/cafamilypatternsofinheritance

bit.ly/rrswanuksupportandinformation

bit.ly/yourgenomehome

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Notes

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SWAN UK is a support network run by the charity Genetic Alliance UK, offering support and information to families of children with undiagnosed genetic conditions.

GENETIC ALLIANCE UK

Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by rare, genetic and undiagnosed conditions. We are an alliance of over 200 patient organisations.

RARE DISEASE UK

Rare Disease UK is a multi-stakeholder campaign run by Genetic Alliance UK working with the rare disease community and the UK's health departments to effectively implement the UK Strategy for Rare Diseases.

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