SWAN UK
syndromes without a name

WHAT DOES GETTING A GENETIC DIAGNOSIS MEAN?

For most families, getting a diagnosis remains just as important to them as their child grows up. Without one families can struggle to access the right support. They have no idea what the future holds for their child or if other children they may have in the future could be affected.

GENETIC ALLIANCE UK

Getting a diagnosis

Although parents generally know that having a diagnosis will not significantly change their child's life, they hope it will give them a greater sense of what to expect.

'Wanting a diagnosis is not about expecting a cure or a magic wand … but knowledge is power.' (SWAN UK member)

Families of children who have been given a genetic diagnosis of a very rare chromosome condition, where the child may be one of only a few in the world known with that condition, can sometimes feel disappointed with the information given to them. Families generally hope that a diagnosis will give them an indication as to what the future holds for their child but this is not always the case.

'I wasn't expecting a cure, I knew a genetic condition was lifelong, but I was expecting an understanding of what it meant for my son's life. What I got was a string of letters and numbers.'(SWAN UK member)

Although families are usually glad they can now explain what is wrong with their child, the technical description of the genetic change in their child's genes or chromosomes does not give them much idea of what to expect from the future. If their child is the only one, or one of a few in the world with this known genetic change, they can still feel quite isolated.

Many members of SWAN UK who have obtained a diagnosis have remained part of our community as their child's diagnosis is so rare there isn't any obvious other support group for them to join.

However, while these rare diagnoses do

not always tell the family much about what they can expect in their child's future, it does at least give them a reason for their child's problems – even if it is unlikely to be understood by many people outside of the field of genetics. Many families say that without a diagnosis they feel like they are treated like a 'neurotic parent' or told that their child will 'catch up' or 'grow out of it'. Having a diagnosis gives them a way to respond to this kind of comment.

The support group Unique offers information and support to families with rare chromosome disorders and, where possible, will link together families whose children have the same rare condition.

How important is having a genetic diagnosis?

Having a diagnosis is really important to families on a practical and emotional level. Here are some of the main reasons for needing a diagnosis:

To have a sense of what the future holds

'No diagnosis means no prognosis.' (SWAN UK member)

Without a diagnosis families have no idea what the future holds for their child – will they walk? Will they talk? Will they have a shorter life expectancy?

'I hate not knowing if my childs' problems will be life-limiting or just how bad the regression will be.' (SWAN UK member)

Having a diagnosis could help identify potential treatments or health issues that need to be monitored in future. Without a diagnosis it is hard to know whether new symptoms are something to be worried about.

'Every time she is feeling unwell, has a sore head or has a pain somewhere … I wonder, is there something more serious going on inside her than just the "norm". Fear caused by not knowing what to expect in her future is the hardest thing.' (SWAN UK member)

To have the child's or young adult's needs taken seriously

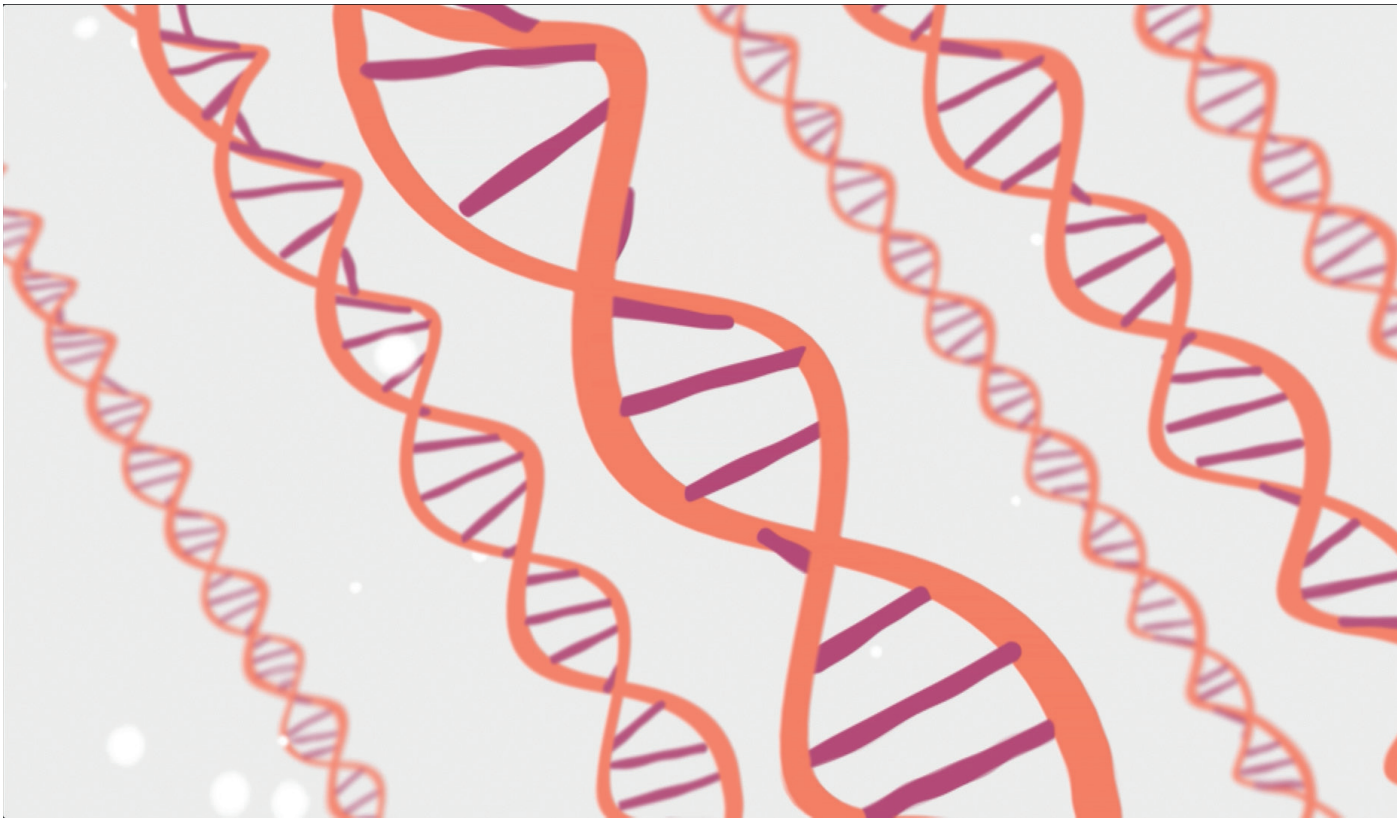
Many families, even those whose child has significant disabilities, can find it hard to get people to take their needs seriously. Sometimes people assume that because the condition is undiagnosed it isn't as serious as a named condition or that it will improve in the future.

'I want an answer to stop the "maybe he will grow out of it" comments.' (SWAN UK member)

Some families are met with disbelief when they say their child is undiagnosed, and it is very difficult to explain the impact it has.

'Not having a "set" of expected impacts from a diagnosis means constantly having to explain what the issues are. It can be exhausting.' (SWAN UK member)

To know if other children in the family will be affected



‘I want another child but without any answers it seems like such a gamble.’ (SWAN UK member)

Without a diagnosis it is impossible to know if the condition is inherited or just a one off. This means families don’t know if future children will be affected by the same unknown condition. This affects not only the parents of the undiagnosed child, but also their siblings, aunts, uncles and cousins.

To access service and support

‘I had to almost beg for [a] hospice place – no diagnosis so no prognosis, and this is despite the amount of times he’s been resuscitated.’ (SWAN UK member)

Families often find it hard to access services

without a diagnosis. Forms or assessment criteria often have a box that asks for the child’s diagnosis and families struggle to know what to write.

‘When you have the “diagnosis” box on the endless paperwork you have, you end up listing all issues but there’s no room for them all! With a diagnosis you can just pop that down and it’s done!’ (SWAN UK member)

‘Accessing any kind of support or professionals is always a fight as she doesn’t fit into the criteria.’ (SWAN UK member)

In a recent SWAN UK survey 53% of respondents said they have struggled to access social care support because their child is undiagnosed.

The recent Rare Disease UK report found that:

- 7 out of 8 respondents affected by undiagnosed genetic conditions did not feel they had been provided with enough information and support throughout the diagnosis process.
- 73% of respondents without a diagnosis felt it had been a barrier to accessing

treatment.
- 4 out of 5 respondents felt that being undiagnosed had been a barrier to receiving appropriate coordinated care.

Why does genetic testing take so long?

If the laboratory knows exactly what genetic change it is looking for, because somebody else in the family has the same condition, or because the laboratory knows which area of the gene to look at, it has a much easier task. The test may then only take a week or two.

However, if no genetic change has previously been found in the family, or, if there are a number of genes associated with the condition, it will need more work to get a result.

Instead of focusing on one area of a gene the laboratory may need to analyse the whole gene or more than one gene. This can be a very long process and can take many months. Exactly how long will depend on a number of factors such as how big the gene is and the facilities available at the laboratory.

For example, the condition Duchenne muscular dystrophy is caused by changes in a gene called dystrophin, one of the longest genes known. There are thousands of different possible genetic changes that can occur, and therefore finding a family's particular change can be a very long and laborious process. On the other hand, in the case of Huntington's disease, changes in the crucial gene always occur in the same small region. Therefore scientists know exactly where to look in the gene and so the test is fairly easy and much quicker.

The quality of the DNA is also an important factor. Sometimes laboratories first have to check the DNA of someone who is deceased in order to identify the particular genetic change. If the DNA from the deceased person is poor quality, this can double or triple the time it takes to find the mutation. In some cases it may not be possible to complete the analysis because there is not enough DNA.

What if my child doesn't get a diagnosis?

A significant number of children having genetic testing will not receive a diagnosis, or a variant of unknown significance might be found. This can be very difficult for families and it is important you have support. By joining the SWAN UK community you will be able to talk to other families in the same situation who understand how challenging it can be.

Useful links

undiagnosed.org.uk

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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