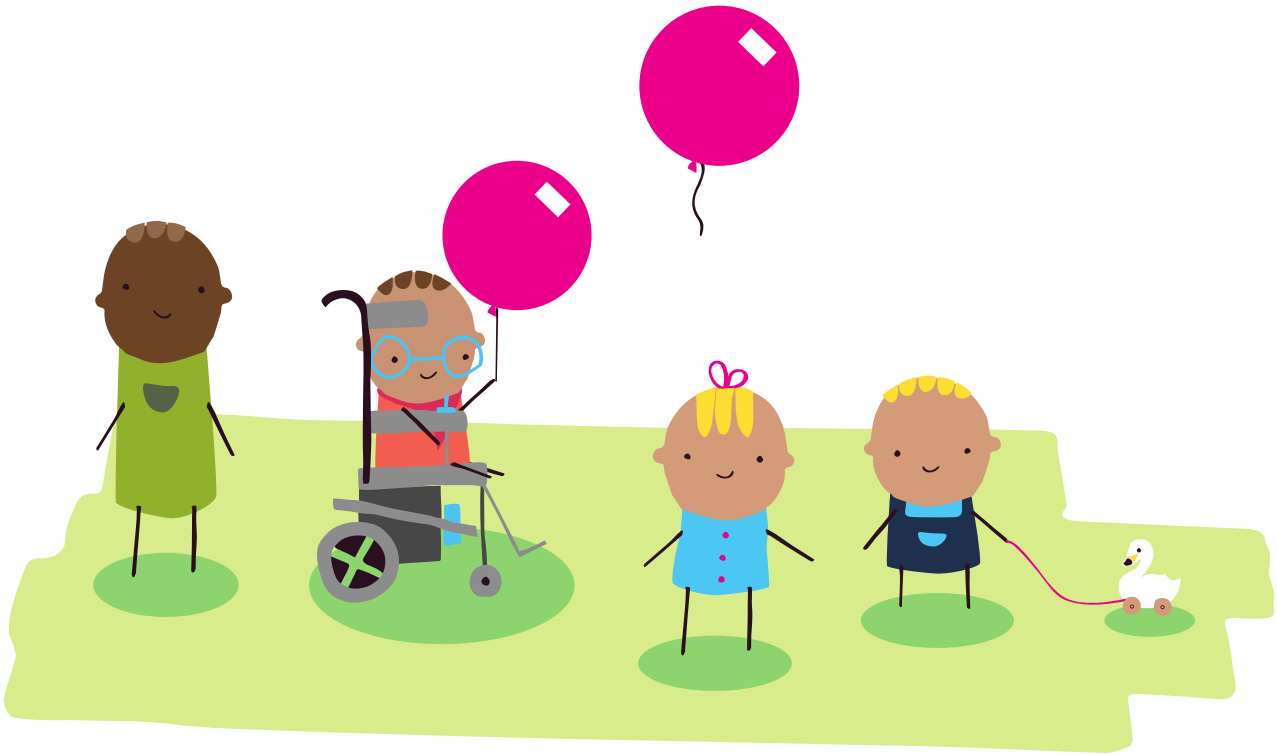
SWAN UK
syndromes without a name

WHAT DOES SWAN OR BEING UNDIAGNOSED MEAN?

SWAN stands for ‘syndromes without a name’. It is not a diagnosis, but a term used when a child or young adult is believed to have a genetic condition and testing has failed to identify its genetic cause. Syndromes without a name are also referred to as undiagnosed genetic conditions, unknown genetic conditions or undiagnosed genetic disorders.



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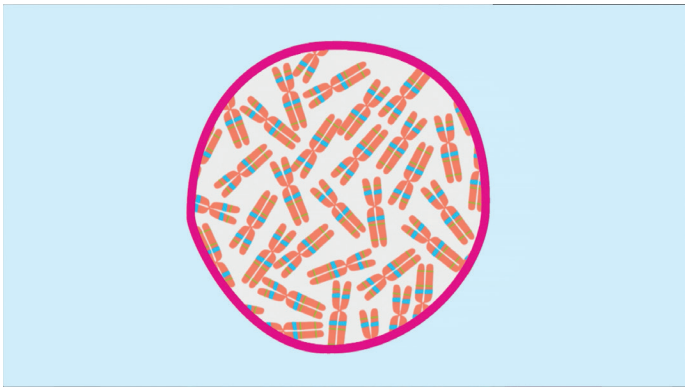
Some children affected by a syndrome without a name might be described as having global developmental delay or failure to thrive. They might have learning disabilities and/or physical disabilities. They can sometimes have complex medical needs and may have epilepsy. Some children may not have any learning disabilities but be physically disabled whilst others are physically unaffected but have learning disabilities.

Children affected by a syndrome without a name can have a range of different symptoms and each child is likely to be affected differently.

Some of the other symptoms children might be identified as having include:

- hypertonia or hypotonia (problems with muscle tone)
- hypermobility or hypomobility (problems with joints)
- feeding difficulties, such as problems with swallowing – many children may be tube fed
- unusual features
- respiratory problems (difficulty breathing)



Why are some children and young adults undiagnosed?

It can be really hard for families when they have no answer to the question 'what is wrong with my child?' or 'why is my child always ill?'

There are three main reasons why a child's genetic condition remains undiagnosed:

- It is the 'rarest of the rare'– a condition that has never been seen before and therefore isn't tested for.
- It is an unusual presentation of a known condition – the child's symptoms might be different to those of other children with the same condition. The condition is therefore not tested for because it doesn't appear to be that condition.
- Sometimes when genetic testing is undertaken genetic changes are found which are of 'unknown clinical significance', for example, changes are found in the child's chromosomes but these are not thought to be the cause of their difficulties. Sometimes this is because the change has been inherited from one of their parents who is unaffected by the condition, or because, although the genetic material has moved around, it all appears to be there.

'My son's microarray (a type of genetic test) showed changes but nothing they could see was responsible for his global developmental delay. Because there are so many changes in his chromosomes it seems likely that one of them is causing his difficulties but if there are any deletions they are too

small to be identified. That basically leaves us in the position where we don't have a diagnosis but we do know/suspect it is genetic.' (SWAN UK member)

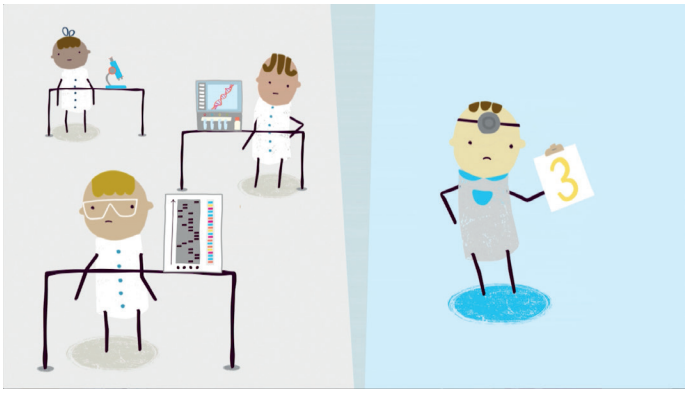
How long can a child or young adult be undiagnosed for?

Most people are surprised when they find out how long children or young adults can remain undiagnosed for – many families wait years for a diagnosis and some may never get one. A Rare Disease UK report published in 2016 found that nearly half of the respondents who identified as undiagnosed had been waiting over five years.

72% of SWAN UK members' children are taking part in genetic research studies which are likely to take years to provide results.

How common is it to have a syndrome without a name?

Approximately 6,000 children are born in the UK each year with a genetic condition likely to remain undiagnosed. Currently around 50% of children undergoing genetic testing in the UK won't get a confirmed diagnosis.



It is thought that about half (50%) of children with learning disabilities and approximately 60% of children with congenital disabilities (disabilities which are apparent from birth) do not have a definitive diagnosis to explain the cause of their difficulties.

Useful links

bit.ly/rarerealityreport

bit.ly/rrswanuksupportandinformation

bit.ly/yourgenomehome

bit.ly/rrjoinswanuk

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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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