SWAN UK
syndromes without a name

UNDIAGNOSED GENETIC CONDITIONS

Information for teachers and school leaders

This leaflet provides you with information on undiagnosed genetic conditions and explains how having an undiagnosed genetic condition can impact a pupil and their family's experiences of school.

GENETIC ALLIANCE UK

Introducing SWAN UK

SWAN UK (syndromes without a name) is the only dedicated support in the UK for families of children and young adults (0-25) affected by undiagnosed genetic conditions. We are run by the charity Genetic Alliance UK.

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

There are three main reasons a genetic condition can remain 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 clinically to be that condition.
- Sometimes when genetic testing is undertaken genetic changes are found which are of unknown clinical significance, for example, a change (or changes) are found in the child's genes but it is not possible to say definitively that this is the cause of the child's condition. These changes are called variants of unknown significance.

While some may go on to eventually receive a diagnosis, many will remain undiagnosed. SWAN UK seeks to support these families in a variety of ways. We produce leaflets to help families understand more about genetics and genetic testing, run information events, organise whole-family days out, and facilitate peer support for parents through our online forums. The following leaflet lets you know more about undiagnosed genetic conditions, life with an undiagnosed child and how we can help you to provide support to a child with an undiagnosed genetic condition in your classroom.

For more information about the support we offer visit: undiagnosed.org.uk.

It is estimated that approximately 6,000 children are born every year with a syndrome without a name - a genetic condition so rare it is unlikely to be diagnosed.

Supporting pupils with an undiagnosed genetic condition

A rare or undiagnosed genetic condition can affect a child's ability to learn, the way they learn and communicate, and the coping mechanisms and strategies they will use to meet the demands of the curriculum and school life.

For some conditions, for example autism spectrum disorder, multi-sensory impairment and Downs syndrome, there are tried and tested methods of supporting learning and communicating in the classroom. For children who are undiagnosed however there can often be more trial and error involved in working out how they can best be supported to access an appropriate curriculum which fully meets their needs. With undiagnosed

conditions presenting so many unknowns, it is important that families and professionals work together in order to achieve the best possible outcome for each child.

As part of the SWAN UK big ambition that every family affected by an undiagnosed genetic condition has the support they need, when they need it, regardless of whether they have a diagnosis or not, we hope to work with schools and teacher training providers to increase understanding of undiagnosed genetic conditions.

We will be producing a more detailed guide on how to support children affected by undiagnosed genetic conditions in the classroom and hope to create opportunities to work in partnership with schools and develop new approaches to teaching children affected by undiagnosed genetic conditions and effectively supporting their families.

We would also love to come and visit you and your team to talk more about the issues affecting undiagnosed families and the support we can offer.

To find out more about our plans and how we can support your school contact info@undiagnosed.org.uk or visit undiagnosed.org.uk.

The value of diagnosis

For some children affected by medical, physical and learning disabilities, a diagnosis can be made relatively quickly and easily through genetic testing. For others it may come after a long wait, and sometimes, after multiple misdiagnoses.

Although receiving a diagnosis can be a difficult time for families, it can also have its advantages. Having a diagnosis means that families and professionals may have an understanding of the cause, likely symptoms and prognosis of a condition. It can allow professionals to meet and anticipate an individual's needs, and in some cases dictate treatments. A diagnosis can also allow families to connect with others going through similar experiences and, if they wish, join relevant support networks.

One of the most important things that professionals working in the education sector can do is recognise the value of diagnosis.

Often families are told in a very well meaning way that 'we don't label children' and that 'we treat children based on symptoms not diagnosis', however sometimes in the absence of a diagnosis a child's symptoms are not fully understood. A diagnosis can open doors. It can enable professionals to understand the underlying cause of a symptom and predict how said symptom is likely to progress. When there is no diagnosis to act as a guide, then strategies and interventions often require more thought and creativity.

For more information about getting a genetic diagnosis visit: bit.ly/gettingadiagnosis.

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Living without a diagnosis

Some conditions are so rare that doctors struggle to identify them through standard genetic testing.

These conditions are generally described as a 'syndrome without a name' (or SWAN for short). It is thought that around half of children with learning disabilities and approximately 60% of children with congenital abnormalities (differences which are apparent from birth) do not have a definitive diagnosis to explain the cause of their difficulties.

For children and families who do not have a diagnosis, life can be full of unknowns. For example, no one knows why the condition has occurred or what its prognosis is. This lack of prognosis means that parents often have questions which can't be answered, such as:

- Why do particular symptoms occur?
- Will the condition get worse?
- Are there likely to be further complications as the child gets older?
- Is the condition life limiting?
- Will other children in the family be affected?

Find out more about living without a diagnosis by reading the family stories on our website: bit.ly/SWANUKfamilystory.

How does being undiagnosed affect children and their families?

Often children with undiagnosed genetic conditions are under the care of multiple consultants and medical departments – sometimes these departments can be based in different hospitals.

The number of appointments, therapy sessions and clinics that children with an undiagnosed condition may have to attend can have a significant impact on family life and also affect school attendance. Some families have reported that they struggle to access certain services, such as therapies and respite care, because their child's condition is not recognised or fully understood. Additionally, managing their child's condition can put parents under huge emotional and financial strain.

Parents of children with a diagnosed condition are often able to access support networks where they can seek advice and encouragement from other parents who can empathise with their situation. However for parents of children without a diagnosis, appropriate support networks can be hard to find as they don't know where they 'fit in'.

Siblings can also struggle with the impact of having a brother or sister affected by an undiagnosed genetic condition. They have no answers to explain or help them understand the difficulties their sibling has. If their brother or sister has complex health needs they may also have experience of frequent emergency hospital trips or ambulances being called, all of which can then impact on their emotional state and ability to concentrate in school.

Families tell us that lack of a diagnosis can make it difficult for children to access educational support through a SEND support plan or EHCP. Their specific needs

may not be understood well, and could be difficult to describe or explain without the help of a diagnosis.

Sometimes terms like Global Developmental Delay can be confusing or easily misunderstood as a condition in themselves. Often this is a symptom that can indicate that there is an underlying genetic condition which has yet to be identified. The term 'delay' is sometimes taken to imply that their development will catch up and can also create delays in assessment or recognition of need.

This can make it more difficult for families to request that their child is assessed for an EHCP, or to gain access to an appropriate school with the right level of support. Without a diagnosis and associated symptoms decision makers may not fully understand the long term nature of the child's condition.

The struggle to access adequate support can lead to strained relationships between families and education authorities and/or schools. Working in collaboration with other professionals and the child's family can be particularly helpful in the absence of specific guidance.

Developments in genetic testing

In recent years, whole exome and genome sequencing has begun to transform the way that genetic conditions are diagnosed and, where possible, treated.

Many undiagnosed children and their families will have been invited to take part in nationwide research studies such as Deciphering Developmental Disorders (DDD) Study and the 100,000 Genomes Project. The ability to sequence the whole genome is set to revolutionise the way that doctors practice medicine, but there is still a very long way to go. The current process is very lengthy and many families wait years for news and with a diagnostic rate for sequencing of around 30%, for many families there are still then no answers.

To find out more about the difference between exome and genome sequencing visit: bit.ly/rrexomeandgenomesequencing.

Understanding the limitations of a rare genetic diagnosis

Whilst a diagnosis can be invaluable when it comes to accessing support, an extremely rare diagnosis can leave families feeling the same as when they were undiagnosed.

Often a child with an undiagnosed genetic condition can receive a diagnosis of an extremely rare genetic condition that little (or nothing) is known about. If this is the case, many questions will be left unanswered, such as how the condition will develop or whether the condition is likely to be life-limiting.

For more information about family experiences of a rare diagnosis from genome sequencing visit: bit.ly/rrexomeandgenomesequencing.

Case study: Benji

Benjamin (Benji) is a very content, loving little boy. He loves watching lights and shadows, such as sitting under a tree watching the leaves move against the sky. He also enjoys the feeling of air on his face and he loves riding in the car with the window open, or with rain pattering on the roof. He likes music, particularly bells and jingly-sort of noises. He loves water: swimming, hydrotherapy and bath-time.

SWAN life

Benji was identified as having a syndrome without a name three weeks before his birth, when anomalies in his brain were detected.

Within the space of 24 hours we went from an uncomplicated pregnancy to MRI scans, consultants, and palliative care plans, and we were offered the option to terminate the pregnancy. I realised then that life was going to be very different for our whole family, whatever we decided to do.

Benji is now five and doing great! He has microlissencephaly which means his brain is very small and very smooth.

This creates several challenges for Benji. His motor control is limited and he has severe reflux (his stomach contents come the wrong way out of his stomach and up towards his mouth) and an unsafe swallow so any liquid that is in his mouth tends to go into his lungs and cause chest infections. Benji is epileptic and visually impaired. He is on more than a dozen different medications and has twice-daily chest physio and nebulisers to keep his chest clear. He is also unable to regulate his own temperature meaning he really struggles in hot and cold weather. Similarly he cannot control his sleep cycles and tends to follow a different sleep pattern from the rest of the family.

Benji is non-verbal and we have really struggled to find ways to communicate with and understand him.

Although he cannot yet use his hands to work a switch, he has worked so hard at his head and trunk control and now has the ability to turn his head to either side to control a switch with his cheek.

This totally awesome news means he can now make a choice between two options, and represents a huge amount of effort on the part of Benji, his teachers, support workers and occupational therapists.

We can work on this to develop a more complex, step-by-step communication method.

Being undiagnosed

To try and find the genetic cause of his disabilities, Benji is on the Deciphering Developmental Disorders (DDD) project and the Scottish Genomes Project (the Scottish arm of the 100,000 Genomes Project).

Our main reason for undergoing testing was to find out the risks of any future children having the same condition as Benji. However, the results took too long for us and we went ahead and had his little sister anyway! Because we had no diagnosis, this pregnancy was an incredibly

stressful time for us. I really hope that we will find out that Benji has a de novo genetic change, and therefore his sisters will not be carriers of any 'faulty' genes and won't have to worry about their own children inheriting the same condition.

A diagnosis would also help us to plan for the future. Being undiagnosed, we don't know what Benji's long-term prognosis might be.

I don't know whether to spend our time and energy planning for the long-term (trust funds, guardianship, care plans, and so on) or to use that time making memories in case we don't have very long with our son.

A diagnosis would make it so much easier to explain Benji's condition to people. It would make it easier to fill in forms, and easier to get funding from organisations that need a 'diagnosis' box filled in before they will even look at your application. A diagnosis would mean the chance to find other people with the same condition as Benji, learn from them, share our stories, and feel less alone in the world.

Finding SWAN UK

Fortunately, about a year after Benji was born we discovered SWAN UK. Those early days were a complete blur of caring for Benji 24-7 whilst also looking after his big sister who was only one year old when he was born. I remember feeling completely alone and ill-equipped as we struggled to make life-changing medical decisions with no-one to turn to for advice or friendship.

Now, the SWAN UK family provides us with support, understanding, answers to stupid questions, advice on serious questions, offers of help when stuck, a place to offload equipment we no longer need and emotions we can't hold in, family days out, new local friends, a feeling of belonging, … oh, and balloons! Benji loves the balloons!

When Benji was first born I was scared, confused, and angry that I had been thrown into all these new roles of doctor, nurse, carer, teacher, advocate, without any training whatsoever. I felt very guilty about the impact having a disabled child would have on our family, especially our daughter Jackie. I didn't know how to even explain Benji's condition or what was happening to us. I felt very alone. Now, I feel I have grown into all those roles and relish them! I feel capable of fighting for whatever Benji needs, and able to help and advise others in similar situations. I relish the changes he has brought to our lives, and I am immensely grateful for what he has taught me. I have made new friends and have a new, more inclusive outlook on life. I can't imagine life without Benji.

Looking forward

I know that my future will never be the same as I will continue to campaign, advocate, and work in my community to make it more accessible and inclusive for everyone. If we lose Benji, there will be an enormous hole in our lives, but I'm so glad we had the privilege to know and love him and to join the SWAN UK family. Learning from other parents and from disabled people has been the best thing about the journey and has totally changed my outlook on life.

SWAN UK is my tribe. It's my friends and family, and it's where we belong.

We look out for each other and help each other both virtually and physically. Although we came into SWAN UK through my son's lack of a diagnosis, I know that even if we get a diagnosis, we'll always be a part of SWAN UK.

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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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