RARE SYNDROMES

This fact sheet has been written by parent carers for parent carers.

What are rare syndromes?

A condition is classified as 'rare' if it affects fewer than five people in every 10,000. However, in some cases, families may find that their child has been diagnosed with a condition that is so rare that there are only several cases in the whole of the UK, Europe, or even the world. This can make parent carers feel isolated, as they may feel that no one truly understands what they and their child are going through.

However, for some rare syndromes, international support groups have been established to link people around the world so they can help each other and share information. Parent carers whose child has a new diagnosis of a rare syndrome may find it useful to start with Unique, or Syndromes Without a Name (SWAN), see page 2 for details, which hold information for many of these organisations.

Getting a diagnosis

It can be very difficult for families to get a diagnosis for their child. The first step is to request a referral to your NHS Regional Genetics Centre. You can do so through your Community Paediatrician or GP. The process involves medical staff taking samples from your child with the aim of identifying a genetic cause for their condition. See page 2 for contact details.

It's important to remember that even with genetic testing, there are still many conditions that are so rare that clinicians are unable to give it a name. Some children's features and symptoms will be unique, while others will fit more than one condition.

Medical advances, particularly in genetics mean that new conditions are being identified all the time. A major clinical study 'Deciphering Development Disorders' has been collecting genetic information from around 12,000 children since April 2011. By using new testing methods and techniques, it hopes to increase the chances of finding the causes of rare syndromes and the diagnosis rate for children with these conditions.

What if a specific diagnosis is not possible

Sometimes clinicians are unable to give you a reason and name for your child's difficulties. This can be very frustrating, as parent carers often want to know why their child has a disability and what caused it. Instead specialists may describe the characteristics of the condition. For example, they may say that a child has a 'learning disability' but not be more specific about the cause. Parents also want to know what may happen to their child in the future and what the implications might be if they want to have more children or if their other children want children of their own one day. Read our companion fact sheet on 'Living without a diagnosis' for more advice - see page 2.

If a child is not meeting two or more developmental milestones at an appropriate age and the cause is unknown, doctors may say that they have 'global development delay'. This means they may have difficulties with their motor skills, speech and language, cognitive or social and emotional skills.

Types of treatment

Some parents worry that if their child has an incredibly rare syndrome that they will not be eligible for support. However, you should be able to access the support of the same services as other families whose children have disabilities. It is worth remembering that many strategies and techniques work for all sorts of different conditions, so these can be adapted to help your child, too.

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The types of treatment needed for children with rare syndromes will vary according to their individual symptoms and needs. However, some of the following therapies may be suitable:

- **Occupational Therapy (OT)** – can help your child gain independence and master everyday tasks such as eating and going to the toilet.
- **Physiotherapy** – intervention to help a child to maximise their range of movement and improve posture.
- **Speech and Language Therapy (SALT)** – can be useful in helping children speak or develop alternative means of communicating such as sign language, for example.

Ask about

- **Information, Advice and Support (IAS)** – Amaze supports families of children and young people with SEN and disabilities in Brighton and Hove. We have a helpline, publications and a website. We also run workshops and courses and can offer one-to-one support with EHC planning and making DLA and PIP claims. Call Amaze on: **01273 772289** or visit: **www.amazebrighton.org.uk**.
- **Compass Card** – a free leisure discount card administered by Amaze for 0 to 25 year olds with SEND registered on The Compass. Call Amaze on: **01273 772289**.
- **Independent Support** – Amaze's Independent Supporters provide extra advice and support to young people and parent carers who are going through the process of getting an EHC Plan in Brighton and Hove and Sussex. For Brighton and Hove Independent Support, call: **01273 772289**. For Sussex Independent Support, call: **0300 123 7782.**
- **Making a claim for DLA** – if your child's care or mobility needs are significantly greater than the needs of their peers you may be able to claim Disability Living Allowance for them. Find out more at: **www.gov.uk**. Amaze can give you advice and practical support with making a claim. Call the helpline on: **01273 772289** or email: **helpline@amazebrighton.org.uk**.
- **Integrated Child Development and Disability Centre (Seaside View)** – may diagnose, treat and support your child. Assessments will usually be via a health or education professional but you can contact Seaside View direct on: **01273 265780**.
- **Making a claim for DLA** – if your child's care or mobility needs are significantly greater than the needs of their peers you may be able to claim Disability Living Allowance for them. Find out more at: **www.gov.uk**. Amaze can give you advice and practical support with making a claim. Call the helpline on: **01273 772289** or email: **helpline@amazebrighton.org.uk**.

Further reading and useful links

- **Children Living with Inherited Metabolic Diseases (Climb)** – go to **www.climb.org.uk**, call their freephone helpline: **0800 652 3181** or email: **info.svcs@climb.org.uk**
- **Contact A Family** – has a guide, '***Living with a rare condition***' which can be downloaded from their website: **www.cafamily.org.uk**. Also runs 'Making Contact', a networking service that matches families whose children have rare conditons. Email the family linking officer: **louise.derbyshire@cafamily.org.uk** or visit: **www.makingcontact.org**.
- **Deciphering Development Disorders** – a major clinical study on rare syndromes. Go to: **www.ddduk.org** for more information.
- **Genetic Alliance UK – www.geneticalliance.org.uk**, tel: **0207 704 3141**, email: **contactus@geneticalliance.org**.
- **'Living without a diagnosis' fact sheet**: Download this Amaze fact sheet from **www.amazebrighton.org.uk/resources/publications/fact-sheets**
- **Local Offer** – The local authority's online listing of all the services and support that are available to families with children with SEN and disabilities in the area. Visit **www.brighton-hove.gov.uk/localoffer.**
- **Orphanet** – lists European patient organisations for rare conditions. Go to: **www.orpha.net.**
- **Unique** – go to: **www.rarechromo.co.uk**.
- **Syndromes Without a Name (SWAN UK)** – go to: **www.undiagnosed.org.uk**.