Amaze FACT SHEET Updated Oct 2024

RARE CONDITIONS

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

What are rare conditions?

A condition is classified as 'rare' if it affects fewer than 1 in 2,000 people. 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 the Genetic Alliance, see page two for details, which hold information about genetic, rare and undiagnosed conditions.

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 service. 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.

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 collected genetic information from around 12,000 children. By using new testing methods and techniques, it hopes to increase the chances of finding the causes of rare syndromes; and improve 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. See our companion fact sheet, ***Living without a diagnosis***, for more information. This can be very frustrating, as parent carers 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 may also want to know what might happen to their child in 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.

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 named 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.

RARE SYNDROMES

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

- **Amaze - SENDIASS (Special Educational Needs and Disability Information, Advice and Support Service)** offers impartial, confidential advice on anything to do with SEND for 0 to 25 year olds. Parents, carers, children and young people under 25 with SEND living or going to school in Brighton & Hove or East Sussex can use the service. Email **sendiass@amazesussex.org.uk**, call **01273 772289** or visit **https://amazesussex.org.uk**
- **Amaze - Parent groups & befriending** – one to one befriending and regular support groups for parent carers across Brighton & Hove and East Sussex. For details of all our groups, visit **https://amazesussex.org.uk/parent-groups-and-befriending,** call: **07484 051755** or email: **marie@amazesussex.org.uk**
- **Child Development Teams -** consultant paediatricians, specialist nurses and therapists who assess and support disabled children up to 11. Ask your GP, health visitor, your child's school or another professional working with your child for a referral. In Brighton & Hove this service is based at the **Seaside View** Child Development Centre. In East Sussex, it is based at **Community Paediatrics Parkview** in Bexhill, Kipling Outpatient Dept in Hastings and satellite clinics. Therapies and equipment services in East Sussex are provided by the Children's Integrated Therapy and Equipment Service (CITES). Contact Therapy One Point on **0300 123 2650** or visit **https://www.eastsussexchildren.nhs.uk/about/childrens-integrated-therapy-services.**
- **Contact's Rare Conditions Network** - national charity Contact has set up a network that brings rare condition support groups together to share ideas and expertise, and strengthen group relationships. They also run free workshops for families whose children have rare conditions. Visit **https://contact.org.uk/help-for-families/information-advice-services/health-medical-information/rare-conditions/contacts-rare-conditions-network/**
- **Disability Living Allowance (DLA) –** if your child's care or mobility needs are significantly greater than the needs of their peers you may be able to claim DLA for them. Visit **www.gov.uk** and search for DLA. Amaze can also give you advice and support with making a claim. Visit **https://amazesussex.org.uk/benefits-advice** or call our SENDIASS helpline on **01273 772289.**

Further reading and useful links

- **Amaze's *Living without a Diagnosis* fact sheet -** see **https://amazesussex.org.uk/resources/fact-sheets/**
- **Contact's Rare Conditions information**. Contact publishes a guide, '*Living with a rare condition*' which can be downloaded from their website here. **https://contact.org.uk/help-for-families/information-advice-services/health-medical-information/rare-conditions.** They also collate and publish an A-Z of all conditions with explanations and support information.

RARE SYNDROMES

- **Deciphering Development Disorders** – a major clinical study on rare syndromes with regular updates on the project. Go to: **https://www.ddduk.org** for more information.
- **Genetic Alliance UK**– **https://geneticalliance.org.uk,** tel: **0300 124 0441**, or email: **contactus@geneticalliance.org.uk**. Also runs **Rare Disease UK**: **www.rarediseasе.org.uk;** and **Syndromes Without a Name (SWAN UK)**: **www.undiagnosed.org.uk.**
- **Orphanet** – lists European patient organisations for rare conditions. Go to: **www.orpha.net.**
- **Unique** – a rare chromosome charity. See: **https://rarechromo.org**, tel: **01883 723356** or email: **help@rarechromo.org**.