Reaching Families FACT SHEET Updated October 2023

RARE SYNDROMES

This fact sheet has been written by parent carers for parent carers.
Clinical information has been approved by West Sussex practitioners.

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 the Genetic Alliance, 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.

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 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. 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.

Many parents also want to know what the future will hold for their child, and what the implications might be if they want to have more children. For more advice, see the 'Living without a diagnosis' fact sheet, which can be downloaded from: **www.reachingfamilies.org.uk**.

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. 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

- **Disability Living Allowance (DLA)** – your child may qualify for DLA, a state benefit that will help with their care. For further information visit: **www.gov.uk/disability-living-allowance-children**.
- **NHS Regional Genetic Centres –** the centre for West Sussex is South West Thames Regional Genetics Service, c/o Department of Medical Genetics, St George's University Hospital. For information go to: **www.stgeorges.nhs.uk/service/specialist-medicine/clinical-genetics**.
- **Portage** – a home-based educational programme tailored to a child's individual needs. Available to very young children. Crawley and East Grinstead: **01293 572480**, Chichester and Horsham: **01243 536182**, Mid Sussex: **01444 243150**, Worthing: **01903 242558**.
- **Reaching Families** – provides training, information & other fact sheets related to this topic. We also offer benefits advice, peer support, a Facebook group & handbook (see below) for parent carers of children & young people with SEND in West Sussex: See: **www.reachingfamilies.org.uk**.
- **West Sussex Children's Services** – support from health and social care. If your child is under 18, contact the Integrated Front Door (IFD) for West Sussex Children's Services. Tel: **01403 229900** or email: **WSChildrenservices@westsussex.gov.uk**. The IFD is also an entry point for the Children with Disabilities (social care) service for children & young people with severe and/or complex disabilities. Eligibility criteria apply – See: **https://westsussex.local-offer.org/services/265**. For over 18s call the Adults' CarePoint: **01243 642121**, or email: **socialcare@westsussex.gov.uk**.

Further reading and useful links

- ***Making Sense of it All: From Birth to Adulthood*** – Reaching Families' handbook for parent carers of children and young people with SEND in West Sussex. This provides essential information on money matters, including claiming DLA and PIP, as well as social care, health, leisure, travel, education and employment. Go to: **www.reachingfamilies.org.uk/guides.html**.
- **Contact** – has a guide, *'Living with a rare condition'* which can be downloaded from their website: **https://contact.org.uk**. Their rare conditions information officer can also assist families to find information on their child's condition and can be contacted via the helpline: **0808 808 3555**.
- **Genetic Alliance UK**– **https://geneticalliance.org.uk**, email: **contactus@geneticalliance.org**, tel: **0300 124 0441** They also run **Rare Disease UK** and **Syndromes Without a Name (SWAN UK)**.
- **Metabolic Support UK** (formerly known as Climb) – go to **www.metabolicsupportuk.org** or call their helpline on **0845 241 2173**.
- **Orphanet** – lists European patient organisations for rare conditions. Go to: **www.orpha.net**.
- **Unique** – a rare chromosome charity. See: **www.rarechromo.org**, tel: **01883 723356** or email: **info@rarechromo.org**.
- **West Sussex Local Offer** – go to **https://westsussex.local-offer.org** for local services, support and details of Short Breaks providers for children & young people aged 0 - 25 years with SEND.