RARE DISEASE DAY®

RARE DISEASE DAY IN A COVID-19 ENVIRONMENT:

THE BEACONS OF LIGHT ALONG THE PATIENT JOURNEY

For many people and families living with a rare disease, their journey can be very complex. At times, that journey can even be scary, frustrating and confusing. However, when your life is touched by rare disease directly as an individual or as a family or carer, it can often mean entering into a new community of dedicated people who ensure that you are not alone and unsupported in that journey. Here we shine a light on just a few key individuals and organisations who's essential support and guidance illuminates the patient journey, provides a network of knowledge and support and ultimately, improves quality of life.

For many people, the start of the journey doesn't always begin with a diagnosis: on average, rare disease patients can wait 4 years to receive a diagnosis with some waiting over 20 years(1). With 8 out of 10 rare diseases considered to have a genetic component(2), a key stakeholder to help facilitate an accurate diagnosis is the **Clinical Geneticist** who can become involved at several different stages:

> *(My role can begin...) If a couple are planning on becoming pregnant and there is a rare disease in the family that's genetic, if we find something on the scan that suggests a genetic problem; or, immediately post-natally with delivery and when a baby is in the special care unit; or, I can be involved when someone is 5 years old and they are having learning difficulties at school and they need to find out why.*

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New gene sequencing methods have recently opened doors to earlier diagnosis, though, with the multitude of possible gene mutations, clinical interpretation in terms of diagnosis or severity is not always possible. Evaluation of subtle physical attributes (such as facial features) to aid in clinical diagnosis is an important part of consultations by clinical geneticists. Due to the COVID-19 pandemic, this has been an aspect that has been particularly challenging, as face-to-face consultations to assess the patient are essential. The very sensitive and emotive conversations with parents and families are also difficult in the virtual space, with no additional professional training provided for this new approach. A close relationship between the **Clinical Geneticist** and **Genetic Counsellor** helps to provide support for parents to start to make informed choices and life strategies.

> *"They are professional communicators and have counselling skills as well...If we have a patient who is struggling with a diagnosis, we would involve a genetic counsellor as a counsellor, not just a genetic communicator."*

Clinical Geneticists and **Genetic Counsellors** are part of the multi-disciplinary team and network of dedicated professionals who ensure continuity of care and treatment for people with rare diseases. Children with rare diseases can often have specialist developmental, behavioural and educational needs. Prior to 1975, children with learning disabilities were dealt with by the health system and did not have the benefit of education. Today, the role of schools, like **Park Lane Special School,** Macclesfield, Cheshire, is vitally important to provide education as well as life skills such as shopping, cooking and cleaning, which really impacts on quality of life. School **Head Teacher, Lorraine Warmer,** pointed out that it's the pupils at Park Lane that make her school so special!

> *"The children make it such a positive place, what you see is children and young people who are inspiring because they are doing so much to overcome the difficulties they have, but they are doing it in a positive way – there's lots of smiles, there's lots of laughter, there's lots of enthusiasm for learning."*

Children can start at Park Lane from the age of two and stay until the year in which they turn 19. While their education, health and care plan focuses on what the children can't do, the school explores what they can do and like to do, through a multi-disciplinary team of **Teachers, Teaching Assistants** and specialists from **Speech and Language Therapists, Occupational Therapists, Physiotherapists** and **Music Therapists.**

The school has remained open during the pandemic and technology has been key for children who needed to stay at home for them to access music therapy sessions and share artwork with their classmates. The school's open-door policy and support through communication boards enables parents to provide the best learning environment for their child and to help parents and families cope with some of the challenges such as specific sensory and behavioural needs.

The school becomes part of the extended family with strong bonds forged between families and teachers, as a child can be at the school for 17 years in total. This makes occasions like the leaver's assemblies very special indeed – a time when pupils and parents come to terms with the ending of school and a reflection on how much the children have developed and achieved through their lives at Park Lane – before the young adult is guided on the next part of their complex journey, "They deserved to be celebrated."

Another vital stakeholder in the rare disease community are the specialist charities providing tailored disease-specific guidance and support, for children and their families. **Chairman Charles Blockley,** of the **Cornelia de Lange Syndrome Foundation UK and Ireland,** and parent of a child with Cornelia de Lange Syndrome (CdLS), passionately explained that the mission of this charity, run by volunteers, is to ensure early and accurate diagnosis of CdLS, promote research, and enable individuals, families, friends and professionals to make informed decisions and plan for the affected person's present and future. In CdLS, clinical diagnosis is paramount, as, despite identification of seven mutations, only 70–80% of people have a genetic diagnosis.

> *"One of the issues with CdLS being so rare is that it's sometimes very hard to get clinical support within the community. Typically, your GP may have heard of it but probably hasn't dealt with it before."*

A major achievement of the charity was to orchestrate development and publishing of treatment guidelines in 2018, in the journal, Nature Reviews Genetics(3). The guidelines were cascaded to professionals to aid both identification and diagnosis by clinicians and GPs. Importantly, the charity worked hard with their scientific advisory committee to provide an accessible summary which could be provided to families to facilitate both understanding and communication between patients and healthcare professionals. During the pandemic, the charity has connected with families increasingly through social media and facilitates their interactions with healthcare professionals and other families via their virtual annual conference.

> *"We held our first virtual congress in October this year, which was very successful...it reached families that sometimes can't actually travel to the in-person conferences."*

As with all our beacons on this, their mission was not halted by the pandemic, only adapted to the virtual environment. Our beacons have shown that despite the challenges of the pandemic, having a life touched by rare disease does not mean you are alone. Epidemiological facts and figures may describe individuals as one unique person in tens or hundreds of thousands but thanks to the commitment and dedication of a wide range of volunteers and professionals, the rare disease community is stronger than ever.

RARE IS MANY
RARE IS STRONG
RARE IS PROUD

References:
1. https://www.raredisease.org.uk/wp-content/uploads/sites/7/2019/02/Illuminating-the-rare-reality-2019.pdf (last accessed February 2021)
2. https://www.eurordis.org/sites/default/files/publications/princeps_document-EN.pdf (last accessed February 2021)
3. Kline, A.D. et al., Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. Nat Rev Genet 2018;19:649–666. https://doi.org/10.1038/s41576-018-0031-0