Dr Beverly Searle, Unique's CEO spoke at the RDUK reception in the Houses of Parliament to celebrate rare disease day and the launch of the England Statement of Intent regarding implementation of the UK's Rare Disease Strategy. Here is what Beverly said:

Thank you for this opportunity to share some of my own personal experiences of being affected by a rare disease and what I hope the English Statement of Intent will mean for patients and families affected by rare diseases.

As you have heard I am the CEO of Unique, the charity working with 11,000 families affected by a wide variety of rare chromosome disorders. But I'm also a hybrid! Let me explain. For many years I had worked as a post-doctoral research biologist with a special interest in genetics and biochemistry. My home life seemed blessed - a loving husband, a healthy first born son and a second baby on the way. Sadly, my professional and personal lives were about to collide in the most spectacular fashion. From the moment my daughter Jenny was born prematurely it was obvious that our lives would change forever. Jenny was born with a severe midline facial cleft and brain abnormality but what had caused these defects? Unlike many families seeking a diagnosis, we were lucky to have a diagnosis of a rare chromosome disorder within a week. However, we were shocked to find that there was no information about the lifetime effects of Jenny's rare disorder and no defined care pathway telling us how to treat her. Although our consultant paediatrician was very supportive, neither he nor we could find a specialist centre or individual clinician or researcher expert in Jenny's chromosome disorder who could advise us and coordinate the specialist care she needed in the long term. We found ourselves unwilling participants on a terrifying roller coaster into the unknown, lurching from one health crisis to another. Life with Jenny was like learning to be a medical doctor the long way round. Every year would open several new chapters in the medical books and all this in a child who was profoundly learning and physically disabled. At one time we were visiting nine different hospitals to see doctors specialising in one or other of Jenny's many health issues. Along the way we were lucky enough to find some hugely talented healthcare professionals expert in Jenny's individual symptoms but no experts in her disorder who could coordinate her, and our, care holistically. You can imagine the immense strains it put on all aspects of our family life. We eventually joined the then fledgling support group Unique where we could meet other families in similar situations. Fortunately my husband is an IT specialist with expertise in databases, so together we created Unique's registry /database in 1996 in order to collect information about the lifetime effects of all our members' rare chromosome disorders. In fact Unique's database has turned out to be by far the best source of information for us about Jenny's chromosome disorder, helping us and her clinicians diagnose new health problems along the way. As Jenny moved from adolescence into adulthood, we were very disturbed to find ourselves falling into the abyss of transition when we lost most of our paediatric support with little to replace it. Hardly any adult healthcare professionals would or could pick up the baton and it was left to Jenny's GP to try to coordinate her care with the goodwill of her wonderful professor of paediatric endocrinology who agreed to keep her on his books for lack of a suitable alternative in adult services. As it turned out we didn't need adult services for long because, very suddenly and unexpectedly, Jenny died in her sleep just over 2 years ago when she was 21 years old.

So what do I hope that the English Statement of Intent will mean for patients and families affected by rare diseases? Most of all I want to see improved patient-centred services with a clear understanding of what patients and families can expect from the NHS, wherever they live in England and at all stages of life. These services should include:

1. A rapid and accurate diagnosis, avoiding the hugely stressful diagnostic odyssey experienced by many families and the endless rounds of unnecessary and frankly wasteful doctors' appointments, tests and inappropriate treatments.
2. Equity of access to timely and appropriate information for rare disease patients across the whole of England, including the opportunity to sign up to database/registries for all rare diseases

3. A clear personal care plan for everyone with a rare disease, integrating health, social care, education and any other essential services, thus leading to better, holistic care for them and their families
4. A patient-centred, coordinated approach to treatments and therapies, as well as specialist healthcare and social care support
5. Creation of specialist expert clinical centres, whether physical or virtual, disseminating appropriate information and advice to patients and their clinicians locally, and promoting and participating in world-leading research into rare diseases and their treatments and therapies
6. Empowering and supporting patient organisations like Unique so they can contribute their considerable understanding and knowledge of rare diseases, and their psycho-social consequences, to help develop improved services.
7. Better education and training for health professionals so that they recognise the possibility of a rare disease when making a diagnosis.

Implementation of all 51 commitments of the rare disease strategy offers a great opportunity and huge benefits not only to patients and their families but to the NHS as we work together to perfect services for, and research into, rare diseases. It's incumbent upon us all, from patients and families and their representative organisations through all levels of the NHS and government, to continue to collaborate to make it happen as soon as possible.

Thank you for listening.