The importance of research when you have a rare disease

#rarediseaseday

A rare disease patient support group is running its first-ever research project, in collaboration with the Patient Led Research Hub on the Cambridge Biomedical campus.

The idea for the study came from the patient support group Ring20 Research and Support UK CIO, which was set up to make it easier for families affected by Ring Chromosome 20 Syndrome [r(20)], to get in touch with each other and to raise funds for research.

The study comprises developing online surveys and a workshop to be held in May, to discuss findings and plan a trial.

The group's co-founder Allison Watson said: "R(20) is an ultra-rare disease with no known cure, affecting fewer than 150 people worldwide."

"We want to see if Ketogenic Dietary Therapy (KDT) is an effective treatment."

The group now supports more than 100 families worldwide, including Ella and her family.

Ella's mum Kirstie said: "Initially the seizures could be controlled by drugs but then on a family holiday Ella was back to square one, having 40-50 seizures a day." Seizures increased to 80 a day before Ella was diagnosed with r(20).

Research

Ring20 co-founder Allison Watson said: "We hope our findings will provide a basis for future clinical trials into whether KDT could be a beneficial treatment for r(20) patients.

"At last we feel we are able to do something proactive for our patient families."

Ella is now on KDT but support is a continual struggle.Kirstie says: "Every day is a struggle – getting dressed, having medicine, just doing the things that we all take for granted. We don't know what the future holds for Ella, all I know is that she is one special little lady."

Patient Led Research Hub

ring 20 research & support uk

@Ring20UK

ring20researchsupport.co.uk