NHS
Central and South
Genomics

WHOLE EXOME SEQUENCING: ELI

We have seen significant developments in genomics over recent decades, and the science continues to progress at a rapid rate. It's crucial to remember that each advancement has life-changing impacts on people across the world: people like Eli and his family. We caught up with Anna, Eli's mum, to learn about their story.

Eli was born full term in February 2022, the youngest of three children. For the first eight weeks of his life, he experienced no complications, but a couple of months in he started to develop some issues. He struggled to feed, for which he saw a lactation consultant, and had a rattling sound in his throat. One day over Easter 2022, his parents noticed his head was continually dropping.

Anna stated, "We saw this and of course started frantically googling. A Facebook page for Infantile Spasms UK came up, where we saw resources showing what a baby looks like during a seizure. It seemed to match, so we rushed to hospital with Eli.

"They confirmed he was having infantile spasms. We ended up staying in hospital for a week, where Eli started treatment."

Anna continued to research the issue and discovered that infantile spasms can sometimes occur with no genetic or neurological reason: she hoped this was the case for her son. His treatment was working and an MRI in June 2022 showed no issues. As part of the process, Eli was also put through genetic testing. Having been told results could take up to six months, his parents put it out of their minds.

However, shortly after the clear MRI results, Eli's parents were contacted by their neurologist, who told them the testing had identified something: a variant of the CYFIP2 gene, causing microcephaly, epilepsy and severe intellectual disability. A report was sent with the full diagnosis of CYFIP2 Arg87cys.

Anna stated, "It was the most devastating thing I have ever read. All of a sudden, my beautiful 4-month-old boy wasn't going to have the life we thought he would have. He'd probably never learn to talk or walk.

"In the early weeks, we didn't understand – we knew what genetics was, but we'd never experienced anything like this. It was so tough, and I was crying every day. My friends and family tried to be positive: though they were just trying to help, I remember thinking, 'just give me my grief!'".

Anna chased up an appointment with a Clinical Geneticist. Though she describes the appointment as "devastating", she notes it gave her and her family the knowledge they needed: they learnt that this variant had only been identified in 2017, with just 20 other children in the world with the same diagnosis.

It also offered an opportunity to understand the genetic element better: with two older children, they wanted to be clear if this was something that could affect them or their future children.

After the diagnosis Eli's family began to adapt to their new normal, with Anna stating, "What helped me as a person the most was going back to work. I'd been spending so much time reading things. I went through periods of complete fixation, on things like measuring his head. Going back to work made things more normal for me."

She also joined a Facebook group for affected patients and their families, which provided community and understanding.

This community has not only provided support, but potentially life-saving information. Tragically, three children with the same diagnosis as Eli passed away in recent years due to aspiration pneumonia. Having learnt that the condition often causes issues with feeding and aspiration, Eli's parents chose to pursue a gastrostomy, and he is now fed through this. Anna commented, "Had we not known, we probably wouldn't have looked for this and pursued the gastric tube."

Eli still struggles with seizures, and this is something his parents are keen to fix, with Anna stating, "We never really worry about the genetics side anymore, because it's not something we can change, and Eli wouldn't be who he is without his specific genetics. The epilepsy feels like something we can address."

Now two years old, Eli has continued to develop. Whilst developmentally delayed, he is able to roll over, use his hands, and choose things through eye gazing. He has not developed speech but is babbling, as well as being able to sit upright independently for a few minutes and stand in a standing frame for around an hour a day.

Eli's family are grateful for his diagnosis, with Anna noting the benefits of "Finding our genetic family". She stated, "Without a diagnosis, we would be constantly worrying about what was wrong with him – we wouldn't have any answers.

"Having an answer, as brutal as it was at the time, ultimately helps. We have access to services; Eli was in speech and language therapy and physiotherapy by 6 months and already has an education, health and care plan. We were able to apply for disability living allowance and Eli has a paediatrician.

"We can plan for the future, and crucially we are able to talk to our older two children about how and why their little brother is different."

Looking back to the time when Eli was diagnosed, Anna is keen to ensure she is able to support others who find themselves in a similar position.

"Having someone to validate my grief would have really helped me. But over time, you do come out the other side, enjoying your child for who they are.

"Watching him develop has been amazing. He has the most wonderful laugh and the most magical smile. He is the most amazing little boy in the world, and I wouldn't be without him."