

I am number 17
Rebecca and Hannah's Experience

It's a difficult thing, to admit to yourself that you think there is something wrong with your child. It's harder again to say that statement out loud to your loved ones or health professionals. It becomes a battlefield when those professionals repeatedly dismiss your concerns. It took strength I didn't know I possessed to persevere for 18 months in the face of accusations of Munchausen's by proxy and statements such as "I can assure you Mrs Pender, there is categorically nothing wrong your child, she is just lazy" and "you're a paranoid, sleep deprived first time mum".

I started to question my own sanity, whether my daughter inability to crawl, walk or babble by 18 months was simply down to bad parenting. My heart and my gut knew differently and so I soldiered on. We wiped the slate and changed our medical practice and our health visitor. I invited the new health visitor to our house to see my daughter and I let it all out. Then came those three words I will never forget. "I believe you". I finally had an ally. Blood tests were ordered to rule out genetic causes in the first instance. And so, we waited.

We were sitting in the chiropractor's office having just spent another weekly appointment trying desperately to help my girl when the phone rang. It was her new paediatrician. She mentioned the blood tests and how the Microarray picked up small variants on Chromosomes and most of the time these were benign, didn't mean anything however Hannah's findings were significant. I stood there and let the tears fall. A mixture of heartbreak that my daughter did have something wrong with her and it was confirmed my child was disabled, and of relief that my daughter did have something wrong with her. I felt vindicated, I wasn't crazy, paranoid, delusional. I wasn't projecting and most importantly I was right and had advocated so strongly on her behalf and it paid off.

I cannot change our story or our experience, instead I hope to use it to show how things can be done differently and that by working together we can prevent scenarios like that happening.

In the last 5 years we have battled healthcare, social work, homelessness, DLA tribunal, education and public perception of my daughters' ability to be fully included. I've personally battled anxiety, PTSD, post-natal depression and chronic psoriasis stemming from chronic stress. I was in fight or flight mode for almost 5 years. I can't change what we have been through, but I am determined to help ensure that we change the narrative for families that walk the journey behind us.

It's very important to understand that I am not here to vilify our healthcare system. Its world class and has saved my daughter's life on countless occasions and she is kept alive and healthy because she has access to daily meds, oxygen and emergency care at point of need. We are very lucky. But that doesn't mean there are not improvements that can be made in practice and the attitudinal culture of hierarchy which sometimes exists.

Something needs to fundamentally change within our system. There needs to be more awareness surrounding rare diseases and conditions and what they mean for people. I would say patients but all too often the person is lost behind the diagnosis.

I wear many, many hats in my day to day life. I work full time as well as writing, advocating, raising awareness for rare genetic condition, with a side of inclusion activism thrown in for good measure. 95% of the time though, my favourite and most treasured hat I wear is being Mum to my three beautiful daughters, Hannah 5, Molly 3 and Daisy 1. The 5 percent it's not my favourite is because, to be honest, I have three children under 6.

The reason I do what I do, the person who gives me the drive and determination to change the narrative is my eldest Daughter Hannah who is almost 6 years old.

She is a daughter, a sister, a granddaughter and Nanas Hannah. She is a cousin, a friend, a student and a teacher. She is a bringer of light in what sometimes seems like quite a dark world. She was conceived in our hearts for 5 long years before she was actually conceived. She is an actress, a musician and singer, a selfie queen, a snuggle monster and a wind-up merchant. She loves music and curious George, she loves school and friends and dancing, handbags and dressing up and her

favourite past time is seeing how far she can wind up her little sister. She loves people and being included. She knows when she has been left out. She loves food but especially chocolate and gravy. She teaches unconditional love, patience, gratitude, resilience and strength. She also happens to have profound and multiple learning disabilities (PMLD). Hannah's primary diagnosis is Inv Dup Del 8p, a rare genetic condition with only 65 known affected worldwide. She also lives with a severe learning disability, epilepsy and complex arachnoid cysts in her pineal gland. She is non-verbal although EXTREMELY vocal, double incontinent, hypermobile and hypotonic. She has seizures which go into status epilepticus* and becomes life threatening in the blink of an eye. But first and foremost, she is Hannah.

We absolutely must "raise the bar" but first we must allow families to see the bar and know where it stands so we can raise it together, collaboratively and change the narrative. We can build a new path where every voice is valued and respected and families feel involved and central to their care.

Let's learn to listen to parents, listen to the experts in their own bodies and their own children. In Hannah's case I am her voice as she doesn't have one. Listen to people describing their symptoms and asking for help. Let's think outside the box rather than take a wait and see approach. Let's stop allowing parents to question their ability to parent and allow them to advocate on their child's behalf and be truly heard. Let's take a step back and look at the bigger picture and truly place the best interests of these people at the heart of their care. Let's not keep making the same mistakes of the past and stop an unnecessary rollercoaster of misdiagnosis or refusal to diagnose therefore stopping the access to the right medical and emotional support required to allow them to flourish.

** Status epilepticus is when a seizure lasts longer than 5 minutes or when seizures occur close together and the person doesn't recover between seizures (source: Epilepsy Foundation).*

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