Alice by Charlotte

Our twin girls, Alice and Verity, were born in April 2017. Alice was diagnosed with Prader-Willi Syndrome when she was 3 weeks old. It's hard to think back to those early days now that she is a happy 2 ½ year old toddler, but I hope that sharing our story will help other families dealing with a new diagnosis.

Alice and Verity were very much wanted IVF babies and, as I was expecting twins, I was closely monitored throughout my pregnancy. I spent a couple of weeks in hospital with pregnancy-induced hypertension before having a C-section at 35 weeks (because of concerns about Verity's growth), so when they arrived and all appeared well I couldn't believe my luck.

Whilst Verity was very alert and fed well, Alice was very sleepy, but I was assured that some babies were just tired after birth and no one seemed concerned. An hour or so later though the doctor noticed that Alice was a bit floppy, so she was taken to the Neonatal Intensive Care Unit, where she ended up spending the next 3 weeks, hooked up to monitors and fed through an NG tube (not what you expect when you are expecting). When Alice's condition didn't improve after treatment for a suspected infection, and an MRI came back clear, we were told she probably had a genetic condition, most likely Spinal Muscular Atrophy. They began planning palliative care as it was unclear how long she would live. In the meantime, we took Verity home without her sister.

When the spinal muscular atrophy test came back negative I breathed a huge sigh of relief. Although I knew they would now test for other, rarer, conditions, I couldn't help but think that maybe they would come back negative too. After all, I thought this sort of thing only happened to other people. A couple of days later though the doctors told us they had a result and this time it wasn't negative – Alice had Prader-Willi Syndrome. The doctors didn't know much about the condition. In fact, one consultant, who was retiring that week, told us that he had only come across it once before.

Perhaps it would have been a good idea to stay away from google (which really does show outdated information and worst case scenarios). Inevitably though, I read everything I could and finding out what Alice could face in the future broke my heart. Those early days were incredibly difficult as we tried to come to terms with Alice's diagnosis. I was just so sad that this had happened to Alice and to our family. To be honest I couldn't imagine ever feeling happy again.

Looking back, I was grieving for the life I thought we were going to have and I think I needed to go through that to move forward.

Gradually, I did start to feel better. Reaching out to the PWSA helped. Patsy from the PWSA came to visit us and I remember her telling me about all the things her daughter (who has PWS) liked doing. I think at that point I was able to start seeing Alice is more than just her diagnosis. I also joined a Facebook group for parents of children with PWS and, having felt so alone because no one I knew had been through anything like this, I found a group of people who knew exactly what I was going through, which was a huge comfort. I've made some wonderful friends through that group.

Alice got stronger and came home and after a while, we were able to take the NG tube out. She got more alert and started to smile and laugh. Although it took her longer to do these things than her twin sister, when she did it was absolutely amazing.

Alice and Verity are now 2 1/2 and life is good. All the appointments that are so overwhelming at the start are just part of our routine now. I went back to work when the twins were 1 (which is what I had always intended) and they attend a mainstream nursery where Alice is thriving. A couple of months ago she took her first steps and she's now walking around confidently. Her speech is coming along too. She enjoys going swimming and trips to the farm and we've even been on holiday.

We've also gone on to have another child. I was quite anxious during the pregnancy, but Estella is now 3 months and has fitted into our family perfectly. The twins really love being big sisters. Having another child has really brought home to me what a hard time we went through when Alice was born and I feel really proud that we got through those early days.

I know we will face more challenges in the future, but I feel much more confident that we will handle those just as we have handled those we have faced so far.

If I could go back in time to when we received Alice’s diagnosis and tell myself one thing, it would be this – There’s a lot to look forward to, I promise.