

When Jacob was diagnosed by Gemma Byrne

When I eventually got over the shock of having a boy (Alannah was our first born and for some reason I was convinced I was having another girl! She is non PKU) and the speed at which he eventually arrived, I was so happy to take my adorable baby home. From day one he was a real mummy's boy, wouldn't sleep unless he was in my arms or could hear me! We were home from the hospital for one night, trying to get to grips with our new routine when we got that dreaded phone call from the screening unit in Holles Street. They put us in touch with Temple Street who asked us if we could come in first thing the next morning. We spent that night crying and looking at Jacob, just wondering what we had done wrong, feeding him milk that I knew was doing him harm. My heart was broken. The next day provided some relief as well as raising some new questions for us! I remember a nurse coming in and saying "I'm sure you're wondering what he can have to drink at his debs" and I just thought, his debs oh my god this is really for life. Then in the midst of all the facts and figures someone said "the hopes and dreams you had for your little boy the day he was born are still the hopes and dreams you should have him, PKU doesn't change that".

So every day we spent in the hospital brought new information and more questions. I was quite sick after having Jacob and so my husband stayed in the hospital with him. I used to lie awake in my bed just thinking it all over and blaming us as parents for giving this condition to him. And then we got to finally take him home. I was terrified. The nurses said "doing the bottles and taking his bloods will all become routine" and I remember thinking they were crazy, how could any of this become the norm. But it does.

When we got home I just wanted to stay in the house with my little baby. I spent hours examining his face, his eyes, his hands and toes just looking to make sure they were normal. As he grew I would compare him to every baby I saw, even though I wasn't a first time mum, it all seemed really new and different. Friends and family were very supportive but I hated all the questions because I just didn't want to think about it and I didn't want other people to view him differently.

I remember he had a turn in one of his eyes (as most kids do, even my daughter did) and just blowing it all out of proportion. I was lucky to have a really good GP who was always there to listen to me and always took my concerns on board. And the support from all the team in Temple Street was phenomenal. There were weekends when we would call up about the most ridiculous concerns but they listened and they advised where they could.

Jacob developed at an outstanding rate. My only comparison was his older sister and their cousins and he walked and talked before all of them! To this day he could buy and sell them all. It took a while for me to realise that I didn't have a kid with PKU but that I had a beautiful, healthy baby boy. It took me a while to show him off and let other people dote on him the way I did. But it did come.

Has this had a big impact on our lives? It sure has. Nothing is straightforward anymore. You soon realise how big a part food plays in our lives and in particular our social lives. We have just learnt to be prepared, use the exchanges wisely (currently Jacob is on 4 exchanges). But we have found good places to eat out as a family and ways of having family meals at home which work for the all of us.

Could things be worse, of course they could. Of all the conditions to have this is very manageable. Are there days when you just tear your hair out because you can't think of another way to make vegetables look appealing, there certainly are! There have been days when I just can't take saying no to Jacob any more but I am his parent, I have to be strong for him now as this is something he will have to deal with for the rest of his life and it's my job to give him the strength to do that.

A lot of the time I feel sorry for my daughter. She gets told "no" a lot, so as not to make Jacob feel left out. If she wants another bowl of cereal I often say, "Try some toast" as I don't want Jacob to get upset because he can't have any more cereal. But then there are times when she will come to me and say "mum I don't mind having toast, I don't want Jacob to feel left out". She knows about PKU, taking bloods, the hospital, his "special" milk and is also very aware of most of the foods he can't eat. She always asks me if Jacob is allowed certain things, like she might get some sweets at a party and will ask me if he is allowed to have one.

I am a full time working mum. I went back to work after 6 months with both of the kids, with Jacob it was more stressful with the added worry about food but we were just very clear to the crèche and au pair what was required and if ever in doubt to call. It took me months to be able to let my own mother have him over night, ridiculous stuff, she is probably more capable than me, but you can't help but worry.

Jacob is now 2 and a half. He is the funniest person I have ever met. Sometimes he drives me crazy when he won't eat his food but a lot of that is just his age and unfortunately for him he can't have the same luxuries with food that his peers can. He has a wicked sense of humour and is the most sociable person I have ever met, going for a walk with Jacob takes double the time as he stops to says hello to EVERYONE! He loves trains, cars, movies, dogs, and his sister (sometimes) and he just loves being a kid.

What do I think the future will bring for Jacob? I don't really know but what I do know is that the team in Temple Street will be there for me and for Jacob when the time comes.

