



# Alicia

## LIVING WITH MACROGLOSSIA, SEVERE SPEECH DELAY, BECKWITH WIEDEMANN SYNDROME, & HEMIHYPERTROPHY

I don't think the Doctors and Nurses understand how thankful I will always be for looking after my baby....

***“I wish our voices would be listened to a lot more.”***

Alicia was born 9 days overdue via emergency C section. Absolutely gorgeous, big brown eyes looking up at us. As new parents we didn't know if we over worried about if she drank too much of her bottle or had way too much wind. Fast forward a couple of months and we are down at the Public Health Nurse. She noticed Alicia's tongue was quite large and her speech was delayed for being just over 2 years old. We took her advice and went to the G.P who referred us to a Paediatrician in Tallaght Hospital in Dublin.

We waited maybe 6 months maybe longer. During the waiting for this appointment we weren't sure exactly why we were going to see her or what she was going to do but we had to have her speech checked and also the fact her tongue was large and always sticking out. We eventually got our appointment, we went and spoke to her and the outcome was that she had macroglossia and severe speech delay. She would make a referral to speech therapy. We were again on the waiting list for this appointment. This took months to get but once we started we did block appointments, meaning we might have 1 or 2 appointments a week!

We were also referred to Genetics in Crumlin Hospital in Dublin to have this investigated. After weeks of waiting we were called back in to see the doctor who confirmed there was a reason she had a large tongue. She had a condition called Beckwith Wiedemann Syndrome.

This is an overgrowth condition but also can cause Wilms Tumours. This meant Alicia would now have to have ultrasounds every 3 months until she is 8 years old. She will also get bloods done when her ultrasound is being done. This was a huge shock. How was this not picked up at birth? The reason is she didn't have any of the other characteristics of this syndrome when she was born.

We attended speech therapy when the appointment was sent out to us and the outcome was that Alicia had severe speech delay and macroglossia which she understood was affecting her speech.

We went back to the paediatrician doctor and she referred us to Temple St Craniofacial Team. Here we met Dr. Murray. He agreed her speech was delayed because of her tongue. In 2017 Alicia had a Tongue Reduction. I won't ever forget how afraid I was that this might not work but within months Alicia started putting words together. This took a lot of speech therapy and a lot of help from myself, her dad, my parents and anybody that was around Alicia. Without this surgery Alicia might not be talking today. We are forever grateful to Temple St and Dr. Murray's team there. They're our heroes!

As Alicia grows so fast we also got a referral to a dietician in case I need advice and to keep an eye on her growth. This thankfully is going well.

A year or 2 after Alicia;s diagnosis we got another diagnosis of Hemihypertrophy. Some children with this condition it is noticeable. Again with Alicia it was not noticeable. I was called to collect her early from playschool one day as she had been walking with a limp. I brought her to the GP who sent us to A and E. We were told it was irritable hip and if it was no better to come back. Unfortunately back we went a week later. No better. Eventually after x-rays we were told it looked like this was Hemihypertrophy. We were referred to Orthopedics at Crumlin Hospital. We have since been attending Physio but thankfully all is going well.

Alicia is now 7 nearly 8. We have had ultrasounds and bloods every 3 months. Thankfully we have had them all clear since her first one and nothing became a problem. We attend Crumlin twice a year.

Due to COVID a lot of our appointments and our check-ups with speech therapy have been put on hold. While this is extremely agitating I am happy to say we have kept safe and avoided Covid coming into our house as we live with my parents (Alicia's Nana and Grandad).

Life has been busy and I don't think the Doctors and Nurses understand how thankful I will always be for looking after my baby. It isn't easy when you have a child with a medical condition. The most frustrating thing is when you get a letter to read they are on a waiting list. I understand it is not their fault but when you are worried about your child this upsets you. I wish our voices would be listened to a lot more.