

Liam's Story

WITH 17q12 DUPLICATION

Our son, Liam, is almost 4 and has the duplication. Liam was a totally normal pregnancy and the first few months were right on par. When Liam was about 6 months I started noticing he didn't "feel" right, I brought it up to his pediatrician, and they concluded I was clearly crazy and he was "fine".

However, I persisted and then Liam started missing milestones. Finally, the doctors took notice and sent us to a neurologist. Liam was diagnosed with severe hypotonia (low muscle tone). Then came trying to find out why, a blood test and a wait told us he had the 17q12 duplication.

Unfortunately having a diagnosis only meant more questions with no answers.

We scrounged online and through medical papers and still didn't really learn anything new. However, we did stumble on a conference and decided to attend. It was so wonderful meeting the other families. Liam's hypotonia has been managed with physical therapy. His other symptoms are speech delay, mild cognitive delay, global development delay, obsessive compulsive disorder, anxiety, iron deficiency, and sensory seeking behaviors.

He sees his speech therapist every week, and an occupational therapist once a month. Liam started school last year and we saw so much improvement with his speech.

"Liam is the happiest kiddo and lights up our life. We are thankful we came across the conference and were really able to get involved"

