

“Javier ("Javi") is a sweet, happy, loving boy. He LOVES cars and trucks, and always has a car or truck in hand. He loves to drive them around and line them up. Javi is shy, especially in new surroundings and situations, but is extremely observant. He understands what is going on around him and often wants to participate. He just doesn't always know how to, despite his hard work and willingness to try.

We were typical first-time parents, completely in love with our beautiful baby boy and also full of questions and concerns:

- Why does he have such difficulty eating, as a newborn, at 3 months, and 6 months, and still at 9 months?
- At 18 months old, should he have any words other than babbles? Should he be able to do simple hand signs after 8 months of trying to teach him?
- At 20 months old, should he be able to use a straw? Why can't he figure out how to sit down in a little chair or crawl down the stairs, after showing him over and over and over? Do toddlers fall down constantly for no reason? Is falling more than 50 times a day typical? Why can't we get him to eat more? Why does he choke and not eat at all when he is tired or sick?
- At 2 years old, should he be able to hug us or hold our hands? Should he be able to mimic sounds, like animals, and movements, like sticking out his tongue?

According to everyone we asked, these things were “normal”. They would say all children are different and reach milestones at different times, some children are late talkers and toddlers fall a lot. Eventually people started to point out differences they saw in our son. Why does he walk that way, with his arms up, and why are his hands always in that unique position? We had actually never noticed it, as that was just Javi. Even after starting therapies through early intervention, we saw limited progress. Should an almost 3 year old have more than 5 words after a year of speech therapy? It wasn't until recently, when we received his diagnosis, of a *SATB2* mutation that makes the gene non-functional, that all of this started to make sense.

Javi loved being held but when we'd show him how to put his arms around us for a hug, his arms would immediately fall once we let go. We worked on this for months and months, placing his arms around us and holding them there. Finally when Javi was 2 1/2, he gently rested one little arm around his mother's neck for the first time, giving his first hug. That is how Javi learns, with a tremendous amount of repetition using hand-over-hand instruction and by breaking things down in to small, simple steps and concepts. While some children can learn movement, words, or concepts after practicing them several times, it can take hundreds or even thousands of times before Javi can learn movements, such as sitting down in a chair, making sounds, such as 'ee', or learning a concept, such as matching.

We still have endless questions, many of which there may never be answers for, but it is a great relief to finally have some answers. Despite the little that is known about this extremely rare syndrome, we have learned much more about Javi's condition based on the work of Dr. Zarate and from the other parents of children with *SATB2* mutations and deletions. The other children with *SATB2*-associated syndrome and their parents are an inspiration to us and we learn so much from them every day.

Javi has made tremendous progress over the past 6 months now that we have a better understanding of his condition and how he learns. We are very fortunate that he has an experienced, pro-active, and caring special education pre-school teacher, as well as therapists who understand his needs. We are extremely hopeful for Javi's future, but also realistic. There will be many challenges ahead, but we are confident there will also be great accomplishments that we will celebrate and cherish, such as that first hug."

*Javi's mom*