A mother's struggle to come to terms with her son's diagnosis of autism caused by a rare genetic condition—and her gradual acceptance of his differences.

Sliding down, climbing up

by ***Lisa Kadane*** ⟆ *illustrations by* ***Byron Eggenschwiler***

"Knock, knock, Mommy."

"Who's there?" I ask.

"Banana," Bennett says.

"Banana who?"

"Knock, knock."

"Who's there?"

"Banana," he says again, his smile revealing a gap where his front tooth once was.

"Banana who?"

"Knock, knock."

"Who's there?"

"Orange!" he yells, barely able to contain his excitement.

"Orange who?"

"Orange you glad I didn't say banana!"

My son Bennett is five-and-a-half years old and has just killed his first knock-knock joke. His dad and big sister join me in laughing and giving him high-fives. A couple of minutes later, not wanting the party to end, he says loudly, "I always burp and I always fart!" We all dissolve into giggles again. "That's funny!" he declares, clearly pleased with his ability to be the ham.

Not every family meal plays out this way. Some evenings Bennett zones out and stares out the window into the backyard, or gobbles his pasta and meat sauce in silence. On Easter Sunday he ate two bites of his salmon and excused himself. Five minutes later he came running out of the bathroom, pants around his ankles, and yelled, "Mommy! Wipe my bum!" in front of a table seating seven adults, all, fortunately, family. We laughed it off—

kids do the craziest things, right? But Bennett does odd things a lot, and life with our son can be a roller coaster, especially out in public. One minute we're getting a kick out of his eccentricities, the next minute we're wishing all the people in the restaurant would stop staring at our son, who just belched loudly after finger-painting ketchup all over the white tablecloth.

Bennett has autism caused by a genetic condition, and it's fair to say he marches to the beat of his own bongo player.

When I was in Grade 8 I bought a T-shirt that pictured a line of identical penguins walking in step, but the bird on the end was wearing an Aloha shirt and a lei and doing a little dance. He stood out from his penguin peers. The text on the shirt read, "Rugged Individualist." At the time I thought I aspired to be that bird, but the truth is I just wanted to fit in with the cool kids in class. And so, against my better judgment, I wore blue eye shadow, Esprit outfits and rhinestone jewelry, and I had my hair cut into the 'do-du-jour. I wanted to be noticed, sure, but I didn't want to be *conspicuous*. For as much as society says it encourages individualism, those who are too different are ostracized. Black-and-white emperor penguins flock together, in other words. They're frightened by the little gentoo penguin who's wearing the loud shirt and dancing all crazy.

My son Bennett dances all crazy and, as his mom, I kind of have to follow the beat of his bongo player, too, or I'd go crazy.

Being the parent of a kid with special needs makes me think about degrees of difference—and acceptance, because it too is graduated. Just as soon as I get into a routine that resembles some kind of normalcy, the roller coaster begins another twisted descent. When the days involve endless tantrums and every night features a night terror, I declare, "I never signed up for this" and become angry that my kid isn't like other children. When you meet typical kids you see only what your child can't do and it makes you sad, and jealous. But then the crazy train rolls into a valley, life evens out again and you start moving along the path toward acceptance. "I can do this!" you think. His behaviour improves, his skills advance. "He's making so much progress!" you tell those closest to you.

I suffer through the hard parts, feeling alone and resentful, but I so delight in the small gains, like how Bennett can finally pronounce the letter 'L.' And I pretty much throw a party for the big milestones, like potty training.

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But still we worry about Bennett's future. Will he be unemployed and living in our basement when he's 30? And sometimes, because that spectre is so frightening and depressing, all we can do is joke about it (humour really is the best medicine): Maybe Bennett will open a breakfast place that only serves peanut-butter toast (his favourite meal). Or maybe he'll get a job as a Calgary tour guide, given his eerie GPS-like ability to point out local landmarks, including the Calgary Zoo, the Bow building, both rivers, Canada Olympic Park, the Peace Bridge, the Calgary Tower and Alberta Children's Hospital, where we spend a lot of time.

It was at Children's where we found out the cause of Bennett's autism: a genetic condition called 18q- (pronounced 18 Q Minus). He's missing a small piece in the long arm of one of his 18th chromosomes. This means he has been slow to hit milestones, like walking and talking; it also means he has a difficult time playing and interacting with peers. Since science hasn't advanced enough to fix Bennett's missing genes, we treat the autism.

Bennett attends kindergarten through Renfrew Educational Services, a not-for-profit organization that runs an integrated special-needs preschool and grades program in Calgary. He also receives Specialized Autism Services through Renfrew, and we have a parade of aides and therapists who come to our home and into Bennett's classroom to help him reach his speech, fine-motor, gross-motor, behavioural and play goals. It's his third year at Renfrew and his progress has been remarkable. We are delighted he'll be continuing into Grade 1 there this fall.

To an outsider, Bennett initially comes across as a typical kindergartner: he's cute, likes being silly, loves climbing and sliding at the playground (although he's more awkward than most kids) and relishes tormenting his big sister. In many ways he doesn't fit people's idea of what autism, or a genetic condition, looks like. But if that same outsider spends any amount of time observing Bennett, they'll see he is different beyond his speech and motor delays. He laughs at things that aren't funny. He taps his hands spastically when he gets excited or agitated, a practice known as "stimming" in autism speak. His favourite books have to be arranged just so on the coffee table or he'll start to cry. And unlike most five-year-olds, a trip to Toys "R" Us is a non-event for Bennett because he doesn't really care about Hot Wheels or Lego bricks or Transformers. My son would rather flip through books, watch the same episode of *Blues Clues* on continuous

play, jump on the trampoline to stimulate his body or just cuddle with me on the couch.

"I'm just tired, Mommy. I need to rest," he'll say when the confusion of the modern world and our expectations of him threaten to overwhelm him.

"It's okay, Bennett," I'll say. "Let's just look out the window for a while."

I can't remember exactly when my husband and I stopped reading *What to Expect: The Toddler Years*. Bennett was about 15 months old and still not walking or talking. The book said by that age most toddlers are walking and saying things like "Mama" and "Dada" and "nana" (banana), plus stacking up to 12 blocks (overachievers!). Bennett was still struggling to palm Cheerios into his mouth, refused to even try and use a spoon and had no interest in stacking anything. He had only been crawling for three months after scooting on his bum for two. I closed the book and rolled over in bed to face Blake. "Do you think we should be worried?" I asked.

"I don't know," he answered. But I think we both knew something was wrong.

Only one child in 40,000 in North America is born with 18q- (by comparison, Down syndrome, the most common chromosomal disorder, occurs in one out of 800 to 1,000 babies). When we received the diagnosis we registered Bennett with the Chromosome 18 Registry & Research Society. According to their records, he's one of four people with 18q- in Alberta; there are only 20 registered cases in Canada. Autism is much more common, affecting one in about 150 children worldwide, though the American organization Autism Speaks pegs its incidence at closer to one in 50. It's more prevalent in boys.

When my husband and I decided to try for a second child and play what we used to half-jokingly refer to as "genetic roulette" (because we were, at ages 36 and 35, respectively, somewhat "older"), autism was a remote "what if?" at best; a genetic condition wasn't on our radar—didn't the ultrasound at the end of the first trimester scan for that kind of thing? I just wanted to have another healthy baby, like my daughter had been. "Healthy," I realize now, is mom-to-be-speak for "normal." Of course I wanted a baby free of disease. But here's what else I wanted: a child without behaviour and learning problems. I didn't want to be that mom—the one with the special-needs kid.

And anyway, after having a healthy baby, who really thinks she's going to have a second child that makes

her want to stop reading developmental-milestone books (or, worse, makes her fantasize about trading that second child in for a "normal" one)? In those early years, it was easier to shelve the books and hope Bennett would catch up, than entertain the thought that something was wrong. "He'll catch up. All children develop at their own pace," our family doctor assured me. "He's just fine," averred well-meaning relatives. I wanted to believe them but I knew—as perhaps only a mother does—that something was wrong with my beautiful boy. It would be three and a half more years until we found out what.

Not long after setting down the book, Bennett began to walk—on his tiptoes. My autism radar sounded in alarm, but the fact that he made eye contact and was affectionate helped push those dreaded thoughts aside. One day when he was sitting on my lap I gently grasped his foot and tried to flex it into the "heels-down" position. It wouldn't budge; it felt as though his ankle was fused into a pointe stance, and Blake and I became accustomed to hearing regular "Maybe Bennett has a future as a ballet dancer" remarks. To which Blake would sarcastically reply, "It's what every father dreams for his son."

An orthopedic surgeon at Children's diagnosed Bennett with "idiopathic toe walking," a term for unexplained toe walking, most likely due to short heel cords. To fix this problem Bennett was scheduled for heel cord lengthening surgery and referred to a neurologist to make sure there wasn't something else going on, like cerebral palsy. A brain and spinal MRI looked normal, but revealed delayed myelination. This means the fatty coating around his nerve cells isn't as thick as it should be, which can slow down the speed at which information travels through the nervous system.

This new information was a huge red flag. It got me thinking back to three hours after Bennett's birth, when the on-call pediatrician came into my room at Peter Lougheed Centre to tell me that my infant son had a hypospadias. Basically, his urethra hadn't developed all the way to the end of his penis. Don't be alarmed, the doctor said, it's one of the most common birth defects in boys (and was surgically corrected when Bennett was 11 months old). It can also be a genetic marker in children with chromosomal abnormalities. Taken together with his short heel cords and less insulated nerve cells (two other potential genetic markers), plus his developmental delays, I asked for a referral to a genetics specialist.

In the meantime, we paid for a private speech/language assessment, and his subsequent "severe delay" diagnosis qualified Bennett for a government-funded spot at Renfrew. After a year at Renfrew, Bennett was talking more but still lagged far behind his peers in fine-motor and play skills. Social interactions baffled him. The Renfrew psychologist suggested the school screen him for autism.

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We got the autism diagnosis six months before the call from the genetics department at Children's. The autism fit but it kind of didn't, so everything made a lot more sense when we learned about his 18q- genetic condition. Knowing helped. Bennett's genetic mutation is de novo, meaning it occurred spontaneously and wasn't passed down from either parent, so I no longer wondered if I had somehow caused Bennett's difference when I was pregnant with him.

At the same time, the diagnosis was so…final. The media is filled with stories of children who miraculously "recover" from autism after going on a gluten-free diet, or taking a bunch of supplements, or doing hyperbaric oxygen therapy. There aren't any similar stories about kids who "recover" from genetic conditions after receiving stem-cell therapy. We could try all the weird diets and take advantage of as much early intervention as possible, but nothing was going to change Bennett's DNA.

As a family we had so much to be hopeful about—Bennett was getting the early intervention that's so important, and making progress—and yet, I still often felt (and sometimes still feel) sad. Crushingly sad, and so alone.

Bennett's differences can be an awkward thing to talk about with friends and even relatives. People can't relate; they don't know what to say, or what questions to ask, and so there is silence. Life with a special-needs child seems to exist in two separate silos: there's a world of school and therapy where you talk about everything but nothing is personal; and then there's a normal world, where you try to put on a brave face and present the best of it to those you're closest with, without burdening them with the ugly bits. This is a hard twin highway to navigate. You're living a normal life…except you're not. You're happy for what you have—two beautiful children, a loving spouse, a nice house, a career you're passionate about and a lifetime of amazing travel memories—but despondent over what will forever elude you: a son with a complete genetic makeup, and everything that signifies. There's a name for what I'm going through: "chronic sorrow."

It's a state of constant grieving that families experience when they are coming to terms with a diagnosis. The child hasn't died, but their dream of a "normal" life has.

Basically, what happens when you have a child with special needs is this: your illusion of the perfect life with perfect children is shattered. There won't be hockey games or guitar lessons or lots of friends calling for play dates. Your kids won't be the playmates you'd hoped—our reality has forced our daughter to be mature beyond her years, but it's also taught her empathy, and Bennett couldn't ask for a better, more loving big sister. (At age eight, she already defends her brother to peers: "He's not weird. He has autism.") Beyond his childhood we worry there may not be university or marriage or adult friendships for him. Or a career or independence. The not knowing is scary, and sad. So I have been mourning the experiences I thought I would share with my son. But I try to keep it in perspective, too. Like autism, all of life is a spectrum of good, so-so, bad and "it could always be worse." Half the time we are in a good groove and I'm a proud, and thankful, mama.

This sense of gratitude was hammered home in San Antonio, Tex., on the final night of the Chromosome 18 Registry & Research Society's annual conference, where everyone gathered for a farewell dinner. Blake and I decided to attend the conference last July, with Bennett, to learn more about his genetic condition and to meet families with kids with 18q-. It's comforting to know we are not alone in this… and yet we are. Because every child with 18q- is missing slightly different pieces, and lengths, of Chromosome 18, and because each individual expresses his or her genes differently, no other kid who shares his condition is exactly like Bennett. Some are non-verbal and still in diapers, while the keynote speaker for the gala had just graduated from the Air Force Academy, providing a glimmer of hope for Blake and me.

The conference was an eye-opener. We met 18q- kids with vision and hearing problems, as well as a little girl born with a cleft palate, heart defect and a host of other health and developmental difficulties. We sat through a seminar that discussed the range of health problems often associated with Bennett's condition, from seizures and epilepsy to thyroid issues and growth-hormone deficiency. The fact that Bennett's only "problem" is high-functioning autism made us feel lucky. "Your son looks so normal," a couple of moms attending the conference told me, as though Bennett gave *them* hope.

The truth is, some children with Chromosome 18 conditions have severe physical and mental handicaps. A teenage girl with Trisomy 18 (three copies of Chromosome 18 instead of two) sat in her wheelchair across from me at our table, staring vacantly. Her mom was with her, accompanied by a full-time, live-in aide who fed the girl through a feeding tube while the mom chatted with us. It was depressing to watch.

Seeing the severity of this young woman's disability tested the limits of my comfort with difference. Do I address the girl with questions about her name and age that she obviously can't answer? Or do I pretend she doesn't exist and speak directly to her mother? Am I keeping my face a neutral mask, so the mom can't read my pity over what appears to me to be her sad plight? (Is my reaction to this girl and her mother the same reaction that moms with normal kids have to me and to Bennett?)

I was overcome with guilt and shame for feeling such discomfort, and marvelled at the utterly human and self-interested thought in my head: *I am grateful Bennett is not this girl, that he can feed himself solid food and tell people his name and age when they ask.* "It could always be so much worse," was all I could think as I ate my chicken and rice pilaf with little appetite.

The next time I'm about to throw a little pity party, I told myself, I should remember this mom and her daughter. Or, I should recall the man we met on the first night of the conference—the one whose seven-year-old non-verbal son had a habit of pooping in his diaper and then playing with it. *Buck up, Kadane, it could always be so much worse.*

And so I work hard to celebrate Bennett's milestones, like learning to put his shoes on and hopping on one foot and singing "Bingo," because for Bennett, little triumphs that go unnoticed in other children are huge; they are small steps on his slower, windier road to (fingers crossed) independence. We don't know where the road will go, so we try to enjoy the bumpier ride. I am mindful to embrace and enjoy the things we can do together, like skiing down the bunny hill in Fernie and visiting a dude ranch in Arizona.

I love Bennett fiercely, and I am trying to accept my little gentoo for who he is, and appreciate what he has brought to our life. More patience (most days). More love (every day). And way more crazy dancing.

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