My Daughter Is Not A Syndrome

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I always had trouble memorizing those pediatric genetic disorders. They seemed so obscure, so idiosyncratic. You had to key in on buzzwords like "cherry red macula" or "rocker bottom feet." These days, there are online memorization aids that try to help students by juxtaposing tough concepts with outlandish visual cues. One of them is called *Picmonic*. I can still picture the man with a pickaxe in his knee for Niemann-Pick disease, or Taylor Swift wearing a sash for Tay-Sachs disease. Ridiculous images, but they worked. In spite of my perceived struggles, I honored my pediatrics rotation.

But good memorization skills of pediatric genetic disorders don't arm you against the terrifying uncertainty that comes with the moment you find out your own unborn baby might have something out of the ordinary. I still remember the text I got from my wife as I was leaving my internal medicine sub-internship for the day. She had gone in earlier for a repeat ultrasound. I had Zoomed into the twenty-week ultrasound four weeks before, while working in the Family Medicine clinic. We found out the gender, and they told us everything looked fine, but they could only visualize about seventy percent of what they needed. They scheduled a twenty-four-week ultrasound to get the rest. It seemed routine. My wife's sister went with her.

Still, I felt a tiny bit of trepidation when I texted "How did the ultrasound go?" on my way to my car.

Her response sent my mind racing. "It went. We can talk when you get home."

I immediately called her. The details were confusing. They had trouble visualizing several structures again. But some of the findings were concerning. The ultrasound report mentioned clenched fists with overlapping fingers and rocker bottom feet. The OB had brought up the

possibility of trisomy 18. Most infants with trisomy 18 do not survive their first year of life. Those who do often live with profound disabilities and require immense levels of care. The future suddenly felt terrifying and uncertain. Why was this happening?

When I got home, I fixated on the ultrasound report. I reread the findings over and over. Frontal bossing. Clenched fists with overlapping fingers. Rocker bottom feet. No cardiac anomalies. Size at the first percentile.

But some things didn't quite add up. Frontal bossing isn't a classic feature of trisomy 18. Severe cardiac anomalies usually are. Only one of the feet had actually been visualized. And the first percentile size was wildly different from the prior ultrasound, which had measured closer to the thirtieth percentile. That earlier scan had also described the heart and other major structures as reassuring.

We clung to hope as we scheduled a visit with a maternal-fetal medicine specialist and a level II ultrasound. Rachel had a Natera blood test drawn at the twenty-four week visit, and we anxiously awaited the results. We were sitting in the waiting room just after the repeat ultrasound when the nurse came out to get us.

"Your Natera is low risk for everything," she said.

Rachel and I broke down in each other’s arms.

Relief, but also confusion. If the screening was reassuring, what exactly were we seeing on the ultrasounds?

The MFM physician was reassuring about several things. The heart looked normal. The brain looked normal.

"Those are two pretty important organs," he said.

But he still felt something was off. Something he couldn't quite pin down. We proceeded with an amniocentesis and sent a chromosomal microarray. Then we waited. Weeks later, the results came back: normal.

With each subsequent ultrasound, some of the earlier findings became less clear. The feet that had been described as "rocker bottom" began to look more like high arches. Measurements fluctuated. But the fists remained tightly clenched. They never opened. And the frontal bossing still didn't quite make sense. Our MFM recommended full exome sequencing on the amniotic sample. We submitted a request for prior authorization.

Denied.

We appealed.

Denied again.

Out of pocket the test would cost more than three thousand dollars.

By that point we were already two months from the due date. Even if we paid for the test, the results might not come back before delivery. And if they did, it wasn't clear what we would do differently.

Would knowing change anything?

Medical school teaches you the science of genetic disease. It teaches you inheritance patterns, molecular pathways, and diagnostic criteria. It teaches you how to recognize a syndrome when a question stem lists the right buzzwords. But none of that prepares you for the strange limbo of uncertainty. For weeks, we lived somewhere between reassurance and anxiety. Every ultrasound was a new data point. Every measurement slightly shifted the probabilities. But probabilities are an uncomfortable language when the outcome in question is your own child.

Leading up to the birth, our MFM's leading hypothesis was arthrogryposis, a condition that causes contractures and rigid joints. Because of the uncertainty around the diagnosis, we spent a lot of time thinking about where we wanted to deliver. In the end, we chose a small hospital in a town north of where we live. Our MFM and OB team agreed this was reasonable and gave us clearance to deliver there. The delivery was fast and smooth. I'll never forget the moment I laid eyes on our Naomi, seconds after she was delivered. She was our little girl. Working out her first cry like any newborn. Her welcome call to the world.

Immediately, I could see the frontal bossing from the ultrasound was real. And it suddenly made sense why her hands wouldn't open. She had complete syndactyly of her hands and feet. I looked to the doctor for reassurance. I don't remember his exact words, but as he wrapped her in a towel and handed her to me, he was confident she wasn't in any acute danger. She pinked up immediately. She moved all extremities. She cried. Her heart rate and sounds were normal. I held her in my arms like she was the only thing in the world, my beautiful daughter. Then I slowly laid her on her mother's chest.

A few minutes later, as mom and baby rested peacefully, my mind swirled. Naomi was okay right now, but it was evident she had a genetic condition. One I had never learned in all my hours

studying for exams and boards. I did what one does in 2025 when you need a quick answer, and pulled out my phone and opened ChatGPT.

All I typed was "newborn has frontal bossing and syndactyly". The result stared back at me in bolded text: Apert syndrome. "What on earth is Apert syndrome?" I thought to myself.

A quick Google image search was unequivocal. Naomi had Apert syndrome. I read through the basics, unsure of what this actually meant. In some ways, it felt like I was back in the classroom, learning one of those hard-to-memorize, idiosyncratic genetic diseases. *Apert syndrome is a condition occurring in roughly 1 out of every 65,000 to 100,000 live births. It is caused by an uninherited, spontaneous gametic mutation in the FGFR2 gene that causes the eponymously named FGFR2 protein to become constitutively active. During development, this leads to inappropriate and/or premature fusion of the musculoskeletal components of the head and distal extremities.* Scientifically, it made sense. But Naomi was not a science exhibit. She was my daughter.

We reached out to doctors at the University immediately. The next two weeks were spent going to a variety of appointments that provided answers, but also more questions. A CT scan of her head confirmed she had craniosynostosis. This is the technical term for saying some of the bones of her skull were prematurely fused. Normally, your skull bones are separated at birth like tectonic plates on a globe. As is characteristic of Apert syndrome, Naomi's coronal sutures were fused. We were informed that there are a few different surgical options to correct this in infancy. We needed to decide on a plan.

Throughout my academic career, I have always loved scanning the literature on various topics I happen to be interested in at the time. As a child, I remember my dad—a primary care physician—sitting on the couch in the evenings, reading *JAMA*, *NEJM*, or some other journal. He was always a stickler for "the literature." If a clinical question came up, he would no doubt tie his reasoning to the outcome of some study or another.

A few days after Naomi's diagnosis, he sent me a paper.

It was a recent study from a group at Harvard looking at surgical outcomes in children with Apert syndrome. The paper discussed different approaches to treating craniosynostosis in infancy, including a technique called an endoscopic strip craniectomy. The senior author was a neurosurgeon named Mark Proctor.

My dad suggested I email him.

At first, I laughed at the idea. Why would a Harvard neurosurgeon take heed to an email from a medical student in Iowa about a baby he had never met? Surgeons like that must get dozens of messages every week. He already likely worked 18 hours a day. Neurosurgeons are amongst the busiest, most overworked people in the hospital. Who was I to contact him and ask for advice about my daughter from halfway across the country?

But I had nothing to lose.

So I sent the email.

I told him Naomi's diagnosis and inquired about his take on surgical options. I asked a few questions about surgical timing and whether the endoscopic approach described in the paper might be appropriate in her case. Then I went to bed not expecting much of anything.

When I woke up the next morning, there was a reply waiting in my inbox. It had been sent at 5:07 AM.

"Hi Andrew. Congratulations on the birth of your new baby. You ask some excellent questions. I wonder if it would be easiest to speak and discuss some of these issues directly. I would be happy to talk. Thanks. Mark Proctor."

I read the email twice just to make sure I wasn't misunderstanding it.

We spoke on the phone that afternoon.

He was calm, thoughtful, and generous with his time. He explained the different surgical options, what the risks were, and what outcomes they had seen over the years. Apert syndrome is remarkably rare, but his team had treated many children with the condition. Boston Children's Hospital is one of the few places in the world that runs clinical trials specifically for Apert syndrome. Families travel there from across the country and sometimes from other continents.

The physicians in Iowa had been thoughtful and supportive from the beginning. But the reality of rare disease is that experience tends to concentrate in a few places. Centers that see more cases accumulate a kind of practical knowledge that comes from treating the condition again and again.

By the end of the conversation, the decision felt clear. Naomi needed surgery, and if possible, we wanted someone who had done this many times before.

To my surprise, Dr. Proctor told us they could work Naomi into the surgical schedule.

Within a few days, his team had coordinated the logistics. What had started as a speculative email suddenly became a concrete plan.

In the meantime, life did not slow down.

Twelve days after Naomi was born, I found myself sitting on the couch with my laptop open, trying to finish my residency applications. In one browser tab was ERAS. In another was a journal article about craniosynostosis. In Microsoft Outlook, I was drafting another email to Dr. Proctor. Naomi slept beside us while Rachel and I adapted to our new responsibilities.

It felt surreal.

Eight years of MD-PhD training had prepared me to analyze molecular pathways and experimental data. It had not prepared me to schedule craniofacial surgery for my newborn daughter while proofreading my personal statement. Then again, perhaps it had. Because against all odds, that’s what I was doing.

At the same time, I was still in the middle of clinical rotations. The morning after one of Naomi's early appointments, I was back in the hospital on my urology rotation, scrubbing into the OR for a robotic TURP. Medicine as a vocation demanded its usual sacrifices. But in the background, a different version of medicine had suddenly become personal.

Rare diseases occupy a strange place in medical education. They are memorable precisely because they are unusual. They are often presented as collections of distinctive findings that help students practice pattern recognition. Cherry red macula. Rocker bottom feet. Maple syrup urine. When you are studying for exams, these diseases can feel almost abstract. Something distant. The kind of thing you memorize because it might show up on a board question. But someone, somewhere, is living with every one of those conditions.

Now it was our turn.

For a few weeks things felt stable. Naomi was eating well and gaining weight. We finalized the surgical plan in Boston and began preparing for the trip.

Then during the fall, something else began to worry us.

Naomi's breathing sounded noisy. At first it was subtle, a kind of soft snoring sound when she slept. But over time, it became more noticeable. Babies with Apert syndrome often have mid-face hypoplasia, meaning the middle portion of their face develops differently. That can narrow the upper airway and make breathing more difficult.

Rachel is a CRNA, so airway anatomy is something she understands very well. The more we read about it, the more uneasy we became.

We scheduled an appointment with pediatric ENT at Iowa.

What was supposed to be a routine evaluation turned into an overnight admission. The ENT physicians were concerned about the degree of airway obstruction they were observing. Naomi did well through the night, but they recommended surgical intervention sooner rather than later.

About a week later, she underwent two procedures: an inferior turbinate reduction and a supraglottoplasty to help open her airway.

Watching your infant go into surgery is an emotional experience. As physicians, we are trained to think in terms of likely outcomes and risk-benefit ratios. As a parent, you are faced with the reality that your child's life is in someone else's hands. You must face the decision of handing your child to a surgical team and trusting that they will bring her safely back. Rachel and I had been through this once before. When our older daughter, Helena, was just two weeks old, she was admitted to Stead Family Children's Hospital for pyloric stenosis. We had watched her go into surgery then too, tiny and brand new to the world, and waited for them to bring her back to us. Now here we were again, in the same hospital, with our second daughter.

Fortunately, everything went smoothly. Naomi recovered well and was soon back home with us.

A sleep study performed afterward showed that she also had obstructive sleep apnea. She was prescribed supplemental oxygen at night, given via a small nasal cannula that delivers a gentle quarter liter of oxygen per minute while she rests.

By the time winter arrived, it was finally time for the trip to Boston.

Helena left with my mother the morning Rachel and I departed for Boston. Watching her go was its own kind of hard. She was two and a half years old, old enough to sense that something significant was happening, young enough not to fully understand why her parents were leaving. We had two children now, and we could only be in one place at a time.

We first had an outpatient visit with a Harvard ENT surgeon. Believe it or not, he was a medical school classmate of Naomi's ENT surgeon in Iowa. And his wife was a pediatric anesthesiologist at Boston Children's. He was happy with Naomi's airway status and recommended no other interventions for the time being.

A few days later, we met Dr. Proctor.

He was just as I had imagined him: kind, calm, and confident – the kind of doctor who would respond to a cold email and offer guidance to a family 1200 miles away. He pulled up the imaging sent from Iowa, did a quick physical exam, and reassured us Naomi was in good hands.

The morning of the surgery, we went to the pre-op suite and were assigned a room. Soon enough, the anesthesia team came to take Naomi to surgery. It was already the second time in her young life that she would have a significant operation. I comforted Rachel as she fought back tears while Naomi was carried to the OR.

The surgery itself went quickly. The procedure, an endoscopic strip craniectomy, aims to correct the premature fusion of her coronal sutures. This will allow her skull to expand more normally as her brain grows. When Naomi got back, the impact on her skull shape was already apparent. She rested peacefully in her crib. Rachel and I squeezed next to each other on a one-person mattress. With just 10 days to Christmas, a *Home Alone* marathon played on TV.

Naomi was discharged the next day. My mother flew Helena to Boston two days later. We took her to the ice cream museum and the Boston Aquarium. She soaked up the sights, giddy and unbothered, completely unaware of what the previous few days had held. Watching her, I felt a sense of calm I hadn’t felt in a long time.

Over the next week, we had several appointments to get an orthotic helmet, which will help mold Naomi's skull into the appropriate shape as she grows throughout her first year of life. We also had a clinic visit with an orthopedic hand surgeon. He examined Naomi and told us his team will reach out when the timing is ideal, probably when she is closer to 1 year old.

It was now just a few days before Christmas. We celebrated the holiday with Rachel's parents in Delaware, before flying home to Iowa right before New Year's Eve. Naomi's first healthcare excursion to Boston was over.

There will be more surgeries in the years ahead. Children with Apert syndrome often require additional craniofacial procedures as they grow.

In the months since Naomi's diagnosis, Rachel has found online communities of other parents navigating the same road. Facebook groups where people ask questions, share updates, and offer the kind of practical knowledge that doesn't exist in a textbook. She has had phone calls with some of them. One family she connected with was also planning to see Dr. Proctor. Their child was their first, and they were learning what it meant to be new parents at the same time they were learning what it meant to have a child with Apert syndrome. Rachel talked with them for a long time and shared our story.

I have done my own searching. One evening, I came across a TikTok of a man with Apert syndrome in the Philippines, around forty years old, who looked as though he'd never had any surgeries at all. His hands looked just like Naomi's – complete syndactyly, the fingers fully fused. In the video, he was just going about his day, holding a bowl and spoon and eating his

dinner in the best way he could, getting done what he needed to do. There was nothing performative about it. He was just living life as he knew it.

Rachel found a video of a little girl, three or four years old, whose mother posts videos on Facebook from time to time. In the clip, the girl was putting on her socks. Her hands had been partially corrected through surgery, but it was still slow, deliberate work. She didn't ask for help. She just kept at it, with a look of total concentration and fierce determination on her face. I watched it more than once.

These were not statistics. They were not outcomes or data. They were people.

I sometimes think back to those *Picmonic* images from my pediatrics rotation. The pickaxe in the knee. Taylor Swift wearing a sash. They were clever memory tricks, useful for passing exams.

But what they represent is only the smallest fraction of the story.

Behind every rare disease name is a person. A family. A life that unfolds in ways no flashcard could ever capture.

Before Naomi was born, I had never even heard of Apert syndrome. From my perspective, it was just another obscure entry in the long catalogue of genetic conditions students may or may not encounter on their journey through medicine. Something distant and improbable.

Now it is something entirely different.

But my daughter is not a syndrome.

She is a living, breathing person. She has her own personality and tendencies. She loves to be held and listen to her teddy bear song. If she's not being held, the next thing she prefers is her swing, where she can slowly doze off with a bottle of milk. Her 2.5-year-old sibling, Helena, has affectionately bestowed upon her the nickname "Noh-me." She also has a canine brother Barkley, who thinks tummy time means it's time to throw the ball.

One thing will be certain. While Naomi has parents who work in healthcare, we don't want her to feel defined by a healthcare condition. We will love her for the beautiful daughter she is, regardless of how her genetics affect her physiology. Medical school taught me to recognize syndromes. Naomi has taught me to look beyond the buzzwords and find the person behind them.

Apert syndrome may be part of her story, but it is not who she is.