

Finalist, Fiction

Undiagnosed
By Elizabeth Lewis

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What's Wrong With Her?

“What’s wrong with her?”

Of course, the boy who lived down the street has to be walking by our house right now. Here I am, standing at the end of the driveway with a freshly delivered newspaper in hand. My dad is backing our station wagon into the street and my sister in full tantrum mode in the backseat. At eight years old and tall for her age, anyone can tell she’s too old for this kind of toddler-style display.

“Is she okay? What’s going on?”

He’s still talking. I almost never talk to the other kids in the neighborhood, so I should be excited. Between going to private school and staying busy with way too many after-school activities, I rarely see anyone from the neighborhood. I don’t even know their names. I’m sure they think I’m the weird girl from the oversized house that keeps its curtains drawn. These things stand out around here.

“She’s fine,” I say. I look down and walk around the car, and the boy shrugs and passes by with a final glance at Chloe.

My dad unlocks the doors and I hop in the backseat next to her. Her body snaps like a mousetrap, leaning forward and then crashing back to the gray cloth seats. She’s jumping as best she can with her seatbelt on, which can’t be easy. She bites the meaty part of her left hand between her thumb and pointer finger, and smacks the inside of the door with her other hand. She’s definitely pissed. Was she mad that it was nearly lunchtime and there was no sign of her

usual Saturday fast food? That I'd slept in again and caused the late start? She keeps smacking away, my dad turns on the oldies radio station, and I settle in and watch Neighbor Guy walking down the sidewalk as we pass him.

None of this is new to me, although that doesn't mean I don't notice. Of course I do. It's just that this is our everyday. It's what I know.

Sibling Status

My sister has disabilities. I know, I know – that could mean almost anything. When people hear “disabilities” they think easily digestible categories like Down syndrome or maybe CP. Blindness is It seems straightforward enough.

Chloe's situation is much more complicated. Her disabilities don't have a name yet, although I'm convinced I'll find it one day. I've been able to rattle off the list ever since she was born: *Chloe had auditory and visual impairments, but she still has some hearing in one ear. She was born premature and with a small heart problem, but it's okay. She has colobomas in her eyes, so her pupils are wide ovals instead of circles and she can't see as much as we can.* [At this point, I place my flat hand against both eyebrows to show her range of vision. People appreciate the demonstration.] *She has OCD tendencies and can't talk. She knows a little sign language.* Basically, Chloe has a bunch of different impairments and a few medical issues, and we don't know why.

My dad calls this our elevator speech, but it's definitely a mouthful. People get uncomfortable hearing so much medical information in a casual conversation. I know it catches them off guard to hear it coming from me, since I'm supposed to be talking about things like the World Cup or my dance team or boys.

We recently learned that Chloe is undiagnosed. Well, okay, I guess we knew that. But we didn't know there was a word for it until the geneticist told us. Undiagnosed. Part of me had always felt ashamed that we didn't know what Chloe *had*. When people asked "What's wrong with her?" I could only remember that I was supposed to say "Nothing is *wrong*, she just has disabilities!"

But that wasn't what they wanted to hear. They weren't looking for identity politics, just an easy term to explain the slanted facial features we pretended weren't part of a broader syndrome, her strange refusal to run or jump like other kids, the awkward gait or slightly too long arms. No, this wasn't just a case of needing eyeglasses, hearing aids, and a little speech therapy. There was more to Chloe. We just didn't know that we didn't know it.

I'm 14 and my sister was born when I was six. Adults sometimes say things like "So you don't really remember anything before she came along." It's almost a question, but not quite. I think they're trying to make themselves feel better or find a silver lining in what they think is a tragedy. I always wonder if they don't remember anything from childhood. I have lots of memories from before Chloe was born. Too many, perhaps. Does that make me weird?

My first memory is from around age two. I woke up one night in my crib and was sure there was someone outside of my window, yet I was too small to get away. I cried and cried until I mom came and scooped me up, bringing me into my parents' bed with the old cat who always scratched me.

I remember when my Grandpa died one spring. I took a clear, plastic umbrella with drawings of rainbows and clouds and gleamingly jagged suns, and walked around the front yard in the rain singing songs my mom used to play. Peter, Paul, and Mary. Simon and Garfunkel. Those were always on in the house back then.

I remember driving across the country to live in Oregon for a while. I remember the day I threw up all over my favorite red patent leather shoes at preschool, and how I haven't had apple juice since. I remember going to a restaurant on the coast and eating chowder, which I didn't like at all. I remember pretending the algae growing in our fishpond out back was a school of microscopic fish. My fish.

People who know about Chloe treat me differently. Not my friends, but definitely adults. They act almost like I have superpowers. They say I'm "precocious" and that I talk like a grownup. They remark how I must have to help out so much at home, and ask how my parents are doing. Friends' parents and teachers offer to let me stay with them if my folks need to drive to see specialists, therapists, or other experts out of town. They give silent, knowing smiles and act like there's nothing else to say. But I see them looking at me, just a little extra – a little longer.

Labels

Undiagnosed. It's a fancy way of saying we don't know why Chloe is the way she is. Different. Different is okay, my parents remind me. But she's not normal like my friends and me, I tell them. Don't say normal, they say – a little too quickly. You mean she's not *typical*. Typical is the word we use.

There are lots of rules about what we can and can't say. Some of them are obvious (like the R-word, which is never to be uttered), but a lot of it is more confusing. I don't know why I'm not allowed to say "normal." It comes up in conversation all the time – normal kids, lives, behavior. *That's not* normal. *Act* normal. People say it all the time. But my parents tell me it's a bad word. If it's so forbidden, why do I hear it everywhere?

My mom usually takes me to the Family Disability Resource Center when she goes. It's an organization for a bunch of other parents who have kids – some grown – with different disabilities. It's usually all moms. They go and tell their stories, drink coffee, talk about different therapists, which teachers or school principals they hate the most. They also answer calls, meet with parents, and give trainings on things like disability rights and special ed. Everyone there has a child like Chloe, my parents tell me, although we all know they don't. I've never met anyone like my sister.

Sometimes the moms at the Center try to talk to me about being a sibling. They tell me that I'm doing such a great job and I'm so grown-up. They reassure me that I can't "catch" what Chloe has. (Apparently, the books on disability and families say siblings are worry about this, but it's never occurred to me. Adults are *obsessed* about this one.) They ask if I'm staying busy with school and activities. I get a lot of knowing smiles and Hallmark-style commentary, plus the occasional "If you ever want to talk to my *other* son about your experience I'm sure he's willing." Sure, I'd love to talk to a total stranger about having a sister who's undiagnosed. What teenage girl could say no to that offer?

But I guess they're trying. When I was 10, my mom sent me to a support group for siblings. She promised ice cream afterward, so I begrudgingly agreed to give it a try. Every single kid in that room had a brother or sister with autism. *Every single one!* And not the new kind of autism you see on TV shows like *Parenthood* and hear about on the news, but the old kind – the serious kind that doesn't show up in one in sixty-something kids. And you know what? I was jealous.

Sure, the siblings seemed totally miserable and uncomfortable at the support group, just like me, but they had a common ground that I didn't even know I was missing. They could

relate. And then, of course, I went and ruined it all during my intro: *My sister has visual and auditory impairments, and colobomas in her eyes, and possibly intellectual disabilities but that might be due to communication deficits...* Their eyes glazed over and I became, without a doubt, the outcast in the room.

Over ice cream afterward, I told my mom I wished Chloe had autism. That was my last trip to the support group.

But it's everywhere, this obsession with a diagnosis. A name. It reminds me of school. We all have labels in high school. Jocks, stoners, cheerleaders, art kids, theater people, band nerds. My yearbook tells everyone that I made honor roll both semesters, was on student council, wrote for the school paper, played the trumpet, and ran track. Those are my labels. They give an idea of who I hung out with, how my days were structured, what types of activities interested me and kept me busy.

It's not that different than the posters displayed at the Center of some of the staff members' kids. Big, proud professionally photographed portraits of the different kids, complete with brief biographies and then, in bold letters, the name of their diagnosis and a short description. My mom said they're from the website. I guess the posters are supposed to make the space seem more personal.

Brianna's family has been active with the Family Disability Resource Center since she was a baby. They all love attending our annual conference and special events, and meeting other families. Brianna enjoys swimming, music, and spending time with her pets. She is seven and has lots of friends at school.

Diagnosis: *Spina Bifida.*

Spina Bifida (SB) is a birth defect in which the spinal column does not close completely. It begins in the womb and is a lifelong condition. It is associated with mobility impairments, as well as multiple other conditions. Mothers should begin taking folic acid supplements at least three months prior to pregnancy to help prevent SB.

I ask my mom why Brianna's mother didn't just take vitamins. She tells me it's more complicated than that and we shouldn't always believe what we read. I wonder what would we put on a poster for my sister?

Chloe likes swimming, French fries, and watching Sesame Street on repeat. She loves animals and enjoys swinging on her backyard swing set for hours at a time.

Diagnosis: Unknown. We've never met anyone who looks, acts, or has the same set of impairments and conditions as Chloe. If you come across anyone who does, please contact us via email or telephone. We don't know how often her undiagnosed diagnosis happens or what it means for the future. A doctor once told my mom it was because she drank wine before she knew she was pregnant, and my parents think there was chemical dumping near a park we used to visit before Chloe was born. Maybe that's it? But why would she be the only one?

Registry

After years of tossing around different possible diagnoses, my parents finally took Chloe in for genetic testing last summer. They even got insurance to pay for it, which had been a big worry. I was away at camp when they did it, but they said it was just a blood draw. A few months later, they met with a geneticist and learned that Chloe's disabilities were caused by a genetic mutation. They even know which gene. She has a mutation on the KPC2T gene. And it's *de novo*, which means they don't know why it happened and there's no reason to think it could happen again in the family. But that was as far as they got. The mutation she has doesn't have a name and no one else is known to have it. Just Chloe.

My parents added Chloe's genetic testing results to a registry, which is basically just big storage place for people's genetic testing results. It's coordinated by the main teaching hospital in our state and falls under a bigger government-run program for undiagnosed disorders. In theory, we'll be contacted if someone else shows up with my sister's same genetic mutation. Good old KPC2T.

I know my mom is waiting for the phone to ring. Do they even contact us that way? Who's they? I picture a geneticist showing up at the front door, like the Publisher's Clearinghouse guys on TV. Or what if it's an email that goes to our spam folder and we never find it? A letter that gets lost in the mail? An automated text message or voicemail that we fail to check? My parents are awful about checking their phones.

My mom talks about the genetic registry daily now. She says that I should be thankful to live in a time with this technology. She reminds me that 10 years ago this kind of genetic testing would have cost millions of dollars and been completely out of reach. Now, it's a few thousand bucks and insurance covered most of it. I haven't seen any real payoff yet, though.

I talk about the registry with teachers and other adults sometimes, but to be honest I can only explain it in abstract terms. The elevator speech again. I have no idea how it works or what it looks like. I picture a small man with a mustache turning a handle on a cage, like at a bingo tournament. KPC2T. KPC2T. KPC2T. Chloe's genetic profile is displayed on an overhead projector, and the little man just pulls out number after number from his endlessly turning stash. "No match." "No match." No match yet.

My mom is convinced there must be others out there. *There's no such thing as a singular case!* she declares. Can Chloe really be the only one? Are we the only family like ours? My dad isn't so sure. While my mom spends her nights with a glass of wine combing over Google for any possible lead about Chloe's genetic mutation, my dad focuses more on our troubles with the local school system. They want to say that Chloe has intellectual or developmental disabilities, and this makes my dad furious. "It's a lack of communication, not her brain!" he yells. His voice is low and thick, especially after a drink or two, and carries throughout the house. It's one of the few voices that Chloe can still hear.

He gets so angry on the subject that I have to leave the room. He starts thundering about the ignorant, heartless people at school and the principle who's just a big nobody and wants to feel important. He says he hopes they get cancer or their husbands leave them, that their lives fall apart. I've never heard him talk like this. I'm supposed to hate the school people – I can tell from my parents– but I don't even know them. It seems strange that adults would be out to get my little sister, but my parents are convinced. We spend a lot of time talking about this. Sometimes I help them organize binders of special ed case law for when things get more serious with Chloe. I might be the only student in my grade who knows what case law means, much less *least-restrictive environment*, *reasonable accommodations*, and *low-incidence population*. I'm still not clear on *due process*, but it's a term that's been coming up more and more.

Hospital

My first memory of my sister probably isn't real.

After my mom went into labor, my parents left me at home with my grandmother. A day or two later, my dad came by the house to bring me to the hospital. They walked me down to the nursery so that I could meet my sister.

“She looks different than the other babies in the nursery,” they cautioned. “She has a cleft lip, so there's a gap in the middle of her mouth.” Armed with this knowledge, I was able to spot her immediately amidst the sea of wriggling, seemingly identical little pink newborns in the room. I loved that Chloe was different. She stood out and she was mine.

In my memory, my mom notices some stickers on Chloe's bassinet that the other babies don't have. She sends my dad off to ask the nurses, and, for me, this is when everything began. It is the moment that leads in a direct line to the present. But I don't think it actually happened.

How could it? I now know that Chloe had to be in the NICU after birth, which is where they send the newborns with health problems. I wasn't allowed inside, since kids were too germmy. So how did I make this memory?

The doctors didn't think Chloe would survive. My parents didn't name her, since someone told my mom it would be easier to lose a child if it hadn't yet been named. I have a vivid – and very real – memory of explaining all of this to a shocked teaching assistant on the playground at my school that week.

I remember the house filling up with bouquets. Everyone sent flowers, but not many people came by. My sister was premature, but not by much. My mom says she knew something was “wrong” with the pregnancy. I think she just means different than with me. She mentions that her belly got bigger earlier in her pregnancy with Chloe than with me, and a family member said that's a surefire sign. Her doctor said it was nothing unusual, but she still holds onto it.

“Don't all pregnant women have big bellies?” I asked the first time she shared this with me. “This was different,” she responded.

Even eight years later, she still spits with anger – literally – when she talks about the doctor who delivered Chloe. Her regular doctor was on vacation that night. This guy made it clear he wanted to be anywhere but the hospital. She never forgot it. I wonder if she believes things would have turned out differently had he been available. I've heard similar stories from the other moms at the Center.

After Chloe's birth, we traveled to hospitals and specialists all around the country to find answers. My dad says this is why I do so well in school now. I learned the language of doctors when I was young. He tells me there are lots of studies showing that young kids who learn more words end up smarter and do better on tests, and I definitely heard more words than regular kids.

I would go into the offices with my family and be the cute little girl who asked grown-up questions and helped with her disabled sister. The doctors loved me, the therapists got a kick out of asking me to be their special helper. I probably shouldn't have been there at all, but what else could my parents do?

When I was in second grade, I learned about cells and atoms at a summer science camp and became completely convinced that I could see them. I told my parents that my eyes were so strong I could make out cells moving through the air. They were just paranoid enough that they mentioned this to my sister's ophthalmologist, one of the best around. I remember hopping into his exam chair at the massive hospital with the adjoining hotel and floating walkways suspended between buildings. He did a quick exam, told me I was seeing nothing more than tears and goo in my eyes, and proudly proclaimed that my eyes were so good I would never need glasses.

Three years later, I couldn't see the board in school and had to get my first pair. My eyes have gotten worse by the year ever since.

Screen Time

It seems silly to say that we found out Chloe was undiagnosed, but that's how I see it. My parents don't seem to care about this at all. Ever since they got the genetic testing last year, they've been obsessed with finding out what Chloe "has." My dad hopes a diagnosis can help us make her better, although disabilities don't have cures. They aren't sicknesses.

Anyway, we all secretly know mystery cures are a thing of the past. We tried them all. Special mouthpieces to help her talk (glorified retainers, if you ask me), facilitated communication, keyboard attachments for the home computer, speech therapies, psychologists, medications, different teachers, pictograms, weighted blankets, therapeutic horseback riding.

You name it, we gave it a shot. Some of the experiments were more fun than others, but none were free, easy, or convenient. I don't complain, though. What's the point?

When we learned Chloe was undiagnosed and not just a mutt (a term I'm definitely not allowed to use), I realized that there were millions of other kids like her and families like ours. Not just like us in terms of Chloe's box-shaped face, flat nose, refusal to play with kids her age, or curious habit of dipping all food in her drink before eating it. There were other families who also didn't know. Who were undiagnosed. Everybody else *didn't* have Down syndrome or autism or Angelman syndrome or whatever else my moms' friends' kids had. We weren't lacking something. Science just hadn't caught up with us yet.

I'd always felt embarrassed that we didn't have a diagnosis for Chloe. I saw people's eyes glaze over as I described her laundry list of impairments and conditions. I worried that I owed them an easy label so they could smile, nod, and not have to listen to me rattle off the elevator speech again. Instead, they would give me these looks of intrigued, mildly bored pity, and I'd respond by being so optimistic and chipper about the whole thing that that my parents were always reminding me that I didn't have to "hold it all in." But isn't growing up about holding things in? Feelings, knowledge, jealousy, anger? About learning to control ourselves and put on a good public face? I often wished my parents could hold it all in a little more. Or at least not share every little thing about Chloe with me. I've watched my friends with their families and I know it's weird for my parents to treat me like an equal partner. *But I'm 14 now, I have to remind myself. I'm not a kid anymore.*

My parents weren't as excited as I was about the undiagnosis angle. They didn't think it was a revelation at all and had no interest in connecting with undiagnosed families, unless they could find a kid like Chloe. My mom was getting increasingly obsessed with the computer and

would run to it as soon as Chloe went to sleep at night. Facebook groups, listservs, message boards, parent blogs – she read it all. The doctors always told us not to do internet searches about Chloe’s disability or health, saying it would scare us and give us bad information, but my parents never listened. Not my mom, at least. She just parked herself in front of the glowing desktop, often forgetting to turn on an actual light. Hunched over, legal pad and multi-colored pens to the right, wine glass to the left. I usually left her alone like that.

She’d been talking about it more and more recently. It was hard to discuss anything else at dinner, between my mom’s internet investigations and Chloe’s increasingly epic tantrums – her chair slamming up and down so hard that the lights flickered. I hardly ever got to talk about school or friends anymore with my parents. We seemed to be having more and more fights, although I couldn’t tell you about what. Sometimes my parents remarked that I’d started acting like a teenager.

“So I found this mom in the U.K.,” Mom was telling us one night over tacos. “Her sons background is similar to Chloe’s, but we emailed each other pictures and they don’t look anything alike.”

Code words like *background, condition, symptoms, experience, history*. So many attempts to explain Chloe, who was sitting directly across from me. I listened, looking down and hoping the meal would end soon so I could go to my room and start my homework. We always looked down these days. No one made eye contact at home anymore. I hadn’t noticed until a friend at school pointed out that I was doing it, too.

“And she thinks she knows another mom in Canada whose kid has similar facial features!” My mom is still talking.

“Oh, okay,” I managed to say. I wasn’t going to get my hopes up. I spent most of my childhood getting excited about whatever new therapy that was going to make Chloe like the rest of us, and nothing panned out. As a kid, it does something when you hear enough grownups say they’re going to make something happen and then nothing changes. Chloe never talked. We never really learned anything, but just moved onto the next possibility. At some point, I started to get exhausted by it all. I started saving my excitement for things I could control: grades, improving my running times, that sort of thing.

“Can I get up?” I asked. “I have an algebra test tomorrow.”

DIY

I’m in my room doing my social studies homework, memorizing the branches of the government.

“James!” My mom yells my father’s name suddenly from the office. Even though Chloe can’t hear, we never yell after bedtime. It’s rude.

“James! Come here now!” She’s yelling again, perhaps more forcefully than usual. I hear my dad make his way upstairs to the office. He takes his time. My mom’s kind of a yeller. It’s hard to know when to hurry.

“My God!” I hear him exclaim. What is going on out there? Uninvited, I open my door and walk down the hall. My parents are huddled over the computer. My mom is on Facebook. This is why they’re yelling?

“That’s her,” my dad keeps saying. “Dear god, that’s Chloe!”

They don’t even hear me. I peek between their shoulders and there she is. It’s not Chloe, but I have to pause. I’ve never mistaken my sister for anyone else in her entire life. She’s never

looked like anyone else. But there it is on the screen: another girl, probably a few years younger, longer hair, slimmer eyebrows, but otherwise the resemblance is striking, right down to the coloring. That could be Chloe.

My mom fills us in. It's been a week since my mom first mentioned swapping photos with the woman in Britain, who then put her in touch with the mom in Canada. She and my mom started Facebook messaging each other – questions, descriptions, medical details. Sharing the personal stuff doctors have to keep secret, the things I could recite in my sleep.

“This is how the other mom did it, too!” my mom is saying. “This is how she found her daughter's diagnosis! She told me about the message boards, how she met a mom over her blog and that mom put her in touch with a woman in Ireland...”

I'm can't staring at the screen. Who is that? KPC2T. KPC2T.

“It's all so DIY,” my mom says. “That's what the mom said and she was *right!* Do-it-yourself, I swear. After all these years, all the doctors and tests! Facebook! Can you believe it?”

I can't stop looking at the picture. That's Chloe. It's not, but it is.

We're suspended in this moment of change. The air is gone from the room. My mom is still in front of the screen, her face inches from the monitor as she clicks through photos of the girl. My father leans over her balancing his weight on the back of the old wooden chair, just staring at the screen.

“I never imagined...” he starts to say, more a whisper to himself than anything, but then he trails off.

KPC2T. KPC2T. Undiagnosed, but not on our own. We are not the anomaly. Not the only ones. There is at least one other Chloe. And that means there are more.