“We Weren’t Given a Manual” So We Wrote One: An Irish Traveller Family Navigates Rare Genetics, Systemic Barriers, and Unseen Labour

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DOI: 10.33178/SMJ.2026.10

Abstract

This is a special edition feature submission based on the interview of the family of the only documented paediatric congenital titin myopathy with central, bulbar dysfunction in Ireland.

The Interview

Tucked away in the modest stillness of a north Cork hilltop estate stands a home that runs like a clinic but beats like a heart. Its porcelain sterility—white tiles, white doors, white light—is softened by the brightness of children’s artwork, communion portraits taped beside medical guidelines, and photos of three children growing older in every glittery frame.

The quiet hum of the washing machine is overlaid by the familiar flick and chatter of TikTok videos, as 12-year-old Lily O’Connors* scrolls through her phone at the dining table, her bubblegum-pink nail polish catching the light. Her wheelchair is angled just so, her curled-up fingers resting gently on the table while her mother, Laura O’Connors*, switches on a toy fan to dry the polish. She looks at Lily with an indulgent smile, the kind shaped by routine but tinged with the quiet ache of watching a child grow up both too slowly and far too fast.

“You know, there was no guidebook for this,” Laura says. “We weren’t given a manual like. We were given a child, and another child with worse problems, and we had to figure everything else out from there.”

She doesn’t say it with resentment. Her voice carries the certainty of someone who has spent nearly two decades learning a language spoken in complex medical care and gut instinct. Two of her children, Bobby* and Lily, live with congenital titin myopathy—a rare genetic disorder made even more exceptional with Lily’s ultra-rare presentation of central, bulbar dysfunction – the only ever documented case in Ireland. It took years, multiple hospitals, and Laura’s refusal to give up before they were diagnosed.

She still remembers holding Bobby for the first time. He was too light. His limbs curled inward in ways that made her stomach twist. “They told me it was brittle bones,” she says, “but brittle bones didn’t explain the floppiness. Or the broken shoulder, even though they did a c-section after he stopped moving. Or the way he couldn’t suck. Or how scared I was to touch him too hard.”

Bobby quickly became what doctors called a "mystery." "He had tubes in him all the time. X-rays, scopes, blood tests. I was getting sick of it." Eventually, Laura hit a breaking point. "I said, no more. I wanted answers, but I didn't want to see him suffer just for the sake of another test. Every time I brought him in, he caught something and we'd end up back in hospital."

It wasn't until Lily was born, five years after Bobby, that the family finally got a diagnosis. A muscle biopsy confirmed she had titin myopathy, and retrospective testing revealed the same for Bobby.

"I always thought I'd have at least four," Laura says. "My brothers and sisters all have big families—eight kids, five kids. But that choice was more or less taken from me." After conceiving her third child, she couldn't risk putting another baby through that uncertainty. "With the third, I just didn't know what way it would go." When Lily was delivered, the neonatal team was waiting. "They took her straight away. And I just knew. She had it too."

If anything, finding out the second time was harder. "Now I had an idea of what might be ahead. I was very emotional. But you deal with it. You get on with it."

And she did. For years, Laura chased information. "I know it's rare. But still, there was nothing. At first, I thought Bobby might be the only child in Ireland with it." She only learned otherwise during a conversation with Lily's doctors. "They mentioned two others—one older than Bobby, one younger. That was it. No details. The doctors were always fantastic and knowing that we weren't completely alone was a comfort but that they couldn't tell us anything more because of confidentiality, we were back to square one."

The feeling of isolation lingered on still. "That's why I'm doing this, hopefully others read this article and reach out," she says of the interview. "There were no support groups. I was afraid to let anyone babysit. I didn't want to do anything wrong. Everything felt medical."

"You don't get to clock out. Even when you're sick, even when you're exhausted — you still have to show up."

Eventually, small things began to change. For Bobby, a second-hand tricycle turned out to be the breakthrough no piece of medical equipment could offer. "He'd sit on it and push with his legs. Then he started standing up, sitting down. It built his strength more than any stander." At seven and a half, he took his first steps. "He never wanted a wheelchair—his mind was fine, and he didn't want to be treated differently."

Now 17, Bobby drives, cooks, ties his own laces. "Two years ago, he couldn't even put on his shoes," Laura says. "Now, he's doing more than his older brother sometimes." She credits much of his progress to their oldest child, Jimmy*, who was born without the condition. "He treated Bobby like a little brother, not like someone sick. He'd sneak him sips of his juice or a bite of his ice cream and we'd have heart attacks worried about Bobby aspirating, but he was fine. We had Bobby wrapped in cotton wool, but Jimmy didn't. And I think that helped more than anything."

Lily, though, needs more support. Her condition has always been more severe, and as she grows, so do the demands. At home, Laura runs what she calls a 24-hour unit. Her day starts before sunrise. "I get up and give Lily her [proton pump inhibitor] for reflux. You wait thirty minutes, then start the feed. Hook her up, flush the line, monitor the pump. That's just the first hour."

Then comes dressing, washing, brushing her teeth—all done in Lily's room. "She can spit now, thank God. I rinse her mouth with a syringe of water. It's slower, but it works."

This has been Laura's life for over a decade. "We used to do eight feeds a day. Every three hours, day and night. An hour each time. That's just what it was." Even sleep offered no pause. "I'd be up every two hours, then still have to get the boys to school. Even if I was sick myself, there was no such thing as rest, but it was God's will and having faith that this is what I was meant to do is really important for me."

Now that Lily can eat some solids, there's a bit more room to breathe. "If she eats something decent like gravy and carrots, I'll skip a feed. But mostly she just tastes it, not eats it."

Care doesn't end at the front door. Leaving the house involves accessibility checks, weather, gear, transport. "We don't go many places. Most places just aren't made for wheelchairs."

Housing adds another layer of difficulty. "We waited ten years for a house we could actually live in; we were in a caravan before. No lift, no rails, no pulleys or straps. We can't afford any. I carried her up and down stairs in this house for a decade. Now she's too big. Her dad does the lifting now."

The council offered bungalows, but they were too far from anyone they knew. "I need people around. Family, friends. Someone to grab tea with or just check in. Even if you can't go with them, it's nice to be invited."

Access to suitable housing, especially for Traveller families with disabled children, remains limited. And yet, the cost of housing, physically, emotionally, medically—continues to fall on families like Laura's.

As Lily nears thirteen, the family is adjusting again. "Puberty changes everything. Her care, her mood, her routine. She wants more independence. I'm trying to give it, but it's hard. I'm learning with her."

That shifting space is what led Laura back to the classroom. She's now enrolled in a course in Social Inclusion and Diversity at MTU.

"I always knew I could do more," she says. "I loved school when I was put in a proper class, but by then it was too late. I was behind and I left school very young." As a Traveller child, Laura was placed in a classroom where "You didn't really have to learn. You didn't even have to try. I never got a real chance. I can read but it's hard to fill out forms."

Now, she's reclaiming it. "The course is once a week, but we study on Mondays too. It's for Traveller women like me. We do projects. I get help if I need it. It's for me."

"I want to work. I don't know exactly what yet, but I want to do something... I always knew I could do more."

Her plans aren't fixed, but her direction is clear. "When Lily starts secondary school, the days will be longer. I'll have time. I want to work—maybe home healthcare, maybe something else. I've got loads of experience, that's for sure!"

Laura doesn't see herself as extraordinary. She simply sees herself as a mother doing what needs to be done. "You just get on with it because you are their mother and they are my babies," she says. But by now, the meaning of "getting on with it" is clear. "Getting on with it" means caregiving without rest, learning without help, advocating without backup, and still finding the energy to plan for more.

None of it has been easy. Much of it has been unfair. But what Laura has built, with patience, faith and resilience, is a future for both her children and herself.

This isn't a story about inspiration. It's a story about visibility. About what happens when you refuse to disappear and instead, choose to repair severed connections and reach out for more.

It's also a reminder of what humanism in medicine requires. To see beyond the diagnosis and understand the whole story of a patient, and of the people who stand beside them. Laura's story invites us to listen more closely, to recognize caregiving as expertise, and to honor the lives that shape care long before, and long after, the clinic doors close.

Sometimes, the most essential thing we can offer as physicians—beyond a treatment plan—is the decision to witness a patient's life fully, to honour its complexities and quiet resilience, and to remain present in a way that helps them move toward the future they hope for, on their own terms.

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