Lucy Bolt, Scarlett Bolt

info@europeanlung.org
@EuropeanLung

Patient voice

A process of years: coming to terms with primary ciliary dyskinesia

"This baby has low oxygen saturation"
"There must be something wrong with the equipment, that's a perfectly healthy baby"

The first words spoken about my daughter between the medical student and the paediatrician in the delivery room were to prove prophetic of the confusion surrounding her health over the next 8 years.

Having been sent to the maternity ward with my perfect, full-term, first child, in a bubble of unreality, I didn't understand why the midwives were unhappy with her breathing or keen for her to be reviewed. By the evening, a doctor was calmly telling me they just wanted to take my daughter to run a couple of tests and I should try to get a bit of sleep. That was 10 pm.

When I woke at 5 am, my daughter wasn't there. I asked where my baby was and was taken to the high-dependency unit. I can't describe the feeling of walking into the dim, hushed environment of neonatal high dependency and being shown my baby being given oxygen, with a nasogastric tube and cannula. I didn't understand what I was seeing or why – and, unfortunately, the why was confounding everyone else too.

Scarlett was treated for congenital pneumonia. I was told this was "as rare as hens' teeth" in a full-term baby, but after 2 weeks, her pneumonia was "cured" and we were free to go home. It was to be another 8 years before we met a doctor who joined the dots right back to this unusual, but textbook, primary ciliary dyskinesia (PCD) neonate experience.

Scarlett was a "mucousy child" and I was regularly telling her to cough to clear her chest. By 2 years, I was fairly certain she had a problem with her hearing, and there followed 2 years of hearing tests and talk of glue ear. At 4 years of age, just before starting school, she had bilateral grommets inserted.

From the day the grommets were inserted, Scarlett's hearing improved markedly, followed by her speech. Unfortunately, the payoff was that her ears permanently oozed thick yellow discharge. We were to and from our excellent, but bamboozled, general practitioners. Scarlett was referred to ear, nose and throat (ENT) and hearing services repeatedly, and was regularly on oral and aural antibiotics.

I would discuss with other parents how tired our children were after a day at school, although it always seemed that Scarlett's habit of walking in and promptly falling asleep on the sofa with shoes and coat still on was at the extreme. This permanent state of exhaustion led to difficult years of what felt like constant angry, explosive behaviour from Scarlett. I was frustrated with my daughter and it felt like I was always cross with her. I now feel a lot of guilt about my response.

At age 8, we were back at ENT for one of our regular visits. We saw a different doctor. I went through my usual spiel about her ears and regularly telling her to cough to clear her chest. I think for the first time, this latter point was heard. The doctor asked something that no-one else had asked before, "Did Scarlett have any respiratory distress at birth?"

The early signs of PCD are like many other common childhood complaints. However, respiratory distress in an otherwise healthy, full-term baby is unusual.

During the 2019 ERS International Congress, a speaker impressed on the audience the importance

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Lucy Bolt shares her experiences of raising a child with PCD, and the long and difficult journey to a diagnosis https://bit.ly/37bdYXX

of asking about neonatal history. The earlier someone asks the question, the sooner they can get a diagnosis, manage the condition and minimise the impact on education, social development, and physical and mental health.

The doctor had talked about removing the grommets and trying hearing aids instead, so when he asked me if anyone had ever mentioned cilia to me, I was too busy worrying about how Scarlett would feel about having hearing aids. He then explained that he would plan joint surgery with a respiratory colleague and that Scarlett would have a bronchoscopy while having the grommets removed.

By the time of her surgery 3 months later, I had extensively consulted with Dr Google. I scoured the far corners of the internet reading everything about cilia. I came across PCD and as I read about it, I read about Scarlett. I read a description of my daughter and her health from her birth to now.

After the surgery, the doctors explained that in joining the dots, they had stopped the downward trajectory of my daughter's lung health. They explained that Scarlett's lungs had been full of secretions that they had cleared and that they would be treating her from today on the assumption that she had PCD. Scarlett would be referred to the national diagnostic centre in Leicester to go through the formal diagnostic procedure. Scarlett's lung function was less than 50%. A computed tomography scan soon afterwards showed extensive bronchiectasis in the right lung and a collapsed middle lobe.

That was the day we were introduced to respiratory physiotherapy. That first day, we enjoyed the novelty of the spirometer exercises and bubble pep. Over the last 2 years, this has developed into a normal routine of twice-daily sessions with her oscillating positive expiratory pressive device and saline nebuliser, with rounds of huff coughing to manually clear the secretions from her lungs, as well as twice-daily sinus rinses and azithromycin prophylaxis. She has had 6-monthly elective admissions for proactive intravenous antibiotics – and more recently, her first reactive admission, which was an unwelcome milestone.

Now aged 10, Scarlett is a bundle of tween hormones and physio is, as I am told daily, "boring and not fair". At the time of writing we are in a relatively compliant phase. Scarlett has always done her physio, but for a long time this was only after tears, tantrums and arguments. The disconnect between my fears about her health and her frustration at having to commit so much of her time to something when "nobody else has to" has, at times, felt like it was going to permanently damage our relationship. Some days are still more challenging than others, but on the whole, it's going ok – and I keep forgetting that at 10 years old, it's amazing how stoically she gets on with it and how maturely she can talk about her worries.

I am writing this in Queen's Medical Centre, Nottingham, where Scarlett is in surgery having a portacath fitted. For irrational, emotional reasons,

Scarlett enjoying her new trampoline.

I don't want this to be happening; silly though it sounds, it just makes everything seem all too permanent. Until 2 years ago, I thought I had a healthy, albeit tired and snotty, little girl. Scarlett thinks it's a great idea as she won't have to have a line put in her arm each time she comes in – and she's right of course.

Coming "to terms" is a process of years (I'm not sure we're quite there yet). Most non-medics have no reason to know what cilia are, never mind the impact when they don't do their job properly. Scarlett will be going to high school this year, and we will need to start the process of educating her teachers and classmates about PCD again. It's a tiring process and one that Scarlett will deal with for the rest of her life.

Scarlett, in her own words

There are some good things about PCD, like my mum always said we couldn't have a trampoline until I was diagnosed but then she got a massive one because it's good for helping me keep fit and to clear my lungs. Usually, I swim nearly every day but because of lockdown, I am using the trampoline every day and doing land training on Zoom with my synchro swim team, Aquastars. I have great nurses and doctors who explain things really well to me and I like seeing them – the nurses on my ward are really fun. I wish I didn't have to do physio every day; it's really repetitive and takes up loads of time. I'm just getting used to having a portacath because I just had it put in as lockdown started; it's a bit weird but it's working well and now I won't have to have a long line put in my arm every time I go into hospital.