

Two girls, one genetic 'blip' and a story of hope and courage

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At her holiday apartment, Peggy Heron waited, heart pounding, to meet her daughter's genetic 'twin' for the first time.

Peggy's daughter Lottie, now 15, was born with a genetic disorder so rare that, until two years ago, they thought no other child in the world had it.

Then, out of the blue, the Herons were contacted by an Australian family who said their daughter had the same disorder. And last month the two families - Peggy and her husband Paul, both 42, and Michelle Lawson, 37, and Luke Smith, 34, as well as their children - finally met.

And when four-year-old Georgia entered the room, Peggy burst into tears.

"It felt as if I'd gone back in time and seen my daughter at the same age," she says.

Georgia's piercing blue eyes with black rings round the irises were identical to Lottie's at that age - the parents from both families are dark-eyed. The girls also shared the same slightly stiff walk and similar facial features, including a wide nasal bridge.

"It was uncanny how alike they were," says Peggy.

"I sobbed - stunned at finally meeting the little girl who, although unrelated to us by blood, was so like our daughter when she was small. All the agonies we went through, that family would also have to face. It was a very emotional moment."

One in every 200 babies is born with a rare chromosome disorder - that's many more than the one in 800 with Down's syndrome, the best-known chromosome disorder.



Two of a kind: Georgia, four and Lottie, 15 share a rare genetic disorder

When Lottie was born, doctors noticed she had abnormal reflexes and tested her for Down's, but no chromosomal abnormalities showed up at that point.

However, as she grew, Lottie was slower to reach developmental milestones than other babies her age. She was also awkward physically - holding her hands in fists, while her arms and body were stiff.

Doctors were baffled by her condition. Some diagnosed developmental delay and said it could have been caused by cerebral palsy; others said it was too early to tell if there was anything wrong.

"At a routine appointment with a paediatrician when Lottie was nine months old, he said we should prepare ourselves for the worst," recalls Peggy. "It was devastating."

Two brain scans - at ten and 18 months - came back normal. But Lottie didn't sit up until she was 15 months old and didn't crawl, though she eventually began to walk aged nearly two. She didn't string two words together until age four.

Lottie was having physiotherapy three times a week and occupational therapy to help her develop through play. The family tried to put doubts about her slow development to the back of their minds.

"It was easier at this stage - Lottie was petite and pretty and looked "normal", says Peggy. "Children develop at different rates and we believed her mild physical problems could be overcome, although her doctors didn't ever say that this would be the case."

When Lottie was two, and Peggy three months pregnant with their second child, the Herons decided to visit a geneticist, fearing that if Lottie's condition was hereditary, their new baby might be affected.

"I was cooking dinner, waiting for Paul to come home from work, when the geneticist phoned with our results," says Peggy.

She said it was good news - the cause of Lottie's problem was a chromosome 'deletion', but her parents were fine, and their second child was unlikely to be like Lottie.

Each of us is born with 23 pairs of chromosomes. These are made from a chemical called DNA, the genetic material found within living cells containing information determining their growth, development and function.

Chromosomes are the building blocks of life. Sometimes they can be missing a piece, or contain an extra bit, resulting in a disorder.

Why this happens is not clear.

However, it is thought that these kinds of genetic errors cause between 30 and 50 per cent of cases of moderate to severe learning disabilities.

Although blood tests done after Lottie's birth had come back normal, the tests themselves are standard, first line tests which look only for more obvious chromosomal abnormalities, such as the extra chromosome in Down's syndrome.

The tests done by the geneticists were more sophisticated. Indeed, major advances in genetic testing technology over the past 15 years have led to increasing numbers of people with unexplained learning disabilities being diagnosed with chromosomal disorders.

The news of Lottie's condition was devastating, says Peggy. "It hit me then that Lottie wouldn't "grow out of it". When I told Paul he turned away from me, but too late to hide his tears. It was heartbreaking, the first time I'd ever seen my husband cry."

Paul works as a bond dealer in the financial world and Peggy was Lottie's main carer. She struggled to cope as Lottie's behaviour became increasingly challenging.

"At the mums-and-toddlers group, I kept jumping up to stop Lottie pulling other children's hair or biting them," says Peggy.

"No one said anything but I felt so conscious of other mums' stares that I stopped going."

At home, Lottie had violent screaming tantrums. Her strength was so great that, before she turned three, she managed to pull out the cutlery drawer and hurl it across the room. She also ripped the washing machine door off - while it was running.

"After our baby George's arrival when Lottie was three, I regularly climbed into his playpen with him to protect us from Lottie's attacks," said Peggy. "I felt a failure as a mum."

But a referral to a learning disability behavioural specialist nurse made a tremendous difference. He taught Peggy 'positive reinforcement' techniques, explaining that the family needed to react positively to good behaviour and reduce the amount of attention Lottie received for the bad.

Although Lottie's destructive behaviour improved, her hyperactive and impulsive behaviour continued. She had no sense of danger and would run out in front of cars and put her hands on hot kettles. She also has a very high pain threshold.

When Lottie was four a paediatrician diagnosed hyperactivity and prescribed Ritalin. It was life-transforming.

"After the first dose of Ritalin, she responded to me calling her name," says Peggy. "For the first time ever, she stopped what she was doing and looked at me."

Lottie took the drug for the next nine years, stopping at 13 when it exacerbated her hormonal mood swings with the onset of puberty. Fortunately, by this point Lottie had matured: she was less impulsive and able to cope better with her own behaviour.

But still the family thought their daughter was the only child in the world with her particular problems.

The geneticist had given Peggy the phone number of a support group - Unique - for parents of children with rare chromosomal disorders.

Unique keeps a computerised database detailing the effects of specific rare chromosomal disorders among members, which can be used to link families affected by similar conditions - but there was no match for Lottie.

Then, in 2005, came the surprise phone call from Australia. The woman's next words took Peggy's breath away.

"My daughter has just been diagnosed with the same problem your daughter has," said Michelle, sounding close to tears.

"Just tell me one thing - can Lottie walk?"

"Not only does Lottie walk, she can also ski and ride a bike!" Peggy replied.

At this Michelle burst into tears. It meant there was hope for her daughter's future.

"I'll never, ever be able to express the relief, elation and joy that I felt after talking to Peggy," she says.

"When Peggy said her daughter could walk, I sobbed, relieved."

For the next year, the two mothers exchanged regular e-mails and photos. And, about a year later, the Herons went on holiday to Australia and the two families met up at an apartment the Herons had rented in Port Douglas, Queensland.

"It was an extraordinary moment," says Peggy, "like being in a film. My eyes met Michelle's and we hugged. Then we all sat down and talked. I could see Michelle watching Lottie, fascinated. Then the room went quiet as Georgia walked in - she'd been playing in another room. My sobs pierced the silence.

"The children hit it off. Lottie loves babies and played with Ethan, Georgia's baby brother. Georgia adored our son, George. Like a surreal role reversal, George played older sibling to Georgia, instead of being younger sibling to Lottie. She snuggled up to him as he read her a book."

Lottie now attends a school for children with moderate learning disabilities and functions mentally at the same level as the average five-year-old.

And fortunately, her physical problems, which include stiff hands and taut skin, are relatively mild.

"Lottie still drives me crazy sometimes, pushing my eyelids open when I'm having a lie-in and demanding: "What are we doing now?"" says her mother. "She needs constant stimulation. But she's brought us so much joy and her development has far exceeded our expectations. Our lives are enriched by her disability.

"I remember watching Lottie fail to reach every milestone, thinking: "That's it; she'll never walk." Now, I'm in a position to reach out to a mum on the other side of the world going through the same agonies that I went through and allay some of her fears. And that is such a wonderful thing to be able to do."

Information on Unique can be found at www.rarechromo.org or 01883 330766.