

marie claire

“I Hope My Girls Have Good Lives, Without Fear Of Being Judged For Being Different”

A mum opens up about raising two daughters with a rare genetic disorder to mark Jeans for Genes day - by Kathryn Madden



Maddy and Briella Luk are in fits of giggles. Ed Sheeran plays in the background as they spin and pirouette around the room, little halos on their heads.

The sisters, aged eight and four, were both born with diastrophic dysplasia, a rare form of dwarfism that causes bone deformity and severe joint pain.

marie claire first spoke to their mum, Nicole Luk, last year, who [shared her heart-wrenching story](#) of discovering her baby had an abnormality at 17 weeks pregnant.

The genetic condition is autosomal recessive; Nicole and her husband Bernard are both carriers of the gene so each of their children had a 25 per cent chance of inheriting it. While older sister Lana didn't receive the gene, Maddy and Briella did.

Today the bright and bubbly girls are halfway through a six-month stint at The Children's Hospital at Westmead to have their spines lengthened before surgery. A "halo" or metal ring is attached to their heads along with some weights on a pulley system above, which works to stretch the spine.

"Maddy's curvature has decreased by 40 degrees. And Briella's has decreased by 20. The doctor's really happy with how things are going," says Nicole. "And it's really great that they're going through this together – they've got company and they're not bored. Their bond has solidified."

The procedure, however, has taken its toll on the family. Nicole and Bernard only see each other when they swap "shifts" at the hospital – Nicole sleeps there about five nights a week – and Lana, nine, "misses her family being together".



One avenue Nicole can turn to for support is her Facebook page, [Faith Hope Joy](#) (a title drawn from her three daughters' middle names). "I started a blog when I was pregnant with Maddy as a way of processing everything I was feeling," she remembers. "It was a little bit like a journal – and a way to inform people about what was going on without having to repeat the same story over and over."

Gradually it morphed into a Facebook page, which has connected Nicole with other families living with diastrophic dysplasia, and women enduring similar pregnancies. As such, Maddy and Briella have received cards in hospital from as far and wide as Brazil and Spain.

Nicole aims to help build awareness. Research, she says, is vital to treating rare genetic diseases and finding cures for tomorrow's children; equally important is advocating for people with disabilities. She notes a frustrating recent situation where the family took the girls out to dinner in their wheelchairs, but they couldn't get to the bathroom because of steps. It was a stark reminder of the obstacles Maddy and Briella may face.

"I just hope they're able to have good lives," she says. "To have the opportunity to do the things they want to do without fear of discrimination. Without being judged for being different and having a physical disability. I hope they'll be able to have the education and the career that they want, and find love if they want. All of those things. That's my dream for them."



Today is the 25th anniversary of Jeans for Genes Day. You can donate to the campaign [here](#).