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Sisters with rare genetic disorder undergo halo traction side-by-side

Like most sisters, Maddy and Briella Luk have laughed, cried, bickered and played together during their short lives.

But having diastrophic dysplasia has meant the siblings from north-west Sydney have also shared the ups and downs that come with a rare genetic disorder that affects their bones and cartilage.

Both girls were born with cleft palates and developed cauliflower ear, their limbs have become shortened and their breathing has been restricted because of their small ribs.

Briella, 4, and Maddy, 7, share a rare genetic disorder, diastrophic dysplasia, that has affected their growth and caused spinal deformities.
Photo: Louise Kennerley

But a spinal deformity known as kyphoscoliosis sparked the most recent challenge Maddy, 7, and Briella, 4, have faced side-by-side: Halo gravity traction to help straighten their twisted spines.

Maddy said the pair were fitted with their metal halos - "like what an angel wears" - together "so I don't feel scared".

Their operations at The Children's Hospital at Westmead almost three weeks ago were on the same day.

"They provide a lot of support to each other," their mother Nicole Luk said. "It's company as much as commiseration.

Metal halo devices, attached to Maddy and Briella's skulls with pins, will be weighted to gradually straighten their spines and prevent spinal cord damage and worsening respiratory problems.
Photo: Louise Kennerley

"Because it is such a long process, and a long hospitalisation, it can be really rough on the whole family. Doing it at the same time means we'll only have to do this process once."

The girls will spend the next six months on the orthopaedic ward while Mrs Luk and her husband Bernard commute to their Glenhaven home to care for their eldest daughter Lana, 9.

Metal pins drilled into the girls' skulls were fixed to the halo device, which was then connected to weights on a pulley system that will be increased to gradually stretch out their spines.

Further spinal fusion surgeries will fix their vertebrae in place.

Mrs Luk said the day of the surgeries was nerve-wracking but "magnetic, cheeky, adventurous" Maddy and "quiet observer" Briella were soon "back to their normal selves".

"I think all in all [the girls] have been about as accepting of it all as they could be. I guess they have their own coping mechanisms."

The Luks were living in Hong Kong and Lana was one when they were told at their 14-week scan their second child could have Down syndrome. Tests later showed skeletal abnormalities.

Nicole Luk said her daughter Briella, 4, was soon back to her normal "quiet observer" self soon after surgery.
Photo: Louise Kennerley

"There are about 200 possible genetic causes for that kind of thing and at the time we didn't know exact diagnosis," Mrs Luk said.

Mrs Luk said it was a worrying time because some of those conditions were lethal. The couple was given the option to terminate once doctors diagnosed Maddy with diastrophic dysplasia.

"We knew if it was a condition that was compatible with life that it's just a physical disability, a physical difference and ... the life potential is still great despite those challenges.

"Knowing that it could've been far worse helped us to accept the physical disability because we just wanted [Maddy] to live and have a good life, and she does."

Briella and Maddy will offer each other "company and commiseration" during their months-long stay at The Children's Hospital at Westmead.
Photo: Louise Kennerley

The disorder is an autosomal recessive condition, which means both parents carry a mutated gene. Each of the Luk's children had a 25 per cent chance of being affected.

Briella was born with the condition almost three years later.

The sisters will have to be monitored as their bones continue to grow to adult size and face more medical procedures, but are otherwise healthy.

"A lot of people might look at us and feel sorry for us, and I understand," Mrs Luk said. "But I don't want people feeling sorry for us.

“My kids are happy and in many ways just normal kids. That said, I do believe more research
needs to be done into ways to improve their quality of life, both physically as well as socially.”

The family supports the Jeans for Genes initiative, which this year marks its 25th anniversary.
Funds raised go directly to the Children's Medical Research Institute's research into conditions that affect children including genetic diseases, cancer and epilepsy.

Institute director Roger Reddel said there were more than 6000 known inherited conditions and one in 20 children was born with some kind of birth defect or inherited disease.

He said rapid advances in diagnosing genetic conditions had been "very powerful for families" and researchers were developing technologies to treat, and even cure, some diseases.

Nicole, Maddy, Briella, Lana and Bernard Luk at their Glenhaven home before the girls' surgery.
Photo: Louise Kennerley

"I don't want to give false hope for everybody and some people would be rather sad to know in some cases it won't be fast enough for them.

"For the community as a whole there's a lot of hope and cause for optimism."

Mrs Luk said the family focused on treating the girls' bodies as they were, and she was grateful for doctors and surgical options that meant they could lead a long life as pain-free as possible.

"I love my kids and I would love them whether they're short or tall, but I would love for them to need less time in hospital and to reduce the severity of the procedures they need.

"As a mother, if there was anything to lessen the impact of the condition on their bodies I think I would do it, and you won't have that without research."