MARILYN MANSON SURVIVOR: 'HE'S A MONSTER'

Who
May 24, 2021

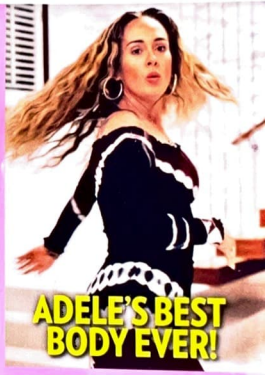
MEGHAN & HARRY

JEALOUSY, LIES & REGRETS

The couple face a shock new crisis as the bitter family feud worsens

J.LO & BEN AFFLECK

INSIDE THEIR SECRET GETAWAY



ADELE'S BEST BODY EVER!

PRINCE FREDERIK AND PRINCESS MARY

WHAT WE GAVE UP FOR LOVE

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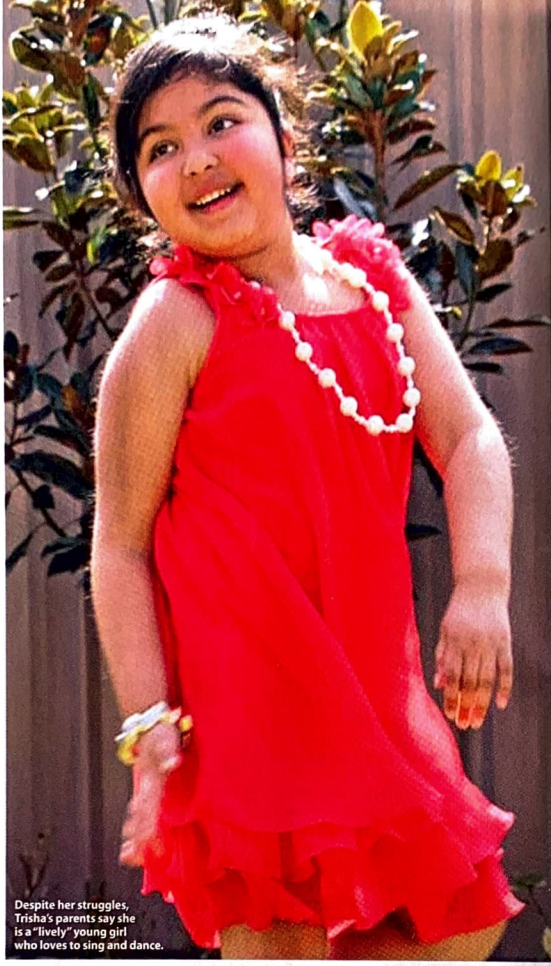
Melbourne Girl's Rare Gene Disorder

OUR FINAL HOPE

TRISHA SAWHNEY'S PARENTS HAVE FOUND A WAY TO TREAT HER RARE GENETIC DISORDER 'AGU', BUT IT COMES WITH A CATCH



Despite her struggles, Trisha's parents say she is a "lively" young girl who loves to sing and dance.

She was born with an ultra-rare genetic disorder that causes a raft of physical and mental disabilities, but there is one trait of the disease that seems particularly cruel for Trisha Sawhney. Within a few years, the 12-year-old will lose the ability to swallow. For a schoolgirl who loves to spend her spare time cooking, and thrives on baking biscuits and cupcakes, it will be a devastating blow.

"Every time we're eating and she coughs, we think that she's getting closer to not being able to swallow," her father, Neeraj Sawhney, tells WHO from the family home in Burnside Heights, Melbourne. "It's always in the back of our minds. It just breaks us."

That symptom is just the tip of a debilitating disease. Trisha has a condition called Aspartylglucosaminuria (AGU), a neurodegenerative disorder in which certain protein molecules are not broken down, leading to a build-up of proteins in the body and causing severe developmental problems (see box). It is so rare that Trisha's is the only known case in Australia.

Now, seven years after Trisha's diagnosis, her parents are finally seeing a glimmer of hope. Doctors in the US have agreed to conduct a clinical trial in an attempt to cure nine AGU patients throughout the world, including Trisha. But the procedure costs $2.6 million, an amount that the patients' families do not have.

"When we first found out Trisha had AGU, we were struggling mentally because we were not able to do anything for her," says Neeraj. "Since we found out there is something we can do, we've been very positive. There is hope."

Since her diagnosis, Trisha has been in and out of hospital with various health issues, including scoliosis.

The couple are determined to find a cure to help save their daughter and help her have a normal life.

FINDING A CURE

"We were devastated the moment we found out that she may not have many years to live," says Neeraj. "But my wife was very positive about finding a cure for Trisha and motivated me to get back on my feet and start looking for solutions."

Born in Melbourne in 2008, Trisha was the first child of Neeraj and Vandana, who immigrated from India in 2004. "We were over the moon," says Neeraj, an ANZ product manager. "Everything was normal and she was doing alright."

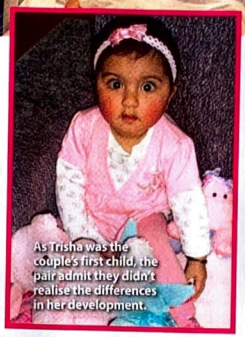
But as the months went by, the couple noticed Trisha's development was slow compared to other babies. "Her crawling was delayed, but not so bad that doctors were concerned," says Neeraj. "She was not walking until 18 months, but again the doctors said, 'Yes, some kids are slow, it's nothing to be alarmed about.'"

Still, the couple, who are in their 40s, could not shake off their concern. When Trisha began her schooling, her speech was underdeveloped and she would not speak to her teachers. It was around this time, that the family's paediatrician sensed an underlying problem.

"At the age of five, Trisha was tested and a urine test came back as positive with AGU," recalls Neeraj. "It was so rare the doctor didn't know what it was. It was devastating. I wanted to quit my job to spend all my time with Trisha. I thought that she wasn't going to be with us for very long."

Since then, Trisha's condition has deteriorated. She struggles to tie her shoelaces, climb a staircase or simply jump on the spot. She is also developing scoliosis and requires surgery to correct the curvature of her spine. "Her hearing and eyesight will be impacted," says Neeraj. "And one day, she won't be able to walk."

Even so, nothing hampers Trisha's zest for life. She loves swimming, watching YouTube cooking channels and she makes her own dancing videos on TikTok. "She's a very happy girl, very cheeky," says Vandana, a former administration worker who is now a full-time mum to Trisha and her younger sister Samika, 10. "She loves cooking and baking, and loves to keep cooking something new."



As Trisha was the couple's first child, the pair admit they didn't realise the differences in her development.

Adds her dad: "Just talking to her is a charm. But it's heartbreaking to see her not being able to read, write and play like her younger sister."

Her only hope is to undergo the clinical trial, which involves gene replacement therapy (replacing the faulty AGU gene with a normal gene). For the trial, the eight participating families – they come from the US, Canada, Switzerland, Spain and France – are crowdsourcing their share. The Sawhneys have pledged to raise $500,000, for which they have created a GoFundMe page (at press time, they had so far raised nearly $100,000).

"It's not easy to ask for money," says Neeraj. "There was a time when we both lost our jobs, but we didn't go to Centrelink – we just did odd jobs and got through it. But we have no choice now. We can't do the clinical trial without help." Help that could save their "beautiful girl".

"Trisha can't run around like a normal child, she's very clumsy and she trips over easily," says Neeraj. "But she forgets about her problems very quickly. She's a very happy-go-lucky girl. You can't help smiling when she looks at you."

• *By Michael Crooks*

To donate, scan the QR code above, go to savetrisha.com.au or gofund.me/2b623b51

SCAN ME

WHAT IS AGU?

AGU is a genetic and neurodegenerative disorder that results in the absence or shortage of the aspartylglucosaminidase enzyme in lysosomes (cells that contain digestive enzymes). As a result, glycoproteins build up within cells to toxic levels, disrupting normal cellular function throughout the body, including in the brain. It is a fatal disorder; people with AGU have a shortened life span. There are around 120 cases in the world, with most cases occurring in Finland. According to the US Rare Trait Hope Fund, AGU causes "delayed speech, attention deficit, clumsiness, restlessness and continuous respiratory infections." www.raretrait.com

COURTESY OF TRISHA SAWHNEY.