

### Jeans for Genes awareness: Nia's story

When Nia was two-years-old, she was diagnosed with DUP15Q syndrome.



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WA News

## Jeans for Genes Day: Nia Collava's life with DUP15Q syndrome

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EACH morning Nia Collava asks her mum whether she will finally get to meet her idol, "Emma" from The Wiggles.

“Wiggles mummy?” the adorable little girl with a rare genetic disorder asks with eagerness every day. And every day her mum, Sarah, has to tell her almost-four-year-old daughter, who has DUP15Q syndrome, that she has to wait until November.

With Nia’s reduced development equating to a two-year-old, waiting so long to see Emma do her ballerina twirls must seem like an eternity.

Nia’s world and that for her mum and dad Domenic, 27, can be complicated. The syndrome causes frequent epileptic seizures, delayed motor skills and speech, and low muscle tone. There are only 70 known cases of the syndrome in Australia and little is known of the disease.

The syndrome, which is non-hereditary in Nia’s case, has also given her autism, which reduces her sociability, except with brother Jack, 2.

But Sarah said Nia responded well to physiotherapy, speech and occupational therapy.

“We don’t know what her future will be like,” Sarah, 26, said. “We don’t have any answers, there’s no text book on this. That’s why research funding for rare genetic disorders like this is so important.

“We need more research to help children like Nia, to give us and other families more answers. We get by because we have to and she’s our little girl — we wouldn’t change her for the world. But we’d like a better future for her.”

The Collavas are helping raise awareness for Jeans for Genes Day on August 4.

<http://www.perthnow.com.au/news/western-australia/jeans-for-genes-day-nia-collavas-life-with-dup15q-syndrome/news-story/80ebd00059eb39bef9cad0b319de6f0a>