Chris Kufner with Lakyen, nine months; Rebecca Nunn with Jack, 5; Dr Miya St John, Mel Anderson with Sarah, 16; and Carina Kufner with Totten, 4, will do Melbourne Marathon to raise funds for research into PURA syndrome. Picture: Wayne Taylor

Finding strength in community

Shannon Deery

When Sarah Anderson was diagnosed with PURA syndrome in 2014, she was the only child in Australia known to have the rare genetic disorder.

A decade on and the 16-year-old is now one of just 26 children known to have the neurodevelopmental condition.

On Sunday, they will band together to run, jog, wheel or walk the Melbourne Marathon to raise funds for desperately needed research into the syndrome.

In just the first year since it was founded, the new PURA Foundation Australia has self-funded two key studies.

Now it is working with the Murdoch Children's Research Institute to research speech and communication for PURA syndrome.

Sarah's mum, Mel, said recognition of the syndrome had come a long way since she had to have a US lab diagnose her daughter

"When you're incredibly rare, your strength is in coming together," Ms Anderson said. "Community is everything and we've built a community that can work together."

Globally there are just 706 children diagnosed with the syndrome, which can lead to developmental delay, decreased muscle tone and speech difficulties.

"Our kids can be quite severe, they're non-verbal, but they're very happy kids," Ms Anderson said. "There is physical disability, intellectual disability and severe epilepsy ... there can be very difficult days, but you find strength in your community. The foundation means we can support families and help initiate life-changing research."

To donate to the team, visit purafoundation.au