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Rare diseases not so uncommon

Terra Sword | 1st March 2012

If there's to be a poster boy for Rare Diseases Day, Goonellabah's Ashton Hayes should be a leading candidate.

Five-year-old Ashton is not only lively and cute as a button, but he's one of only four people in the world with the genetic condition Atypical Rett Syndrome. And this year he started big school.

He attends Wilson Park Public School in East Lismore, which caters for kids with specific purposes like Ashton, who is dependent on a wheelchair for mobility and has limited speech. His mum Angela says like most kids starting school, he can't wait to put on his uniform each morning and get the day started.

"He's absolutely loving it... he gets so excited when we turn into the school driveway every morning he starts to vocalise and he swings his arms around wildly and kicks his legs," Angela said. "He hardly gives me a second glance when I say goodbye and give him a kiss on the cheek," she laughed.

"He has come along in leaps and bounds (since starting school)... while he's very tired and fatigued at the end of the week, he's just a sponge taking in all his new learning, and he loves it."

Angela said marking Rare Diseases Day could potentially be an important day for many parents at the school, who can learn about the new national organisation that was established last year - Rare Voices Australia - and see if they fit the criteria for joining.

After talking with the principal of Wilson Park School, Helen Rae, Angela said more people may be included in the organisation's parameters than they first thought.

"But we haven't connected the dots yet... I would love there to be a local support group get together," Angela said. "When you have a child with such a rare condition it can make you feel quite isolated and alone. If all the people with rare conditions join together all of sudden you're not so alone - it's suddenly common that you have a rare disease diagnosis!"

She said for people who have a child with a disability but no specific diagnosis, which can be a terribly frustrating place for a parent to find themselves, it could create important bonds.

"I think it could be really important for parents and families who don't have a specific diagnosis, such as when the child has a development delay but the root cause is never known. We were lucky with Ashton that his genetic deletion showed up because of a new test, but just a few years before, without that technology available, we would have found ourselves in the category of no known diagnosis... and still searching for an answer," Angela said. "It doesn't change the fact the condition exists, but a diagnosis gives you closure that there's an answer, and you can move forward."

She said being able to connect and talk to other parents and find out information about rare conditions, and what facilities and funding are available would be a wonderful thing.

"The basic human need of belonging and feeling like you belong is really important."

The idea for Rare Voices Australia came from a recent symposium in WA which identified the need for an umbrella organisation to bring together the disparate groups and families affected by rare diseases.

While still in its infancy - the board met for the first time last week - board member Dr Tracy Dudding said the organisation could help raise public and political awareness and lobby for increased funding for research and work towards providing equity in access to care and treatment.



Ashton Hayes, 5, and mum Angela (front) with (rear l-r) Wilson Park School principal Helen Rae and students Shannan Nurse, 17, and Hayley Cameron, 7.

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"It's estimated about 1.5 million Australians have a rare disease - about the same as diabetes. We talk about the paradox that while each of the conditions is rare, it's not unusual people have rare diseases," Dr Dudding said. "People with rare diseases are commonly known as 'health orphans' because for most of them there's limited information about the causes of disease and therapies for them, so they are often neglected."

"One would hope the long-term plan (of Rare Voices Australia) is to have an Orphan Disease Act which would enable allocated funding for research into rare conditions... At the moment rare diseases aren't considered a health priority in terms of research and we'd like to see that change. If there's no research, there's no development of treatment."

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