

# Raising awareness of rare diseases

Ely Pleitez suffers from the rare degenerative disorder ataxia-telangiectasia - more commonly known as A-T. So rare is the condition, there are only some 50 diagnosed cases in Australia. As such, treatment and support options for patients are severely limited. While Ely isn't looking for a cure, she is hopeful that medical researchers can develop treatments to make life easier for her and her family.

Professor Martin Lavin leads a team at the UQ Centre for Clinical Research working to unravel the mysteries of A-T. As Professor Lavin explains, A-T is a multi-system disorder, affecting a variety of body systems. "A-T affects the brain, lungs, liver and immune system. While our research has focused on a number of these organs, we're now concentrating on the lungs - because lung disease kills up to 40 per cent of A-T patients."

Dr Abrey Yeo, a researcher in Professor Lavin's team, is conducting research to profile the microbial species (microbiome) present in the airways of patients with A-T. "If we are able to identify the microbiome, or a trend in the microbial profile of these patients," says Dr Yeo, "then we can develop more targeted therapies using more effective antibiotics."

While Professor Lavin and his team conduct their research into A-T, Ely creates paintings to build awareness and raise valuable funds. Ely's mother Sandra says her paintings have raised thousands of dollars for research and support initiatives. "Through painting, Ely is able to express what she likes," Sandra says. "She feels good that the money she raises helps researchers, as well as organisations such as BrAshAT and Montrose, which support us."

As Ely explains, due to A-T's far-reaching effects, people like her require full-time care. "I need 24-hour support," Ely says, "and I get that from my Mum. She can't work. She can't do lots of stuff with my sisters because she has to look after me."

Sandra says Ely's goal is to continue to raise awareness of A-T, and promote support organisations such as BrAshAT. "A lack of awareness means that support in the public system is very limited for A-T patients," Sandra says. "Even small things are difficult to organise because most people don't know what A-T is. Everything takes a long time, and this is worrying when Ely is sick. ●

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“Raising awareness for research and support costs a lot of money. We want people to look at the organisations that help A-T patients and see what they can do to help.”

