Getting a degree and inspiring a foundation

Murdoch University graduate Conor Murphy has not let Duchenne Muscular Dystrophy (DMD) stand in the way of earning a double degree in Politics and History.

A rare form of Muscular Dystrophy, DMD sees muscles deteriorate progressively over time due to the under-production of the protein dystrophin. The disease stems from a genetic mutation and only affects males.

According to his mother, Lesley Murphy, Conor was first diagnosed at the age of three-and-a-half and was confined to a wheelchair by ten. At 15, he required assistance with feeding, and by his final year at university, Conor was left with only the use of his fingertips.

Despite these challenges, Conor found strategies to keep studying.

“The biggest challenge was developing new ways to get around limitations which were constantly changing and gradually getting worse. I had to develop my memory skills so that I could remember things I wasn’t able to write down in time, as it took me longer to write things, and eventually I couldn’t write at all,” Conor said.

“When I started at uni I could use the readers like anyone else, but by my final year I had to use a book stand and record notes on a voice recorder, which I would type up later on my computer.”

Conor credits Murdoch University for offering support, including flexible exam arrangements, funding arrangements for support workers, good accessibility and providing a special room in the library with technical aids for study.

While he concedes he felt like giving up at times, the desire to achieve something he could be proud of later in life kept him going.

“I wanted to prove my worth as a member of society. I was also conscious of being a trailblazer for others with a similar disability to show the younger guys that life doesn't end at diagnosis and that even with a disability you can achieve anything,” Conor said.

This spirit was part of the inspiration for a newly formed not-for-profit, Rare Voices Australia, which hopes to become the peak body for ‘rare diseases’ nationwide. Its foundation came out of wide community consultation with those living with rare diseases, including Conor's mother, Lesley, who put her experience as a long-time grassroots advocate to work as a co-founder.

Like her son, she is not prepared to sit back and wait for others to do something.

“If you think things need to change, then I believe it is up to you as an individual to ask, ‘what can I do to help bring about the change?’” Mrs Murphy said.
“After the first ever Australian Rare Diseases Symposium in Fremantle in 2011, there was a unanimous call for a national organisation to represent people with rare diseases, but like with most good intentions, no one did anything tangible.”

“So a group of us [people living with a rare disease] decided to make it happen ourselves. We established Rare Voices Australia in February 2012 and have been on a steep learning curve ever since.”

Mrs Murphy said while no internationally accepted definition for rare diseases exists, the generally accepted number of conditions is 7,000 to 8,000, most caused by genetic mutations.

She said Australia was about 20 years behind the rest of the world in terms of having a national organisation and databases but was now in a rapid growth phase and was being assisted internationally to bring our rare disease alliances up to speed with those in Europe and the USA.

As for Rare Voices Australia, its charitable status is now before the tax office, a new website is up and running and Mrs Murphy has been invited to attend the Shanghai Medical Association Society of Rare Diseases conference later in the year.

The organisation also recently held its first community consultation in Sydney on September 13, which brought together nearly 100 rare disease stakeholders to start the hard conversations that will serve to inform a working plan for the next 12 to 18 months.

“We don’t have all the answers. We need to hear what people want us to do, so we can go out and act for them. If they want us to lobby for increased therapies, access to international drug trials or push for research, that’s what we plan to do,” Mrs Murphy said.

“Our mission is coming from the grassroots, from the people who have been knocked back for life-saving drugs, or aren’t getting access to therapies or have spent 20 or 30 years waiting for a diagnosis for their disease while being told they were simply unwell or were making things up or were accused of Munchausen disease.”

Mrs Murphy knows improving treatments will be high on the agenda. She said one problem with rare diseases – especially those that affect only a handful of people in the country – was that there wasn’t a political will or economic incentive to pursue research and drug development.

This is why she sees genetics research as pivotal.

“We’re hoping some of the genetic discoveries being done on certain diseases can be expanded and translated into personalised therapies for other types of conditions,” Mrs Murphy said.

This is where Professor Matthew Bellgard, Director of Murdoch’s Centre for Comparative Genomics (CCG), comes in. His Centre is collaborating with
esteemed researchers from elite organisations nationally as well as in the USA, China, India, Germany and Japan.

CCG analyses DNA across the spectrum, from plants to bacteria, animals and humans, comparing genes from different species (and between individuals within species) to look for corresponding genomic defects.

This cross-species comparative approach, for instance, has led to the successful discovery of mutations in a gene responsible for the most common form of human hereditary colorectal cancers. These mutations were identified thanks to an equivalent gene characterised in the DNA of yeast.

Professor Bellgard said he encountered Mrs Murphy in an indirect way but was immediately impressed with her resolve.

“One of the projects we’ve been involved with is the design and development of web-based national disease registries for the Office of Population Health Genomics within the WA Department of Health and Aging, including those related to neuromuscular diseases,” Professor Bellgard said.

“The first one we built was for Duchenne Muscular Dystrophy. It came from strong lobbying by the Duchenne Foundation and Muscular Dystrophy Foundation members such as Lesley Murphy and other mothers, particularly Deb Robins in Queensland.

“It was a real grassroots effort with mothers all across Australia and New Zealand pushing for it. Lesley wanted Conor and other boys and young men afflicted with this disorder involved in international drug trials.”

Up until CCG built that national registry, no one in Australia knew collectively exactly how many patients had DMD, or their particular gene mutation. An awareness of the challenges facing those dealing with rare diseases prompted Professor Bellgard to look at the bigger picture.

“Instead of building one registry for DMD, we decided to build a robust, web-based framework that could be adapted for use with other rare diseases,” Professor Bellgard said.

“Our goal was to provide a way for as much information as possible to be integrated together – disease types, locations, prevalence, symptoms – to improve clinical diagnoses and inform drug and gene research.

“All of this background information is vital to move forward. If funding bodies and agencies aren’t informed, they can’t make good decisions.”

Professor Bellgard has recently invited Mrs Murphy to join his team at CCG as an Adjunct Research Associate, hoping that together they can help the up to two million people who are too often on the outside of the mainstream medical spectrum.
“Being at Murdoch gives Rare Voices Australia support and shows people believe in what we’re doing. To have the support of Matthew Bellgard, who is passionate about the research, is wonderful,” Mrs Murphy said.

Meanwhile, Conor is pursuing big goals of his own. Since February, he’s been putting his degree to good use, working as a researcher and speech writer for Fremantle MP Melissa Parke.

“I’m working two days a week and really enjoying the experience. But who knows, I may go back to Murdoch University in the future and do more study. I’m thinking maybe Business,” he said.

To learn more about Rare Voices Australia, visit rarevoices.org.au.