

## RARE DISEASE DAY

### *Statement*

**HON MATTHEW SWINBOURN (East Metropolitan)** [5.18 pm]: Tonight, I stand to make a statement about Rare Disease Day. International Rare Disease Day is normally observed every year on 28 February, but this year, given the rarity of 29 February, it falls on 29 February. Obviously, today is not 29 February, but because we will not be back before 29 February, I want to take the time now to make a statement about Rare Disease Day. The theme for Rare Disease Day 2020 is “Bridging health and social care” and the 2020 slogan is “rare is many”. Rare Disease Day raises awareness of rare diseases among the general public and encourages researchers and decision-makers to think more about rare diseases.

A rare disease is defined as a life-threatening or chronically debilitating disease that is of such low prevalence—less than one in 2 000—that special combined efforts are needed to address it. As I said before, it is no coincidence that Rare Disease Day is on 29 February this year. Examples of rare diseases include rare childhood cancers and other better known conditions such as cystic fibrosis and haemophilia, which have symptoms present from birth or, as with Huntington’s disease, with symptoms arising in adulthood. Eighty per cent of rare diseases are genetic in origin, with the age of onset of symptoms ranging from early childhood to adulthood.

Last year, as part of the annual parliamentary research program, I asked a UWA student, Anneliese Ng, to look into rare diseases in Western Australia, particularly in relation to funding. She has produced for me a fantastic and insightful report that gives an overview of rare diseases and their social and economic impact, Western Australia’s capacity to make meaningful contributions to research, and the most worthwhile research opportunities, some of which I will speak about.

I find it interesting that, even though prevalence of rare diseases has to be below 0.05 per cent of the population for them to be called rare, rare diseases are, in fact, not very rare at all. They affect more than 190 000 Western Australians, which equates to roughly seven per cent of the population. I am included in that group, as are two of my sons and my wife, so in our household rare diseases are, unfortunately, exceptionally common. It is the same for many families.

This cohort includes some 63 000 children, and if we think about that number, it is enough to fill Optus Stadium. As I say, rare diseases are in fact not rare in their overall prevalence, but in their individuality. Therefore, the slogan this year, “Rare is Many” is quite apt. It is estimated that there are more than 8 000 rare and undiagnosed diseases, and it is important to recognise those who have undiagnosed diseases because they never get a name for their condition or their symptoms, and they do not get treatment. So, as I say, while the diseases themselves are individually rare, being diagnosed with a rare disease is more common than we might think.

What is worse is that this figure is conservative and it is likely to be much higher, as there are a number of significant issues with the diagnosis of rare diseases. In a survey of 810 adults with rare diseases, researchers found that 30 per cent had to wait five years or more for a proper diagnosis; 62 per cent had to see three or more doctors before receiving a diagnosis, and 46 per cent of those diagnosed were diagnosed wrongly at least once. That is a real struggle, and therefore the theme is “Bridging Health and Social Care”. This theme is directed at bridging the gap and coordinating medical, social and support services to tackle the challenges that people with rare diseases and their families face, particularly in respect of getting a diagnosis and appropriate treatment.

Unfortunately, people diagnosed with rare diseases in WA stay in hospital 24 per cent longer and have 110 per cent more hospital discharges than the general population. Although rare diseases people represent only seven per cent of the population, they contribute a significant cost to our healthcare system, although not through any fault of their own. That is due in part to the lack of appropriate care and treatment that is available to many of them. The cost of discharges related to rare diseases works out to approximately \$395 million annually.

Ms Ng also found that in most cases, unfortunately, only the symptoms of rare diseases are treated, not the underlying disease pathology. For many there is no cure, only treatment. That is a hard thing to face, because when we get sick or when we suffer, we want a cure, not just treatment. As I say, for many there is no cure at all. This is why Rare Disease Day is so important. With seven out of 100 Western Australians expected to be diagnosed with a rare disease, more must be done to address issues with diagnosis and treatment. It is pleasing that Western Australia is a leader in rare disease research. Ms Ng notes that WA was the first state in Australia to develop a rare disease strategic framework, which was released in 2015. Further, the WA Department of Health is one of only two Australian members of the International Rare Diseases Research Consortium, ensuring that WA is at the forefront of international best practice.

Last but not least, WA is the first jurisdiction in the world to have implemented an undiagnosed diseases program in its public health system, which aims to provide definitive diagnoses to those with chronic, complex and typically multisystem diseases. It is also worth noting that next week the federal government will launch its framework. Hon Greg Hunt, the federal Minister for Health, will launch the national strategic action plan for rare diseases.

I commend the federal government for taking that action. I also commend the McGowan government for its support and input into that program, which is really important to help deliver better outcomes for people with rare diseases.

My wonderful parliamentary research student did a great report and made a number of suggestions that I think are worth sharing, including that we should leverage our established advantages and direct part of the WA future health research and innovation fund to rare disease research. One reason it is an effective means of directing funds is that there is in many respects a lot of hard work to do, but also a lot of low-hanging fruit. From the point of view of public policy, rare diseases often do not attract the level of investment that other diseases do because the financial returns for drug companies and research institutions is just not the same. However, we continue to have massive costs in our public health system—and the private system, for that matter—to support those who bear the burden of rare diseases. It would be good to see a significant amount of that money, through proper research and a supported evidence-based approach, allocated to rare disease research.

Some of the most well-known rare disease research in Western Australia happens in the space of childhood leukaemia. We have some of the best researchers in Australia through the Telethon Kids Institute. They are also supported by groups such as the Children's Leukaemia and Cancer Research Foundation to really advance those things. I have spoken before about the advances that we have made in childhood leukaemia and how well our Western Australian researchers have contributed to them. The national plan will be launched next week, and I hope that other states will, like Western Australia, take up the mantle of rare disease research, because that will help to advance and push it forward, as in that old saying, many hands make light work.

In the short time that I have left, I would like to talk about a young man I met with, Andrew Bannister. Andrew posted his story to the Rare Disease Day. He states —

My name is Andrew. I live in Perth Western Australia I am 25 years old

I was born with three Brain Malformations, one is Periventricular Nodular Heterotopia ... the second is Polmicrogyria ...

I apologise for not being able to say that correctly. One of the problems with rare diseases is that the language around it is almost impenetrable to ordinary people. He continues —

... and I also have Cerebellar Hypoplasia. My family and I found out when I was 17 years old I had these three malformations. I didn't talk until I was six and half, before then I used sign language, I started walking when I was four and a half. I am now 25 years old. In late 2008 I started doing para Athletics from 100m to 1500m on a National level. In 2014 I started para cycling, racing a tricycle. In 2015 I went to Adelaide for the National Championship and I got 2 gold medals in the Time trial and the Road race and became the National Champion for my T2 classification. I hope one day to go to the Paralympics and represent Australia. One of my biggest goals is giving back to the community ...

He plans on doing that on Rare Disease Day because he plans to do what he called a 50-kilometre social ride with his cycling group around Perth. Andrew, on your 50-kilometre social ride, I wish you well. On 29 February, I hope that everyone reflects on Rare Disease Day.

**The PRESIDENT:** I just want to say that Andrew is, indeed, a remarkable young man, and that Parliament will be lighting up to acknowledge Rare Disease Day on 29 February.