UWA Drug Treatment Offers Hope For Young Sufferers Of Duchenne Muscular Dystrophy

University of Western Australia
University of Western Australia's Centre for Neuromuscular and Neurological Disorders
Western Australian Neuroscience Research Institute

Professors Steve Wilton and Sue Fletcher.

Some 20,000 chlidren are diagnosed each year with Duchenne muscular dystrophy, a fatal muscle wasting disease, which is caused by errors in their dystrophin gene. Affected children are usually confined to a wheelchair by the age of 12 and succumb to their illness by 30. The disease almost always affects boys.

Professors Steve Wilton and Sue Fletcher developed a treatment at The University of Western Australia's Centre for Neuromuscular and Neurological Disorders (CNND) and the Western Australian Neuroscience Research Institute (WANRI). The treatment is designed to allow the body to make a shortened but functional version of dystrophin - an essential protein involved in muscle fiber function and lacking in patients with DMD.

> *This is the first treatment for DMD that addresses the cause of the disease and the hope is that it will slow its progression and keep patients mobile for longer.*
>
> UWA's Deputy Vice-Chancellor Research Professor Robyn Owens

In 2016, the US Food and Drug Administration granted an accelerated approval for the drug. Sarepta Therapeutics, a US biotechnology company that licensed the development and commercialization rights, now manufactures and markets the drug as Exondys 51®.

The treatment helps patients with a deletion mutation adjacent to exon 51 of the dystrophin gene (about 13% of the DMD population). Treatments targeting different exons are currently in clinical trials, with a treatment addressing exon 53-flanking mutations (another 8% of the DMD population) expected to receive FDA approval later in 2019 and a treatment for exon-45 flanking mutations expected in early 2020.

"What Steve and Sue and the team at Sarepta have done is amazing – to think that the results in a lab notebook at UWA have gone on to be translated into a treatment for a disease where there is currently little hope is amazing," Owens said. "It demonstrates how universities can be the source of great ideas and how partnering with experts in industry can create real impact."

The patented treatment should be useful for DMD patients with a mutation in a particular region of the dystrophin gene and the race is on to develop similar treatments to treat more DMD patients.

Simon Handford, Manager Research Commercialisation at UWA, said the technology transfer journey from laboratory to patient was an extensive process.

"We've been working with Steve and Sue since 2004 and it's so satisfying to have played a small part in the process from discovery to approval," Handford said.

"Our hope is that this inspires other researchers to think about how they are going to translate their research findings – whether in medicine, agriculture, engineering, business – and to think about partnering at an early stage."

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