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Dystonia research receives funding

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NEUROLOGIST and post-doctoral research fellow at Garvan Institute of Medical Research, Dr Kishore Kumar has formed a team to use Garvan's whole human genome sequencing capabilities to study dystonia.

Dystonia is a neurological movement disorder involving sustained or intermittent involuntary muscle contractions that cause abnormal and often repetitive movements or postures, or both. It is the third most common movement disorder.

"Dystonia can affect many different parts of the body," he told Dubbo Weekender. "For example, dystonia affecting the legs can lead to a tendency for one foot to turn in or drag. If dystonia affects the arms, it can lead to pain or stiffness in the hand.

In other cases, the neck may turn or pull involuntarily (cervical dystonia). If dystonia affects the muscles around the eyes, it can lead to 'blepharospasm', which involves involuntary twitching, blinking, closure or squeezing of the eyelids.

"Dystonia can even affect the muscles in the larynx, or voice box, causing difficulty speaking (spasmodic dysphonia). Generalised dystonia affects the entire body and usually starts in childhood. Some forms of dystonia are associated with a specific task, such as writing (writer's cramp) or playing a musical instrument (musician's dystonia)," said Kumar.

As there are few treatments

and no cure for this condition, Kumar's team, which includes Professor John Mattick, Professor Carolyn Sue, Associate Professor Marcel Dinger and Dr Mark Cowley, will reignite research thanks to a donation from the Paul Ainsworth Family Foundation; a cause supported by Paul's family as he suffers from the condition himself.

"Our study will utilise highly advanced gene sequencing technology, known as the Illumina HiSeq X Ten at the Garvan Institute of Medical Research. We think this will be the most accurate and detailed way of studying the genetics of dystonia," said Kumar.

"We will establish a large genomic database of patients with dystonia, making this an innovative and cutting-edge project that other researchers from around the world will follow."

Seventy thousand Australians are impacted by dystonia and the Garvan study will focus on two of the most common which affect the neck and vocal chords. In most cases, the cause of dystonia is unknown, however, several gene changes or faults have already been confirmed as causing dystonia.

"Our team is trying to understand the genetic basis for dystonia. By studying the proteins that these genes code for, we hope to identify new disease pathways, which may lead to a cure for dystonia in the future," Kumar said. "My team is passionate about understanding this disorder and developing better treatments and cures.

"You should visit your GP if you are concerned that you may have this condition. Your GP may decide to refer you to a neurologist for further assessment.

The neurologist can help confirm the diagnosis and may arrange additional investigations, which in certain circumstances could involve genetic testing.

Paul and Valeria Ainsworth, directors of the Paul Ainsworth Family Foundation, said they decided to facilitate the research after learning about Garvan's leading genetic research capabilities.

"Hopefully as a consequence of this research, new options for treatment, prevention or a cure will eventually emerge," Paul Ainsworth said.

"My wife and I are absolutely confident in Garvan's researchers and the technology at their disposal to achieve goals otherwise considered unattainable."

Andrew Giles, CEO of the Garvan Research Foundation said philanthropic support of this nature is vital to sustain innovative research projects, such as the Ainsworth Dystonia Project.

"Philanthropic support like this is absolutely vital to our work. I've been working with dystonia patients for many years and I have seen the debilitating impact of the condition first hand.

"We're very grateful to the Paul Ainsworth Family Foundation for providing important support to a disease area in dire need of more attention. The reality is, for every dollar of funding our researchers receive, we still need to raise another 70 cents in order to sustain research projects. So, the Ainsworth's philanthropy allows Dr Kumar and his team to focus on dystonia, understand the cause and hopefully, find better treatments or even a cure," said Mr Giles.

The Garvan Institute in Dubbo does not conduct this type of research. It is dedicated to researching osteoporosis.



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- Dr Kishore Kumar,
Neurologist and postdoctoral research fellow at Garvan Institute of Medical Research

