Lessons from the 'gene for speed'

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As you prepare to watch the world's best athletes competing at the London 2012 Olympics, have you thought about what distinguishes elite sprinters from long-distance athletes?

The University of Sydney's Kathryn North is an internationally renowned genetic researcher whose work includes the world-first discovery of the 'gene for speed', known as alpha actinin 3 (ACTN3). A common variant in the ACTN3 gene results in alpha-actinin-3 deficiency in one in five people, which influences muscle function and performance.

"Whether or not you have ACTN3 really influences whether you are going to be better at being a specialised sprint athlete or a specialised endurance athlete," says Professor North, who worked with some of Australia's top athletes to make her findings.

"To date, no Olympic sprint athletes at the very short levels of distance, like 100 metres, have been found to be deficient in alpha actinin 3."

However Professor North's research into the way muscles work in elite athletes has wider implications. Her work on the gene started with a desire to see how it might influence muscle disease.

"If alpha actinin 3 influences how muscle works in elite athletes, can it influence disease severity in kids with muscular dystrophies?"

Questions such as this are particularly relevant to Professor North in her role as Head of the Institute for Neuroscience and Muscle Research

based at the Children's Hospital at Westmead.

"We've demonstrated that whether or not you have alpha actinin 3 does influence the severity of the disease. And now we understand the molecular mechanisms, we can start to harness that in terms of directing future therapies."

Her work has also provided insight into genetic risk factors for common health problems in the general population such as obesity. Professor North has shown that the absence of ACTN3 influences skeletal muscle performance, muscle and fat mass, adaptive response to diet and exercise and susceptibility to muscle damage.

"What we've found is that if you don't have alpha actinin 3 you actually gain weight much more slowly on a high-fat diet."

Provided by University of Sydney