Key finding in rare muscle disease

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The finding is in the current issue of *Annals of Neurology*, a leading international neurology journal, in work led by Professor Nigel Laing and Dr Kristen Nowak of the Laboratory for Molecular Genetics at the Western Australian Institute for Medical Research (WAIMR) and done in collaboration with a number of European researchers.

Professor Laing said his team had discovered a number of children across Europe who, despite a complete absence of the crucial skeletal muscle protein actin, were not totally paralysed at birth, and managed to have some muscle movements.

"This intriguing finding led us to look deeper and what we saw is that while these children do not have any skeletal actin in their skeletal muscle, they have another form of the protein, known as heart actin, in their skeletal muscles," said Professor Laing.

"It appears the more heart actin they have in their skeletal muscles, the more movement they have."

Professor Laing said the finding was providing much excitement.

"Before we are born, we all have both skeletal muscle and heart actin in our skeletal muscles, but around the time of birth, we switch off the heart actin – and right now, it's a mystery why this happens," he said.

"We have long believed that if we could find out how to switch the heart actin back on in the skeletal muscle we could use this to create new

treatments for these devastating muscle diseases.

"What's remarkable is that these children's bodies have performed this 'switching on' process naturally, presumably to help counteract their condition, and if we can tap into the science of how they've managed to do that, we could perhaps use that information to help other affected children."

Children with these muscle diseases have no skeletal actin because of recessive mutations in the skeletal actin gene which "knock out" the gene function.

The fact that the mutations are recessive means that both the unaffected parents of the patients are carriers of the disease.

Professor Laing's laboratory was the first to discover mutations in the skeletal muscle actin gene which cause muscle diseases.

Source: Research Australia