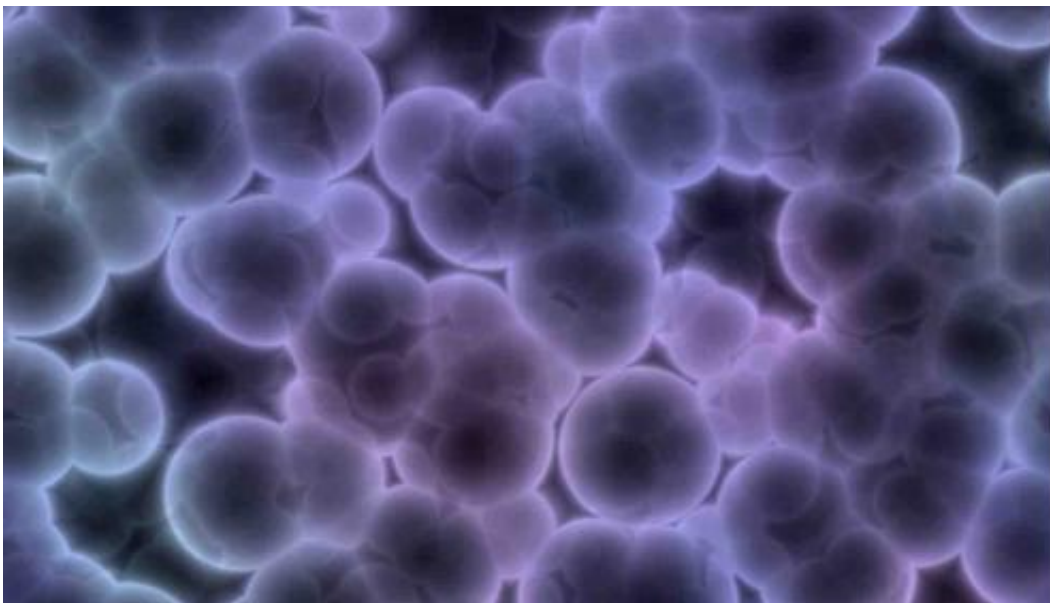


Identifying candidate treatments to delay the onset and progression of rare neurodegenerative disease

June 25 2014



A Victoria University of Wellington researcher is one step closer to identifying candidate treatments to delay the onset and progression of a fatal paediatric disease for which no effective therapy currently exists.

Work by Dr Andrew Munkacsi from Victoria's School of Biological Sciences and Centre for Biodiscovery, in collaboration with Professor Mengjie Zhang from the School of Engineering and Computer Science,

and Dr Stephen Sturley from Columbia University, is delivering new knowledge about the rare neurodegenerative disease, Niemann-Pick type C (NPC).

NPC is a monogenic disease caused by a defect in one of two different genes that affects approximately one in 150,000 children worldwide. Those affected are typically born without symptoms, but within a few years exhibit dementia similar to Alzheimer's disease and usually die before reaching adolescence.

Using exome sequencing, a strategy that selectively investigates important sequences of genetic material in all 23,000 human genes, Dr Munkacsı is analysing DNA samples from siblings in Australia, the United Kingdom and United States who have NPC disease to identify underlying disease gene mutation.

"Affected siblings, by inheritance, have the same mutation in the disease gene, but the onset and progression in the cohort we are studying is different. What we hope to do is identify genes associated with disease severity," says Dr Munkacsı.

Dr Munkacsı has been researching NPC for the past nine years. Through his investigations with Dr Sturley, using a yeast model of NPC disease, they have demonstrated that there are genes other than the disease-causing genes that modify [disease severity](#). This strategy has been successful, and has identified a drug that will be further tested in a human clinical trial in the United States.

They are now going to the next level and conducting the first genome-wide analysis of sibling pairs affected with NPC disease.

"Once we identify which genes regulate the onset and progression of NPC disease, we can work towards targeting those [genes](#) with drugs. Our

goal is to identify drugs already on the market as the children do not have the time to wait for new drugs to be developed and approved.

"As terrible as Alzheimer's disease is, at least persons affected live a healthy life for 60 to 80 years. Children affected with NPC disease deserve a chance to live a [healthy life](#)," says Dr Munkacsi.

Provided by Victoria University

Citation: Identifying candidate treatments to delay the onset and progression of rare neurodegenerative disease (2014, June 25) retrieved 19 April 2024 from <https://medicalxpress.com/news/2014-06-candidate-treatments-onset-rare-neurodegenerative.html>

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