Faulty gene linked to obesity in adults

August 18 2017, by David Stacey

Credit: University of Western Australia

Groundbreaking new research linking obesity and metabolic dysfunction to a problem in the energy generators in cells has been published by researchers from the Harry Perkins Institute of Medical Research and The University of Western Australia.

The study, published in *Science Advances*, found that reduced function of a gene that impacts the breakdown of fats resulted in adult-onset obesity and fatty liver.

Head of the Mitochondrial Medicine and Biology Laboratory at the Perkins, Professor Aleksandra Filipovska, said the study provided new knowledge that would enable the development of specific drugs and treatments to overcome the impact of obesity on normal body function.

"Our new model is helping us test drugs that can lessen the burden of obesity and fatty liver disease," Professor Filipovska said.

Lead researcher Kara Perks said that in normal metabolism fat and carbohydrate in our diets was broken down to produce the energy that our bodies required by the cellular energy plants, known as mitochondria.

"Our study showed that a fault in a mitochondrial gene had major impacts on normal metabolic function," Ms Perks said.

"If mitochondria are compromised or damaged, the breakdown of fat and carbohydrates is poorly regulated, which can lead to adult-onset obesity.

"We investigated the impact of mitochondrial dysfunction, in relation to a specific gene known as PTCD1, required for energy production and the breakdown of fats and carbohydrates.

"Healthy adults will have two copies of the PTCD1 gene, but we looked at what happens when there is only one copy, and we learned that PTCD1 is vital for the breakdown of fats and energy production. When one copy of this gene is lost, it results in obesity, fatty liver and ultimately heart disease.

"Our research also showed that PTCD1 affects the way that mitochondria are formed, impacting their shape and making them less connected with each other."

Excess weight and obesity are major risk factors for cardiovascular disease, diabetes, some musculoskeletal disease and cancers. The prevalence of obesity among Australians and most western societies has been increasing for the past 30 years, and this research offers useful

insight into one genetic cause of obesity.

More information: Kara L. Perks et al. Adult-onset obesity is triggered by impaired mitochondrial gene expression, *Science Advances* (2017). DOI: 10.1126/sciadv.1700677

Provided by University of Western Australia