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Media Statement

CLUE TO TREATING CHILDHOOD DISEASES

West Australian researchers have made a breakthrough which has the potential to lead to development of drugs to reverse effects of a devastating childhood disease.

Researchers at the Western Australian Institute of Medical Research (WAIMR) and the University of Western Australia have identified two new proteins that may be the key to treating mitochondrial gene defects known to cause severe and debilitating diseases, particularly in children.

Mitochondrial disease is estimated to affect one in 200 Australians, leaving them with symptoms including muscle fatigue, hearing loss, diabetes, heart disease and stroke.*

WAIMR group leader Dr Aleksandra Filipovska and her team had uncovered two new proteins that control mitochondrial gene expression.

“What we have discovered are two proteins – PCTD1 and PTCD3 – which together regulate how mitochondrial genes are activated in cells,” she said.

When genes are ‘turned on’ they make RNA (ribonucleic acid) which acts as instructions for the gene and initiates proteins to carry out important functions in the cell.

“Now we know these two proteins are what ‘turn on’ mitochondrial genes, they offer us a clue as to how we might be able to control mutated genes which cause mitochondrial disease,” Dr Filipovska said.

“The identification of these two proteins provides excellent targets for the development of drugs that might reverse the effects of mitochondrial gene defect diseases.”

Mitochondrial disease occurs when there is a mutation in the mitochondrial genome. Because the expression of mitochondrial genes is poorly understood, these diseases are not easily treated.

“Mitochondria are known as the powerhouses of the cell, and are responsible for producing 90 per cent of the energy needed by our body to sustain life,” Dr Filipovska said.

“There is currently no treatment for diseases caused by faulty mitochondria and so it is very exciting that this research may lead to treatments for these diseases which can be extremely devastating – even life-threatening, particularly in children.

“Faulty mitochondria have already been linked to Alzheimer's disease and type 2 diabetes, so there is a possibility that such treatments might also have broader applications in the future.”

The research was published in the August editions of *Nucleic Acids Research* and *FEBS Letters*.

The team's findings were made possible after postdoctoral scientist Dr Stefan Davies last year joined Dr Filipovska's laboratory from the University of Otago in New Zealand, bringing important expertise in mitochondrial biology.

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*Source: Australian Mitochondrial Disease Foundation website - <http://www.amdf.org.au/>(.)

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