

Discovery Profile



MitoCanada funds research to advance **Rapid Diagnosis** of Mitochondrial Disease

Reducing diagnostic timing from years to days
through generation sequencing

Research Project: **MITO-FIND**

Mitochondrial Functional and Integrative Next generation Diagnostics

Individuals with mitochondrial disease will see, on average, eight doctors before receiving a diagnosis. Traditional methods of diagnosis include painful and invasive muscle biopsies, the results of which can take months to come back to the clinician.

At the University of Calgary, Dr. Aneal Khan conducts leading edge mitochondrial research focused on rare and inherited metabolic and genetic disorders.

MitoCanada granted \$75,000 to Dr. Khan for the MITO-FIND project. These funds were also matched. This innovative research study was designed to implement a rapid process to diagnose mitochondrial disease using next generation sequencing. The funding was applied to perform gene sequencing on patients presenting either in hospital or in clinic with a suspected mitochondrial disease. This included mostly reagent costs but also labour to run the gene sequencing.

Dr. Khan developed sequencing methods to provide next-day diagnostic results from gene sequencing for

patients suspected of a mitochondrial disease. His methods eliminate the need for a muscle biopsy in the majority of patients and, in some cases, shorten the time to diagnosis from two years to two days.

These mitochondrial-targeted rapid sequencing methods can be applied to any sample from a simple cheek swab and provide results to anyone in the world.

Mitochondrial diseases have high morbidity and mortality rates. While the diagnostic task was made easier by the MITO-FIND project which sets a new standard for diagnosing mitochondrial disease, it is important to note that until a cure is found, we need to continue efforts to find effective treatments, improve access to diagnosis across Canada and help families living with mitochondrial disease. We need to understand how to improve lives of patients using existing drugs as well as developing new drugs.

“MitoCanada
Has been instrumental in
proving an opportunity to
advance the art of
diagnosis of mitochondrial
diseases in Canada.”

– Dr. Aneal Khan



Dr. Aneal Khan

Professor of Medical Genetics
and Pediatrics, University of
Calgary Cumming
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*Thank you supporters for your confidence in us.
Your donations allow us to support life-saving
researchers like Dr. Khan and
offer hope to those living with -- or at risk of
developing -- mitochondrial disease.*

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