

# Discovery Profile



## MitoCanada partners with MITO2i

fund mitochondrial research that could improve quality of life for those with **with autism spectrum disorder**

**Dr. Ingrid Tein** | *Director, Neurometabolic Clinic, Hospital for Sick Children*

*Identification of Candidate Genetic Susceptibility Variants in the Carnitine Transporter and Carnitine Biosynthesis Gene Families in Autism Spectrum Disorder: A Novel Precision Medicine Target*

### Autism spectrum disorder (ASD) occurs in 10-20 % of mitochondrial disorders.

ASD is a neurodevelopmental disorder that can cause significant social, communication and behavioural challenges. Those living with ASD have difficulty with social communication/interactions and may engage in repetitive patterns of behaviour.

ASD occurs in approximately 10-20 % of people affected by mitochondrial disorders. Genetic mutations that are important in neurodevelopmental genes can explain approximately 20 % of ASDs. Metabolic factors may also impact up to 10-20 % of children. This includes abnormalities in the mitochondria and carnitine (Cn)-dependent system.

L-Cn is a safe vitamin that is important in generating energy for the brain, protecting it from excessive free radicals, and helping neurotransmission.

It is often used to treat mitochondrial disorders. The brain has 3 Cn transporters. Dysfunction of one is associated with ADHD and ID. Certain children with ASD have had a positive response to Cn, particularly those with defects in the Cn pathway.

**The earlier we can identify and treat children at risk, the greater the effect on brain development and quality of life.**

Dr. Tein's research aims to identify genetic risk variants in the Cn transporter and Cn biosynthesis gene families. It also plans to identify clinical risk factors leading to Cn deficiency in a group of children with ASD. The approach will select children at risk for carnitine deficiency for potential future clinical trials in order to select those who may have a positive response to Cn. The hope is that this will lead to improved social communication, attention, and learning for a subgroup of children with ASD.

*Your donations allow us to support life-saving research and offer hope.*



mitocanada

*This research is a collaborative initiative funded jointly by MitoCanada and the Mitochondrial Innovation Initiative (MITO2i)*