Mitochondrial disorders (MDs) are the most common group of inherited metabolic multi-organ diseases. MD patients show exceptional clinical variability yet commonly present with neurological or muscular symptoms. Globally, MDs account for 1 in 2–5000 births, and treatment options are extremely limited. Diagnosis of MDs is both challenging and time consuming: it may start with genetic testing of a pathogenic mtDNA variant, and frequently necessitates many subsequent approaches such as neuroimaging, cardiac evaluation, genetic testing for a nuclear gene pathogenetic variant, and muscle biopsy to name a few. Thus, there is a significant unmet need for effective, specific, rapid and sensitive methods for diagnosing MDs in the clinic to determine if a patient has an increased risk for a mitochondrial disorder.

Translating a unique metabolic fingerprint into a powerful multi-biomarker

By studying patient cohorts with severe metabolic diseases, researchers at the University of Helsinki have discovered a subset of novel biomarkers that can be used for sensitive and rapid diagnosis of MDs in the clinic.

Blood metabolites as biomarkers for mitochondrial diseases: A readout of four distinct metabolites serves as a powerful multi-biomarker (sorbitol, alanine, cystathionine and myoinositol) that can distinguish patients with mitochondrial disorders from other groups.

Benefits:
✓ Rapid, easy and more accurate diagnosis in the clinic
✓ Enables tracking and follow up of disease progression over time
✓ More specific and personalised treatment options
✓ Achieve reproducible results with a kit akin to ELISA
✓ Bypass the need for an invasive muscle biopsy or genetic testing

Breakthrough Publication

Gen Tamjar
Commercialisation Officer
+358 50 377 3384
gen.tamjar@helsinki.fi