NON-CONFIDENTIAL SUMMARY

INVENTOR(S): **JOHN VINCENT**

CAMH TECHNOLOGY ID: 001-2009/002-2010

BUSINESS OPPORTUNITY

The Centre for Addiction and Mental Health is seeking partners to in-license these technologies, either individually or as a bundled portfolio to strengthen genetic diagnostics of intellectual disability.

Novel Diagnostic Marker for Intellectual Disability

Market Need

Intellectual disability (ID) is a devastating neurodevelopmental disorder with a profound impact on affected individuals and their families. The disease has a prevalence rate of about 2%, typically resulting from genetic mutations. ID may be present as the sole clinical feature (non-syndromic (NS)), or may be present with additional clinical or dysmorphological features (syndromic (S)). While autosomal recessive (AR) forms of NS-ID are believed to be more common, few genes have been identified so far. Thus identifying novel diagnostic markers for NS-ARID would be highly valuable for genetic testing. The genetic testing market is expected to reach $1.2 billion - $2.54 billion by 2016 with a CAGR of 7-9%, and the US and Canada hold the largest market segments of 30-33% and 13-15% respectively. To this end, the technologies described below will be of great benefit to this growing market.

Technology Description

Through homozygosity mapping and whole exome sequencing our scientists have discovered a nonsense mutation in the TRAPPC9 gene (Figure A). The truncating mutation in TRAPPC9 was co-published in the same issue of The Am. J. Human Genetics. Importantly, the TRAPPC9 findings have now been replicated and validated by six independent groups. Furthermore, mutations in TRAPPC9 in a non-consanguineous population have been found (Marangi et al. 2013). Our findings have been replicated in independent studies.

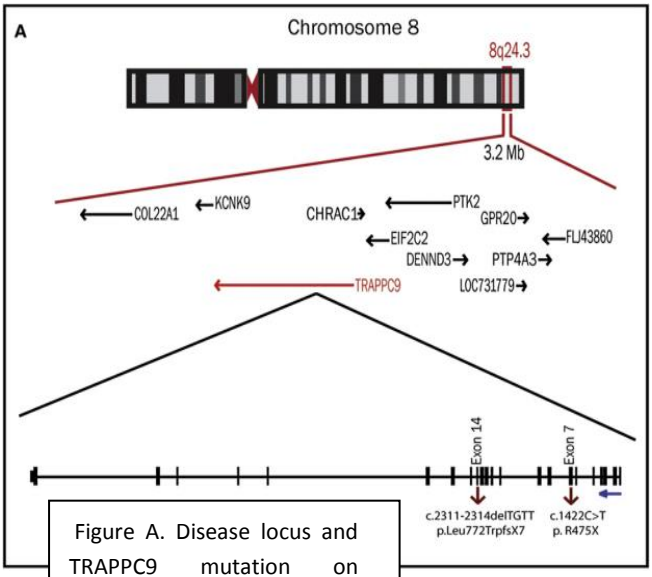

Figure A. Disease locus and TRAPPC9 mutation on chromosome 8q24. 23-q24.3, published in Mir *et al.* 2009.

Stage of Development

Whole exome sequencing, confirmation of mutations by Sanger sequencing, segregation analysis in families, and control populations have been analyzed, and mutations are not found. In the future, these markers will be highly valuable for genetic testing of intellectual disability and will have a positive impact on genetic counseling for families with intellectual disability. Lastly, investigators are examining the prevalence of the TRAPPC9 mutations in a Canadian cohort to determine the validity of these genes as diagnostic markers for ID in a Canadian outbreeding population.

Advantages

- Novel mutations can be used for ID diagnostics
- Predictive value of nearly 100%, allowing for the direct diagnosis of disease
- Specific mutations within novel genes for ID have not been associated with any phenotypes previously

Notable Publication(s)

Mir et al (2009) The American J of Hum Gen 85: 1-7

Khan et al (2012) The American J of Hum Gen 90: 856–863

Intellectual Property

TRAPPC9: Patent granted in US and applications filed in CA

FOR MORE INFORMATION CONTACT

Dr. Klara Vichnevetski
Director, Industry Partnerships & Technology Transfer
klara.vichnevetski@camh.ca
416 595-6056

Centre for Addiction and Mental Health
33 Russell Street, Suite 4039
Toronto, Ontario, Canada M5A 2S1
www.camh.ca