Market Need: Intellectual disability (ID) is a devastating neurodevelopmental disorder with a profound impact on affected individuals and their families. The disease, typically resulting from genetic mutations, has a prevalence rate of about 2%. Autosomal recessive (AR) forms of ID are believed to be more common but few genes have been identified so far. Thus, identifying novel diagnostic markers for NS-ARID would be highly valuable for newborn and predictive genetic testing. The genetic testing market is expected to reach upwards of $2.54 billion by 2016. 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 1) and a missense mutation in the NSUN2 gene. Importantly, the TRAPPC9 findings have now been replicated and validated by six independent groups. In addition, our investigators have just discovered mutations within four novel genes not previously associated with NS-ARID. One of these genes, FMN2 has been found in two consanguineous families affected with NS-ARID (Law et al. 2014). These markers would strengthen the genetic testing menu of a diagnostic company, and may support therapeutic interventions for intellectual disability.

Stage of Development:
- Investigators are currently examining the prevalence of the TRAPPC9 and NSUN2 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% allows for the direct diagnosis of disease.
- Mutations in TRAPPC9 in a non-consanguineous population have been found, as demonstrated in a recent publication by Marangi et al. 2013.
- Specific mutations within novel genes for ID have not been associated with any phenotypes previously

Notable Publication:

Intellectual Property:

Business Opportunity: CAMH is leading the commercialization of these technologies. We are seeking partners to in-license these technologies, either individually or as a bundled portfolio to strengthen genetic diagnostics of intellectual disability.