

New genetic test for cause of intellectual disability to be launched

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TORONTO - For the second time in as many months the Centre for Addiction and Mental Health (CAMH) has signed a licensing agreement with US-based Athena Diagnostics Inc. to market a new genetic test.

The test was developed by CAMH scientist Dr. John Vincent as well as independent clinical researcher Dr. Muhammad Ayub. It is based on their research into genetic causes of [intellectual disability](#). The test is being developed by Athena Diagnostics Inc. of Worcester, Massachusetts, part of publicly held Thermo Fisher Scientific Inc. CAMH and Athena Diagnostics signed a licensing agreement in January 2010 that could make the test available to physicians for clinical use in Canada, the US and Japan by January 2011.

The test analyzes a small sample of blood or saliva for the CC2D2A gene mutations that cause about 10% of cases involving a type of intellectual disability, similar to and including Joubert's syndrome. As an autosomal-recessive disorder, if both the mother and father carry a mutated copy of the gene, Joubert's syndrome can occur in one out of four of their children. The current research by Drs. Vincent and Ayub is based on findings in families where cousin-cousin marriage is common.

Intellectual disability affects about one in 100 individuals worldwide.

Genetic counselling for families

There are many types and causes of mental retardation or intellectual disability. It is estimated that up to half of all intellectual disability is caused by genetic mutation. Down syndrome is among those that are relatively straight-forward to diagnose through syndromes, or physical signs. In contrast, non-syndromic types of intellectual disability do not have physical signs, and so are more difficult to identify.

Genetic counsellors will assist physicians to interpret the results for each client and family. "Many affected families are eager for information that will help them understand the cause of the condition, as well as the risk of passing the disorder on to their offspring," adds Dr. Vincent. "Our test will provide more accuracy to physicians who are trying to identify these hard-to-diagnose cases, and help them offer appropriate genetic counselling to family members who request it."

Dr. Vincent is a scientist at CAMH whose research focuses on the genetic causes of inherited diseases, including autism and intellectual disability as well as bipolar affective disorder.

Licensing partner

"This is the second such licensing agreement with Athena Diagnostics that CAMH has signed within a few weeks. It is an exciting time for CAMH to partner with Athena Diagnostics in bringing new tools to the clinicians who are helping clients and their families learn more about the risk factors associated with inherited conditions," says Dr. Bruce G. Pollock, VP of Research at CAMH.

"Dr. Vincent has a remarkable research and publications record in this area. We are proud of his work and of the support that the CAMH Research Program is bringing to the individuals who have, or are at risk of having, inherited conditions," Dr. Pollock adds.

The cost of the test - which will be available only through clinical services - has yet to be determined. The original research was supported by funds from the Ontario Ministry of Research and Innovation.

Provided by Centre for Addiction and Mental Health

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