Ways in which translation of genetic discoveries into diagnostic strategies can benefit individuals with ASD and their families

1. Earlier detection of risk for ASD and related conditions
2. Better understanding of the specific causes of autism with implications for medical management and genetic counseling
3. Targeted individualized approach to ASD treatment
4. Development of drugs/treatments based on new gene/protein targets

Autism Discoveries in the News

THE GLOBE AND MAIL
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Canadian breakthrough offers hope on autism

Lee Marshall, left, and his brother Eric, centre, each has a form of autism. They are pictured with their father, Craig, mother Marie and brother Marc at their Burlington, Ont., home yesterday.

Project makes possible DNA test to identify children most likely at risk to condition

BY CAROLYN ABRAHAM

I don't think it's inconceivable that we're going to be able to prevent autism down the road. The clinical implications of this discovery are unprecedented.

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THE GLOBE AND MAIL
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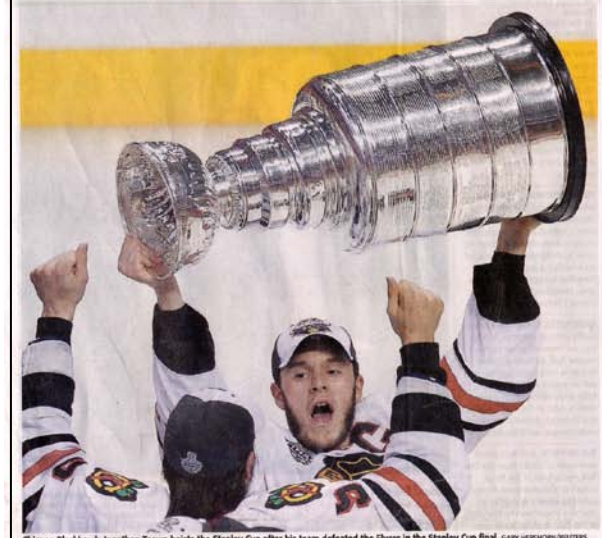
STANLEY CUP PLAYOFFS

Blackhawks break 49-year drought in OT



Chicago Blackhawk Jonathan Toews hoists the Stanley Cup after his team defeated the Flyers in the Stanley Cup final. GARY HERSHORN/REUTERS

Full coverage in GLOBE SPORTS ⟲

MEDICAL SCIENCE

Genetic finding paves way for controversial autism testing

Canadian discovery makes prenatal scan possible, prompting debate over who should have access to the technology

BY CAROLYN ABRAHAM TORONTO

An international effort led by Canadian scientists has uncovered the complex genetic architecture of autism, revealing brand new targets for treatment and making it possible to predict with a DNA test at birth, or even before, about 10 per cent of those who will develop the condition.

But precisely how or when people should have access to such a test could prove to be nearly as tricky as the disorder itself, now seen as the most common serious developmental condition of childhood.

After sifting through the DNA of 1,500 families, members of the Autism Genome Project, a consortium of 120 researchers in 11 countries, have made the humbling discovery that the genetic risk factors for autism are different for each person who suffers from it.

"I highly doubt you will find two families with the same combination of genetic variants," said study leader Stephen Scherer, senior scientist at the Hospital for Sick Children in Toronto.

Autism encompasses a spectrum of lifelong neurological disorders that can vary widely in severity and symptoms. Researchers agree an early diagnostic test could be crucial to countering the condition's worst effects, but also acknowledge it could be used in family planning, or as a prenatal test in which parents may opt to terminate a pregnancy.

Researchers however caution it is too early to use a genome scan to routinely diagnose autism until they can figure out how the genetic glitches they have discovered play out in real life. Their paper, released Wednesday in an online edition of Nature, says that the roots of autism involve dozens of genes fouled up by long stretches of missing or duplicated pieces of DNA. But which genes and which stretches differ from person to person.

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Researcher expects mail-order test soon, even if results will be dubious

To learn more about the genetic quirks they have found, Dr. Scherer, director of the Centre for Applied Genomics, has received $8-million from the Ontario government to run a DNA scan on nearly every child diagnosed with autism in the province over the next three years, expected to be more than 5,000 children. Researchers in the United States and Britain have similar plans, all of it with an eye to developing diagnostic tests.

But in the direct-to-consumer age, the market rarely waits for more research. Dr. Scherer expects people will be able to buy a mail-order test that scans for autism-related genes in the near future – even if the information they can glean from it is dubious at best.

> ***The quicker we get to these kids the better. ... There's that critical time between the ages of 2 and 5.***
>
> Lisa Bond, a mother of two children with ASD

"I get e-mails from U.S. companies every month, start-ups and diagnostic companies, looking for markers for autism," he said, noting the findings for this study are not protected by patent.

In this study, approximately 10 per cent of the 1,000 people with autism tested had mutations in genes known to affect brain function. For this reason, Dr. Scherer believes certain variations within these genes are predictive of an autism disorder.

Still, if families came for testing now, "in the majority of cases we won't be able to tell them anything," he said.

Study co-author Peter Szatmari, director of the Offord Centre for Child Studies at McMaster Children's Hospital in Hamilton, said the prospect of a commercial test "makes me nervous. I don't think we have the science yet to nail it down. ... This isn't one gene, but a profile of genes, a pattern of susceptibility, not cause."

Autism spectrum disorders (ASD) affect one in every 110 children. Some face such severe cognitive impairment they're unable to speak. Others are savants. Most show a preference for rigid routines and repetitive behaviours. But common to all are social deficits that hamper the ability to interact with others.

Currently, children generally aren't diagnosed until age 4 or 5, after a battery of psychological tests. A genetic test could allow parents to intervene with behavioural therapies in infancy. It could also shed light on their chances of having another child with autism, and their children's chances of having an affected child.

Lisa Bond, a single mother of two in Campbellford, Ont., waited years to learn both her children had autism spectrum disorders. Her son, now 14, was 6 when he was diagnosed.

"It was a nightmare for me, constantly going to doctors," she said. "Now they realize the quicker we get to these kids the better. ... There's that critical time between the ages of 2 and 5."

As part of the study, Ms. Bond learned her son has a region of chromosome 16 deleted, which helps to explain why he also has trouble with his spine, walking and swallowing.

Ms. Bond's eldest child Rebecca, now 17, was diagnosed at 12 with Asperger's syndrome, a high-functioning form of autism characterized by normal-to-high intelligence, striking talents and obsessive interests (in Rebecca's case, dinosaur teeth). But she does not carry the same genetic mutation as her younger brother.

Scientists used the latest microchip tools to scan the DNA of nearly 1,000 people with autism and 1,200 controls. Such scans are not designed to pick up specific mutations, but rather cover the whole genome and highlight areas where large chunks of DNA are deleted or repeated like a record skipping.

These types of hiccup mutations are known as copy-number variants, or CNVs. Researchers found that people with autism have more CNVs in their genes than controls.

In all, the study, spearheaded by post-doctoral research fellow Dalila Pinto at Sick Kids, identified more than 100 genes affected in the people with autism, many of them forming part of a network that governs how brain cells grow and talk to each other.

Age of Autism
Daily Web Newspaper of the Autism Epidemic

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June 13, 2010

Scherer of Nature Autism Gene Study Fails to Disclose Pharma Funding As Competing Interest

Senior author in *Nature* autism-gene study fails to disclose funding by MMR manufacturer GlaxoSmithKline as a competing interest.

By John Stone

Prof Stephen Scherer who is the senior author of the autism gene study launched in *Nature* last week holds the 'GlaxoSmithKline-CIHR Pathfinder Chair in Genetics and Genomics at the Hospital for Sick Children and University of Toronto. The title used to be 'GlaxoSmithKline-CIHR Endowed Chair", GSK being one of the defendant companies in the UK MMR litigation.

While this information was tucked away in the paper under acknowledgements it did not appear as a competing interest for Prof Scherer, and was not mentioned in the paper's extensive media publicity. The study boasts a remarkable 176 authors, including Prof Sir Michael Rutter and Prof Eric Fombonne who have given evidence for the vaccine manufacturers, and the US Department of Justice in vaccine litigation, but the disclosures of competing interests amount to barely three lines and do not mention any of these things:

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