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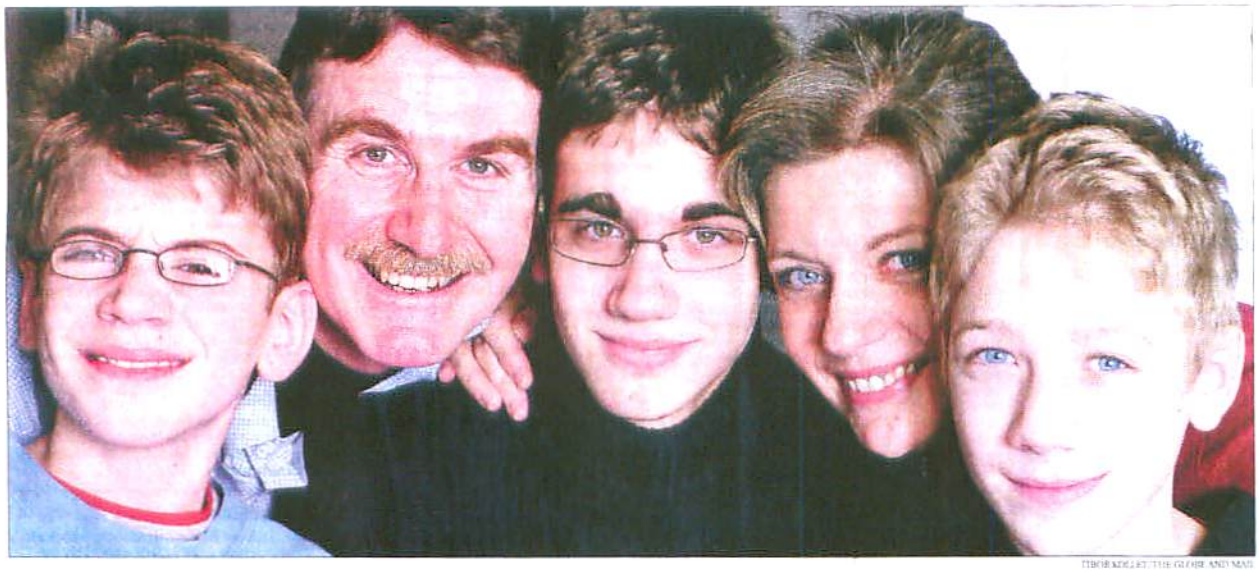
Canadian breakthrough offers hope on autism



TIBOR KOLLEY/THE GLOBE AND MAIL

Luc 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

A massive international effort led by Canadian scientists has homed in on the genes behind autism – a breakthrough that could revolutionize how the mysterious and surprisingly common condition is both detected and treated.

Touting it as the most significant advance in the field in 30 years, researchers say the landmark project has put within reach a DNA test to identify children with autism early enough to counter the condition's worst effects.

"I don't think it's inconceivable that we're going to be able to prevent autism down the road," said study leader Peter Szatmari, director of the Offord Centre for Child Studies at McMaster Children's Hospital in Hamilton. "The clinical implications of this discovery are unprecedented."

Doctors currently rely on psychological tests to diagnose autism spectrum disorders in children at age 2 or 3. But a DNA test could identify those affected as babies, or perhaps even before they are born.

The findings, based on the largest autism DNA collection ever assembled, could also allow parents who have children with autism to learn through genetic screening their chances of having another affected child.

"If you know ahead [of time] of your predisposition to autism, you can make an informed decision," said Marie Folicoeur, a Burlington, Ont., mother who has two sons with autism disorders and whose family contributed DNA to the project.

Using new genome scanning tools, researchers have found that several different autism-related genes can play a role in different families. This helps to explain why no two children – not even identical twins – have identical symptoms.

The researchers have pinpointed at least five areas of the genome that harbour genes linked to autism susceptibility, including those crucial for brain function. They have also found a genetic mutation tied to the disorder in girls – who are four times less likely than boys to develop autism disorders.

The work has also highlighted how autism can spring from genetic quirks not seen in either parent – suggesting that a genetic glitch has randomly emerged in the sperm or egg cells of the father or mother prior to conception.

Co-author Steve Scherer, senior scientist of genetics and genomic biology at Toronto's Hospital for Sick Children, said, "It may be that 5 to 10 per cent of autism cases are arising from these de novo [new] mutations."

The research, released yesterday in an advance online publication of the journal Nature Genetics, is the first part of a two-phase study run by the Autism Genome Project. It involves more than 137 researchers from 50 academic institutions in eight countries and the study of nearly 8,000 people from 1,600 families who have at least two members diagnosed with an ASD.

Dr. Szatmari, who set the ground rules for the unprecedented collaboration that began in 2002, said "the effort has meant the putting aside of individual ambitions to work together as a team."

Autism disorders have only recently been recognized as the most common serious developmental condition of childhood, affecting roughly one in 165 children. Experts refer to it as a spectrum because the complex neurological condition can range so widely in severity.

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PETER SZATMARI,
STUDY LEADER AND DIRECTOR OF THE OFFORD CENTRE FOR CHILD STUDIES AT McMASTER CHILDREN'S HOSPITAL IN HAMILTON, ONT.