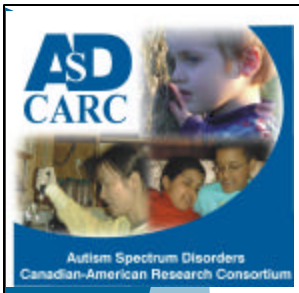

Unraveling the Mystery of Autism: From Genotyping and Phenotyping to Prospective Identification and Prevention

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www.autismresearch.ca or 1-866-ASD-CARC

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A Message from Dr. Jeanette Holden

This Newsletter contains some updates on our ASD Research program – we hope you enjoy reading it.

For those of you participating in our Research, we hope you'll share the extra copy with your friends & members of autism groups to which you belong.

If you find participating in our research interesting, please encourage others to check out our website.

Genetics Studies: We are contacting all families to let you know about our Genetics studies, and how you can take your own "cheek swab" samples to send us for the DNA studies. Details inside.

Prospective Study: During our calls, some families have let us know of a "new addition" to their families – Please keep this study in mind if you are planning on extending your family, or know of friends who are!

On-Line Research: We now have 8 studies on-line, varying from those that help us standardize the diagnosis of individuals with ASDs to studies on children, siblings and their parents. We will be sending emails to all families as we add new studies, and hope that you will be able to complete these relatively short studies as time permits.

Fragile X and Autism Spectrum Disorders Conference: Please see enclosed information about our conference in July, here in beautiful Kingston, Ontario – near the Thousand Islands. This is not only an informative Conference for families, educators and other caregivers, but also a fun time for meeting other families.

Student's Corner: We have introduced this new section, which will feature short articles from students working with one of our ASD-CARC faculty.

Family Corner: Remember that you can spark our interest in new projects by submitting comments to our Family Corner.

Sincerely,

Jeanette Holden

PROSPECTIVE STUDY OF INFANTS AT-RISK FOR ASD

Do you have one or more children with ASD and an infant under 12 months of age, or an infant on the way? Please read about our Prospective Study and how you can help.

The chance of a sibling developing an ASD is higher than in the general population (6% compared to .05%). Therefore, it is expected that a small percentage of infants with older affected siblings will develop an ASD. As a family with one or more children with ASD, parents are already watchful or concerned about their infant's development and may appreciate having a professional who can help them track their infant. Through involvement in the Prospective Study, parents can discuss with us any concerns they have during their infant's first three years and will receive suggestions around stimulating infant development and support regarding assessments and intervention services, if necessary.

Participating families are completing monthly developmental checklists and, every three months, are being interviewed by our Clinical Research Coordinator who completes standardized assessments and discusses any developmental concerns parents have about their infant. Where possible, families are also videotaping their infant's development to help detect early signs of ASD should they occur. Some families are invited to participate in some of our lab-based studies looking at early neurophysiological and social-communicative markers of ASD.

To learn more about this research or sign up to participate, please contact Dr. Becky Ward or Heidi Penning at autism@post.queensu.ca or 1-866-ASD-CARC.

RESEARCH REGISTRY
Jeanette Holden, Project Director

It is clear that not all cases of ASDs are the same, and that there are many subgroups of ASDs, each with specific behavioural and clinical features. Therefore, a very large number of families are needed to identify common features and to subgroup families for genetic studies.

Since its inception in August 2001, more than 600 families have joined our Research Registry, with several coming on board every week. We hope to have literally thousands of families completing our on-line questionnaires, so that we can have a very broad picture of how ASDs affect families and what characteristics may be common to subgroups of families. ***So, please invite other families you know to check out our website and participate in our studies.***

Our Research Registry families have become an integral part of our research program. They give freely of their time by participating in a variety of research projects. From time to time we receive notes of encouragement from our families, often including pictures of their children with ASD.

Below is a description of questionnaires we currently have on-line. ***To maximize our possibilities for subgrouping families, we need to have you complete all the questionnaires at your earliest convenience, regardless of whether your child has the problems or not.***

Our research team draws inspiration from all of you. We are deeply grateful and hope that your efforts will lead us to some answers about ASDs soon!

Current On-Line Questionnaires

- **Pervasive Developmental Disorder – Behaviour Inventory (PDD-BI) (20-30 minutes):** This is perhaps the most crucial questionnaire and helps us subgroup families according to specific characteristics in your affected child(ren). ***To be completed for each child with an ASD.***
- **Diagnostic Information (~ 5 minutes):** This brief questionnaire asks about both recent and initial diagnoses, and developmental disorders. ***To be completed for each child with an ASD.***
- **Sleep Disorders Survey (BEDS) (15-20 minutes):** Sleep disorders are common in persons with ASDs. This questionnaire deals with issues such as length of sleep, difficult sleep issues, behaviours during sleep or at bedtime, etc. ***To be completed for all of your children .***
- **Obsessive-Compulsive Core Dimensions Questionnaire (OCCDQ-T) (~10 minutes):** Certain aspects of personality, emotions and motivations, are present to some extent in all people. ***For parents, adult siblings, and grandparents, when possible.***
- **Autism Quotient (AQ) (15-20 minutes):** Family members often report having characteristics that resemble the autistic traits in their affected child(ren). This questionnaire deals with issues such as social skills, attention switching, attention to detail, communication, and imagination. ***For parents, adult siblings, and grandparents, when possible.***
- **Handedness Test: (10-20 minutes)** One way of assessing how the brain has developed is to test for handedness. A parent asks the child/spouse to carry out several motor activities, such as picking up a raisin or throwing a ball, and records which hand the child or spouse used in each task. A great fun activity on cold or rainy days! ***For all family members.***
- **Social Interaction Subtypes (~5 minutes):** Parents are asked to classify their children into one of three social subtypes. ***To be completed for all of your children .***
- **Prenatal Factors and ASD (~15 minutes):** This questionnaire asks about stress, medications and infections during each pregnancy. Even if there were no complications, we need you to complete this questionnaire to get an idea of how frequent such issues occur. ***To be completed for all of your pregnancies .***

COMING SOON!

- **Life-time Anxiety and Mood Disorders Screening Questionnaire:** This questionnaire is about parental issues related to depression and anxiety.
- **Perspective Taking Assessment:** A brief informal assessment for parents to complete on their affected children to assess challenges in their ability to take someone else's perspective.
- **Gastrointestinal Disorders:** This asks about GI functioning and dietary habits.

A family testing for 'Handedness'

FROM OUR STUDENTS
Each issue will feature a student's "story"

Sonia Robitaille, MSc candidate working at Queen's University with Drs. Jeanette Holden and Doug Munoz

Autism was unknown to me until I babysat a young boy with the disorder. I remember that pictures of his favourite foods were stuck on the refrigerator and when he wanted something to eat, he was encouraged to point at the pictures and make eye contact. I realized that for most of us, it is so easy to communicate but for individuals with autism, it is not always so simple. From then on, I was intrigued by how life was different for them and always wanted to know more. Then came the opportunity to do just that. Three years ago, I started to do research on the genetics of autism with Dr. Holden.

The main objective of my research is to determine whether a gene might be involved in autism. Two genes were chosen, the homeobox A1 (HOXA1) gene and the brain-derived neurotrophic factor (BDNF) genes. Why did I choose these genes? In a group of individuals exposed to thalidomide, a drug administered to pregnant women to treat morning sickness, and now known to cause abnormalities in the fetus, there was an increased number of individuals with autism (at least 30 times higher than expected). Interestingly, these individuals had malformations of the external ear and some form of facial paralysis, similar to mice lacking the HOXA1 gene. It was then proposed that the HOXA1 gene may not work properly in some individuals with autism, and causing ear abnormalities and facial paralysis.

As for BDNF, it has been found that, at birth, some individuals with autism had elevated levels of BDNF in their blood. It was thus proposed that the high levels of BDNF in individuals with autism may be due to a malfunction of the BDNF gene.

Based on my research results, the HOXA1 and BDNF genes differ in persons with autism compared to controls. However, further studies are needed to determine whether these genes are directly involved in causing ear abnormalities (HOXA1) or overproduction of BDNF.

Through my work, I feel that I have contributed, in a small way, to knowledge about autism. I intend to continue working in this field since I believe that our genes hold answers to many of our questions about autism and with technology advancing so fast, the answers may be closer than we think.

Genetics Kits

In the Fall of 2002, ASD-CARC staff began calling families in the Research Registry about participation in the Genetics Research Project.

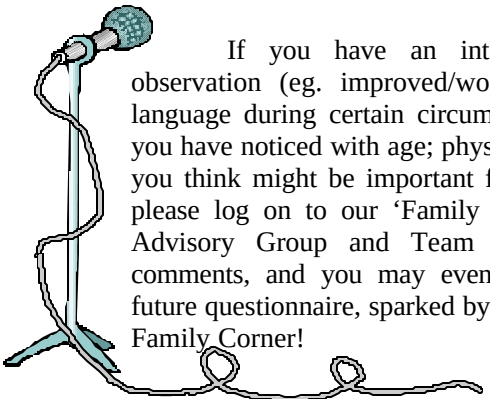
Once a family has agreed to participate, they are sent a Genetics Kit in the mail for cheek swab sample collection. All family members are invited to participate, including parents, all children, and grandparents. The collection is quick and easy and all required materials are provided with the Genetics Kit, including written and video instructions.

Cheek cells are collected from each individual by taking a small brush and rubbing it along the inside of the cheek. The brush is then swirled inside one of the sample collection tubes. The collection tubes are then placed inside the stamped return envelope along with signed consent forms for each family member.

We have contacted more than 300 families and the rest will be contacted in the next few months, and we look forward to speaking with you. We are thrilled by the overwhelming support and enthusiasm for our research from our participating families.

Some of the materials that are in a Genetics Kit. One mother remarked that she and her family felt like "real scientists" as they collected the cheek swab samples!

Family Corner



If you have an interesting anecdote or observation (eg. improved/worsened behaviours or language during certain circumstances; changes that you have noticed with age; physical features; etc.) that you think might be important for us to know about, please log on to our 'Family Corner'! Our Parent Advisory Group and Team members review all comments, and you may even see your ideas in a future questionnaire, sparked by your comments at our Family Corner!

ASD-CARC

(Autism Spectrum Disorders—Canadian American Research Consortium)

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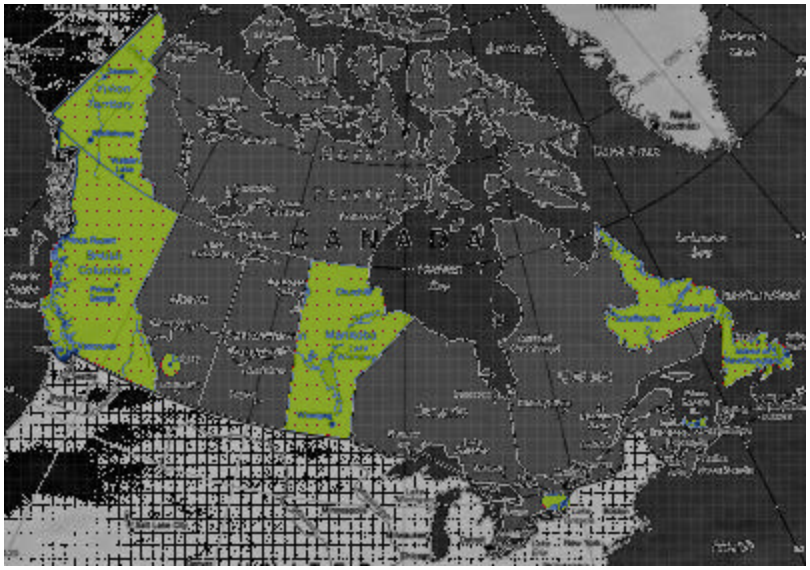
EPIDEMIOLOGY OF ASDs IN CANADA

Hélène Ouellette-Kuntz, Project Director

Information about cases of ASDs is being collected from agencies and schools in different regions of Canada, including BC and Yukon, Calgary Region, Manitoba, Southeastern Ontario, PEI, and Newfoundland & Labrador (see map— dark blue shaded areas are currently included in study). To date, over 1,900 cases have been identified.



Stay tuned for an update regarding the prevalence and recurrence in families in our next newsletter!

To learn more about this research or to sign up to participate, please contact Hélène Ouellette-Kuntz at oullette@post.queensu.ca or call 613-548-4417 X1198

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