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The latest in autism research

By Kelly Dean Schwartz, PhD RPsych, Institute of Marriage and Family Canada Research Fellow

October is Autism Awareness Month and Institute of Marriage and Family Canada Research Fellow Kelly Schwartz attended Autism Research Training (ART) in late August, in Montreal. The following is an overview of the coordinated attempts of a group of Canadian researchers to understand the nature and process of autism.

Autism spectrum disorders (ASD) are a complex group of conditions characterized by impairments in shared social interaction and communication, and the presence of restricted and repetitive behaviours. It is estimated that 50,000 Canadian children and adults are affected by this family of disorders, and researchers and practitioners (physicians, teachers, psychologists) are keen to understand more about this pervasive developmental condition at all levels.

What is ART?

Autism Research Training (ART) was held August 23-27, 2010 in Montreal, Quebec. The goal of the program is “to recruit and train outstanding researchers in the field of autism, in disciplines such as genetics, brain imaging, epidemiology, neurology, psychiatry, psychology, and others.” (ART Program website, 2010)

Three areas presented at the ART Program Summer School—epidemiology, genetics, and intervention—are briefly reviewed below.

Epidemiology

Popular opinion is that autism is affecting more and more individuals than ever before. Psychiatrist and researcher Dr. Eric Fombonne of McGill University has made significant contributions in epidemiology—the study of the distribution of diseases in human populations. Fombonne states that one of the greatest sources of confusion for autism is the misunderstanding between studies focusing on prevalence (the proportion of people affected by autism at one point in time) versus incidence (the proportion of people who develop autism over a period or new cases). Case in point, in 1965 it was estimated that approximately 4/10,000 individuals were diagnosed with autistic disorder. Jump ahead to 2001 (Chakrabarti & Fombonne, 2001) and the prevalence rate is closer to 61/10,000.

Why this perceived increase? Fombonne states that it is partly due to how inclusive studies are in what defines a “case” of autism. Studies that only include a sample of individuals diagnosed with Autistic

Disorder (the most severe form of the disorder) will normally report lower estimates than those who estimate the prevalence of the more robust Pervasive Developmental Disorder. That considered, the best global estimates are that about 1/140 will be diagnosed with autism disorder, Asperger's Syndrome, childhood disintegrative disorder, or Pervasive Development Disorder—Not Otherwise Specified.

While far from an epidemic in terms of increasing prevalence rates, Fombonne indicated that an increase in the incidence (new cases) of autism is more likely the result of better diagnostic criteria and improved awareness and services available for this identified population. Popular opinion would lead one to believe prevalence rates are higher than they are, but it is likely that autism and other related neurodevelopmental disorders are simply receiving more press because of controversial treatments (e.g., hyperbaric chambers, sensory rooms) and much heated debate over its origins, including MMR vaccines and other environmental toxins. The topic of genetics has also been the focus of much discussion recently, and this too was an area explored at the ART gathering.

Genetics

By far the most active area of research in autism is in the area of genetics. Dr. Andrew Patterson, senior scientist and Canada Research Chair in Genetics of Complex Disease at Toronto's Hospital for Sick Children presented current perspectives on just how far we have come – and how far we have yet to go – in establishing reliable links between genetics and autism spectrum disorder. The main goal of genetics research in this area is to explore the molecular contributions of genetic risk factor or mutations that are responsible for understanding the pathophysiology of autism.

The main findings? There are a few twin and family studies that indicate a high heritability (greater than 90 per cent) for a small number of cases. This translates into a small proportion of overall familial risk. Patterson qualifies these modest results by stating that many (if not most) of the studies on which this high heritability is cited do not show much consistency in the gene site amongst twins or families, are often based on small sample sizes, and rarely if ever consider that the environment is greatly implicated in the expression of these genetic markers. This means that even if a series of gene markers were consistently identified in robust samples of individuals with autism, there remains immense phenotypic (i.e., how the genes are expressed behaviourally) heterogeneity that is not explained by isolating such genetic information. Although there is a lot of study in the area of the genetic origins of autism, the practical application of such data for identification and intervention is still in its infancy.

Intervention

If genetics is the most active area of research, exploring the current status of interventions for people with ASD is by far the most controversial. For families dealing with a child diagnosed with autism, choosing amongst the myriad of intervention strategies is as confusing as the disorder itself. Families and governments invest millions of dollars every year in treatments to ameliorate the symptoms of autism, the result of which many are left wondering if such treatment has a measurable impact or not. Dr. Pat Mirenda is a professor in Educational and Counseling Psychology and Special Education at the University of British Columbia and a Board Certified Behavior Analyst, and she presented the findings from a comprehensive meta-analysis of 775 studies (National Standards Report, 2009) exploring the effects of established, emerging, unestablished, and ineffective or harmful treatments.

Mirenda summarized the complex findings: The most effective treatments for autism involve early intervention strategies like behavioural treatment, joint attention intervention, and pivotal response treatments, as well as other behaviour regulation and instruction. Emerging treatments, which are those treatments with less well-established records of evidence-based support, included language training, augmentative and alternative communication (known as AAC, of which the Picture Exchange Communication System or PECS is most well known), and social or theory of mind skills training.

Most notable by its absence from either the established or emerging treatment paradigms are some that are most popular and (likely) most expensive, including auditory integration training, facilitated communication, gluten and casein-free diets, and sensory integration training. The last of these treatments – sensory integration training that includes practices such as deep pressure or weighted vests – has gained the most notoriety in medical and education settings, and yet Miranda notes that there is no empirical evidence to support the validity of such treatments for individuals with autism. This would include the Snoezelen multi-sensory environment therapy rooms that inhabit many homes, schools, and hospitals, the likes of which have yet to be established as a valid, evidence-based treatment for children with autism. Miranda notes that there continues to be a wide gap between best practice and empirical research in sensory and other treatments. This includes pharmacological, biomedical (for example hyperbaric oxygen), and alternative (e.g., equine therapy, rhythmic entrainment, etc.) treatments.

Helping parents discern which treatments are supported by rigorous and evidence-based research would be of great benefit as they navigate these emotional and desperate treatment decisions, as there are many treatments that show little or no benefit to children and families

Conclusions

The ART Program is to be applauded for its mentoring efforts in developing the next generation of researchers and practitioners to become highly productive, ethically responsible independent investigators. The challenge continues to be how we can most effectively disseminate this growing wealth of information down to the “front lines”—parents, educators, and even the children themselves—in short, those who are most directly impacted by this complex and pervasive neurodevelopmental disorder. Canadians can take heart in knowing that we have some of the world’s best researchers who have devoted their lives to ensuring we are making informed and evidence-based decisions for children and adults with autism.

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Notable resources

[Autism Research Training Program](#)
[National Autism Center Standards Report](#)