Aspect Research Update 2013

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Welcome to the 2013 Aspect Research Update. Each year Aspect reviews international and local research into autism spectrum disorder (ASD). In this research update, we highlight research into the prenatal influences and genetic causes of ASD, review the effectiveness of new technologies and look at service provisions for adolescents and ageing adults.

The diagnostic classification for ASD changed in May this year with the release of the Diagnostic and Statistical Manual-5 (DSM-5) (APA, 2013). The most notable feature of the new criteria for ASD was the deletion of the term Asperger’s Syndrome. The new diagnostic criteria for ASD are classified in DSM-5 by two domains of impairment (1) social communication and interaction, (2) restricted, repetitive patterns of behaviour, interests or activities. For an ASD diagnosis to be made these symptoms need to be evident from childhood, and impair daily functioning.

There has been some discussion about how these changes to the diagnostic criteria will affect the support entitlements for individuals who were diagnosed with Asperger’s Syndrome under DSM-IV (APA. Diagnostic and Statistical Manual of Mental Disorders IV-TR, 2000). At this point in time it is too early to state whether these changes to the diagnostic criteria will impact on individuals who are currently receiving support for Asperger’s Syndrome (Buxbaum & Baron-Cohen, 2013; Gibbs, Aldridge, Chandler, Witzlsperger, & Smith, 2012).

Early diagnosis is the key to implementing interventions and support. Currently diagnosis is possible from about two years of age through a variety of screening tools which use parental reports, and assessments of communication, social and behavioural capabilities (Baio, 2012; Barbaro & Dissanayake, 2012; Ozonoff, Iosif, & Baguio, 2010).

The aim is to develop screening and diagnostic tools that can be applied earlier. The progress of the mapping of the human genome is providing valuable genetic information about the DNA of ASD. Our understanding of how an identified change in the genetic make-up of individuals with ASD influences behaviour is still limited. Each year as our understanding of the interactions of these genetic variations improves, the researchers move us ever closer to more effective interventions and treatments.

1. Genetics
This year one research group examined the entire DNA code of 32 families with at least one member who has ASD, and compared them to families who do not have members with ASD (Jiang, et al., 2013). The study reported that the researchers identified significant genetic risk variants in 50% of the families who have members with ASD. These genetic variants were not present in the DNA code of more than 1000 non-ASD individuals whose genetic information was available on a public database. These results
provide a promising potential for the development of clinical tests that could identify individuals who have specific genetic mutations.

Researchers have suggested that in the future ASD may be diagnosed through a blood test in children as young as one-year-old (Chow, et al., 2012). Professor Eric Courchesne reported at the Asia-Pacific Autism Conference 2013, that his research team had identified specific gene networks that can disrupt brain growth in babies. The identification of this ‘genetic signature’ of autism, the researchers suggest, could lead to earlier diagnosis and subsequently more targeted treatments.

A research group in the University of North Carolina have investigated the effect of a group of enzymes known as topoisomerase inhibitors on the development of ASD. Topoisomerase inhibiting chemicals are found in some chemotherapy and antibiotic drugs. The researchers suggest that topoisomerase inhibitors have the potential to cause mutations to genes that have been associated with ASD (King, et al., 2013). The benefit of the identification of the potential effects of these enzymes is that it may offer pregnant women the opportunity to avoid exposure to these chemicals.

The ability to identify specific genes that are associated with particular disorders may also make the process of ASD diagnosis more reliable. Researchers have suggested that children with specific genetic disorders (such as 22q11.2 deletion syndrome) may be misdiagnosed as having ASD (Angkustsiri, Goodlin-Jones, Brahmbhatt, Harris, & Simon, 2013). In a study by Angkustsiri, et al. (2013) researchers found that in the sample of 29 children who were diagnosed with 22q11.2 deletion syndrome, none of these children met the strict diagnosis criteria for ASD. These findings are important because without a correct diagnosis, appropriate interventions may not be provided.

Researchers have also identified genetic factors that are linked to neurodevelopmental and neuropsychiatric disorders. In the Cristino, et al. (2013) study the researchers used publicly available molecular data to analyse the DNA variations of 700 genes for ASD, attention deficit hyperactivity disorder (ADHD), schizophrenia and X-linked (male) intellectual disability. The results from this study identified a common genetic variant influence in each of these disorders (Cristino, et al., 2013). The researchers hypothesise that this finding will lead to new diagnostic tools that will be sensitive enough to assess an individual’s genetic health risk and ultimately provide specific therapeutic strategies for these disorders.
2. ASD Diagnostics and Interventions
Diagnosis is typically made using the Diagnostic and Statistical Manual-5 (DSM-5) criteria (APA, 2013). The DSM-5 criterion relies upon a clinician’s assessment of communication and behavioural competencies. Researchers are investigating new ways of diagnosing ASD using technology. Recently, a research group has reported that they have been able to identify specific electrical brain activity in people with ASD that was significantly different to the control group (Dominguez, Velazquez, & Galan, 2013). The researchers compared nine male children aged between seven and 16 years with ASD, and 10 control children aged between seven and 16 years (six males, four females). The results showed that using a magnetoencephalogram (MEG) the researchers were able to detect ASD with 94% accuracy. They suggested that MEG analysis may in the future offer a non-invasive, quantifiable way of confirming a diagnosis of ASD.

New research suggests that the quality of the ASD intervention that is most important. A recent study that investigated a range of popular and often costly ASD interventions found that it is the quality of the program that makes the most difference to the development of the child (Boyd, et al., 2013). The researchers found that school performance improved when each of the interventions were used. More importantly, the study found that there were no statistical differences between high quality classroom teaching and the proprietary interventions. The study involved 74 teachers and 198 children aged between three and five years of age who were enrolled in public schools.

3. Infancy and Prenatal Influences
The health of a pregnant mother has long been identified as having an effect on the development of a foetus and ultimately the health of the baby. Researchers have shown that when a pregnant mother’s immune system is activated by a viral infection, there is a potential for changes in the development of neural brain cells in the foetus. These changes can impair the brain’s ability to effectively transmit and communicate signals. The researchers used animal models to suggest that the increase in specific molecules that were detected in the offspring, after the mother had been exposed to a viral infection, can lead to developmental changes that may be involved with the increased incidence in conditions such as ASD (Elmer, Estes, Barrow, & McAllister, 2013).

Further evidence of the effect of maternal antibodies affecting a foetal brain was reported by researchers who showed that mothers whose children have ASD were significantly more likely to have specific antibodies in their blood samples than the mothers who did not have children with ASD (Braunschweig, Krakowiak, Duncanson, Boyce, & Hansen, 2013). These findings will allow researchers in the future to develop blood tests and targeted interventions for use during pregnancy with the aim of avoiding the antibodies entering the foetal blood stream.
Mothers who have a low thyroid hormone (thyroxine) levels during pregnancy may have children that are at an increased risk of ASD. Researchers have found that mothers who are severely deficient in thyroxine whilst pregnant, are four times more likely to have a child with ASD than mothers who have normal levels (Roman, et al., 2013). These findings suggest that thyroid function should be tested routinely during early pregnancy. The findings also indicate a need for an increase in community awareness about the effects of low thyroxine during pregnancy.

Inducing and augmenting labour has also been reported to be associated with an increased risk of ASD (Gregory, Anthopolos, Osgood, Grotegut, & Mairanda, 2013). In this United States study, mothers whose labour was not augmented or induced had a significantly lower incidence of children diagnosed with ASD. The researchers suggest that the hormone oxytocin may be contributing to an increased risk of ASD, since up to 70% of women whose labour is induced or augmented are given this hormone. The researchers state that these findings need further validation.

Foetal growth rate that is extremely low, or large, has been reported to be a risk factor for ASD. In a study which looked at more than 40,000 child health records in Sweden, researchers found that babies whose birth weight was above 4.5kg or below 2.5kg were at approximately a 60% greater risk of developing ASD than babies whose foetal development was within the normal growth range (Abel, et al., 2013).

4. Young Adults and Adolescents with ASD
Adolescents with ASD have ambitions and aspirations of leaving their parent’s care and setting up their own home and family (Aspect, 2013a). The We Belong Too study carried out by Autism Spectrum Australia (Aspect) identified that adolescents aged between 12 and 17 years expect to be independent in their adulthood, but they are aware that support is likely be needed for this to occur. The study highlighted the need for support services that provide relevant, easily accessible and affordable transition from school programs. The research also identified that more ASD information, training and support is necessary for employment agencies, employers and tertiary educators so that adolescents can find and retain appropriate employment, in order to have an independent life.

Without consistent and ongoing support, people with ASD often do not reach their potential. Researchers have suggested that young people with ASD typically have a higher unemployment rate and lower income than people with other disabilities or the general population (Shattuck, Narendorf, Cooper, Sterzing, Wagner, & Talyor, 2012; Warren, 2012). Despite the adolescent’s ambitions of a home of their own, the research suggests that young adults with ASD are also more likely to remain living with their parents after high school than people with emotional or learning disabilities (Anderson, Shattuck, Cooper, Roux, & Wagner, 2013).
5. Females with ASD

Autism spectrum disorder is diagnosed more often in males than females, but recently researchers have begun to question whether there is a biological difference between the genders that accounts for this difference. Historically, the majority of neuroanatomical studies have tended to concentrate on researching the male brain.

In a new study, researchers have compared the neuroanatomy of male and female individuals with ASD (Lai, et al., 2013). Using magnetic resonance imaging (MRI), they investigated gender differences in the brain anatomy of people with ASD who were aged between 18-49 years and who have a normal Intelligence Quotient (IQ). The MRI results identified substantial biological differences between male and female brains in people with ASD. The researchers stated that although the diagnosis criteria does not account for gender differences, their research has identified that males with ASD and females with ASD have structurally different brains. The study authors suggest further research is warranted before these findings can be used with established diagnostic criteria.

In another study that looked at the ASD presentation of females, the researchers suggested that teenage girls with anorexia nervosa have a higher number of ASD traits than typically developing age-matched girls (Baron-Cohen, Jaffa, Auyeung, Allison, & Wheelwright, 2013). In this study, the researchers considered that anorexia and ASD might be linked because both disorders involve similar characteristics including rigid attitudes and behaviour. In anorexia, the focus of the restricted behaviour is usually directed towards food and personal weight. The researchers found that girls aged between 12-18 years with anorexia were more likely to score higher on standardised ASD questionnaires than typically developing adolescents. Baron-Cohen, et al. (2013) suggests that anorexia interventions may needed to be reviewed, and that the interventions should include the implementation of coping strategies, rather than concentrating on the patient’s preoccupation with food and weight.

6. Ageing with ASD

Most research into ASD has concentrated on children. A recent report from the United Kingdom has estimated that the prevalence for ASD in adults is comparable to the prevalence in childhood (Brugha, et al., 2011). This finding confirms the view that people with ASD have lifelong persistent impairments that do not resolve after childhood (APA, 2013).

The number of people diagnosed with ASD has increased over the last few years. Some adults may have suspected for many years that they have ASD, but have only recently been diagnosed (Perkins & Berkman, 2012). Data collected in the United Kingdom (U.K.) by the National Autistic Society (NAS) identified that around 70% of people aged over 55 years who are diagnosed with ASD, may have only recently been recognised as being on the spectrum (Boem, 2013).
Perkins and Berkman (2012) reviewed the current literature on the life outcomes of adults with ASD as they age. The researchers concluded that there is very little research available that is appropriate or relevant to aging adults since most of the research is conducted with children and youth. The authors also identified issues such as mental and physical health, long-term accommodation and matters relevant to later-life expectations are lacking from the literature.

In 2013, the NAS released a report that found that older people with ASD are dealing with significant daily life challenges (Boem, 2013). They found that as people with ASD age not only do their health and social welfare issues become more obvious, but that there is also evidence of a lack of understanding of their support needs by the care professionals.

The NAS made many recommendations (Boem, 2013, pp. 30-33) including that:

- Clinicians should ensure older people are not disadvantaged in the diagnostic process by the requirement for a developmental history. This is especially relevant since parental or family accounts of developmental progress may not be available.

- All health and social care professionals, including those working in older age specialisms, and older people’s services, should receive training.

- Funding should be made available for research into a population-based longitudinal study of the health and quality of life of older adults with ASD.

- The needs of older people be considered when making local plans and strategies.

The outcome of this NAS study is that there is an increased awareness of the needs of a newly recognised and increasing population of people who require support. Research is urgently needed for the development of diagnostic, cognitive and behavioural tools. In addition, interventions that are appropriate for people with ASD and who also have other age-related conditions, such as dementia, need research (Mukaetova-Ladinska, Perry, Baron, & Povey, 2012).

7. Technology

Technology can provide an effective means of supplementing communication opportunities for people with challenges with comprehension, including difficulties in following spoken directives (Schlosser, et al., 2013). In this within-subject study of nine children with ASD, the researchers showed that augmented instructions, using cues such as photos, and full-motion video were more effective than spoken cues alone.
These results suggest that technology-assisted communication can be effective for expressing the relationships between people and things in space and time.

Two recent meta-analyses that have reviewed the effectiveness of technology-based interventions have shown that augmented communication devices provide benefits to individuals with communication disabilities.

In one meta-analysis study, which investigated technology-based interventions for children with ASD, the review found that there is evidence of improved outcomes when these technologies are used (Grynszpan, Weiss, Perez-Diaz, & Gal, 2013). In this study, the researchers investigated interventions that used desktop computers, interactive DVD’s, shared active surfaces and virtual reality environments. They found that in pre-test/ post-test studies, the evidence suggested that the children with ASD who received technology-based interventions showed a significant improvement.

In the other meta-analysis study, which assessed effectiveness of augmentative and alternative communication interventions with people who have challenging behaviour, the researchers also showed that these interventions were effective, especially with younger children. The researchers, however, noted that these improvements often occurred in specific situations such as when the interventions occurred in the therapist’s consultation rooms with the therapist conducting the intervention (Walker & Snell, 2013).

This year there has been several studies that have investigated the effectiveness of iPad technology.

A study conducted by Aspect investigated the effectiveness of the iPad for developing core competencies in 58 primary school aged students. The results from the preliminary report found the iPad to be a valuable pedagogical tool for supporting learning in children with ASD (Aspect, 2013b). In particular, the results showed that when using an iPad, 59% of the students achieved an improvement in communication skills. The study used the Proloquo2go application with the iPad.

In a small-scale research project that investigated the effects of video self-modelling on mathematics skill acquisition using the iPad, the results were also encouraging. In this study the researchers (Burton, Anderson, Prater, & Dyches, 2013) found that the students who used an iPad to review and reinforce their own success in solving maths problems showed significant improvements in accuracy in tasks such as, identifying the cost of an item from a price tag and estimating how much change they should receive. The researchers suggested that the main benefit from the iPad was that the students engaged with the mathematics activities because they enjoyed watching themselves performing tasks, especially the ones that they accurately answered.
Video-based instruction was also evaluated in another small-scale study that used the iPod Touch for teaching food preparation skills to adolescent school students with ASD and intellectual disability. This study showed that students increased their ability to perform three different cooking tasks when prompted by an iPod touch. The teachers reported that the iPod technology supported the student’s ability to focus on the task (Johnson, Blood, Freeman, & Simmons, 2013).

Humanoid robots are also being evaluated as a novel method of improving a student’s attention to tasks (Bekele, Critendon, Swanson, Sarkar, & Warren, 2013). Although the results from this small-scale study of preschool children (six with ASD and six typically developing) showed an increase in gaze toward the robot, the conclusion from the authors was that all the children in the study improved their attention to tasks best when prompted by a human.

Conclusions
This research update provides a snapshot of ASD research that is happening around the world. The estimated prevalence of ASD is one in every one hundred people in the community (Baio, 2012). Children are growing up and entering adulthood, and more adults are recognising their own symptoms and seeking out a diagnosis – and support. The outcomes reported in this update provide the basis for new support interventions, technological solutions, and more biological and genetic answers. Together all these findings are helping the community to provide support to people with ASD and their families.
Works Cited


