



AUTISM SPEAKS™
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Advancing research worldwide

Current UK autism research funded by Autism Speaks (2008/2009)

We believe there are four main challenges for autism research: 1) to understand why the number of individuals with autism spectrum disorder (ASD) is increasing; 2) for diagnosis of ASD to be accurate and made as early as possible; 3) to improve our understanding of the susceptibility genes for ASD; 4) to identify the structural and functional differences in the brains of individuals with ASD.

The work of the Autism Genome Project (AGP) is particularly important in the **genetic understanding of autism**. It is an international consortium, currently coordinated in the UK, where genetic data on thousands of families is collected. The AGP is using cutting edge technology to identify areas of the genome where individuals with ASD have greater duplications and deletions in their genetic code, which is seen as a major risk for some cases of ASD. The work of the AGP means we know more about the causes of ASD and what the genetic risks may be for families in the future.

The most effective way of studying **the relationship between genetics and brain development** is by examining microscopic alterations in the cellular and molecular architecture of neural tissue. Dr Payam Rezaie from the Neuropathology Research Laboratory at the Open University uses these approaches and gave a research overview. The original neuropathology studies were plagued by difficulties in gathering together enough brains but with the advent of more organised "brain banking", scientists are identifying brain regions that are different in adults with ASD compared to typical controls. Most significantly, the amygdala (a brain region managing "social" information) and fusiform gyrus (which manages how we look at faces) both are distinctly different in the brains of individuals with ASD. Dr Rezaie commented that researchers must still confirm whether these differences are present in childhood, which can only be archived via international collaboration and an increase in the number of donations.

Another elegant study exploring the relationship between genetics and neuroscience is being conducted by Dr Tom Lavender from the Institute of Psychiatry in London who uses Magnetic Resonance Imaging (MRI) to explore **structural differences in the brains of identical and non-identical twins with ASD**. Identical twins share the same genes, or DNA, so the structural differences between their brains are likely to be caused by divergence in their development or their environment. The study of identical and non-identical twins in this way will help scientists understand more about the gene x environment interactions that cause ASD and how these are reflected in the brain. The team also use an exciting new technique called Diffusion Tensor Imaging (DTI) which can visualise how regions of the brain are connected together. Dr Lavender will explore whether behavioural and cognitive difficulties shown by individuals with ASD can be explained by faulty neural connectivity.

One aspect of cognitive function explored by Dr Simon Wallace from the University of Oxford was **how children with ASD recognise facial emotions and respond to pictures of eyes looking at them**. Dr Wallace reports that the autonomic nervous system of children with ASD is over aroused when they look at eyes gazing at them and suggests that this may be related to underlying difficulties engaging in eye contact and recognising certain facial emotions. His current research uses magnetoencephalography (MEG) which records the tiny magnetic fields when a person's brain is functioning and aims to explore the neural underpinnings of face and eye gaze processing in children and adolescents with ASD. **By identifying these biological differences or "markers" in children with ASD scientists can identify the optimum point in development when educational interventions will be most effective.**

Professor Margaret Esiri is Director of the **Brainbank for Autism and Related Developmental Research**, based at the University of Oxford - the first brainbank for ASD in the UK. It aims to meet the need of neuropathologists such as Dr Rezaie who must study a larger number of brains to produce definitive answers. The Brainbank has been an absolute success and has received 5 brains and over 30 pledges.

Research has shown that if parents already have one child with ASD there is a 10% chance that they will have a second child who will later receive an ASD diagnosis. Dr Mayada Elsabbagh and her team at Brikbeck College are part of the British Autism Study of Infant Siblings (BASIS) network and are following infant siblings who are at increased risk for developing ASD, to look for the “early signs” and behavioural precursors to what will later be ASD. The research uses eye tracking and EEG to identify these early signs and so far 80 families have been enrolled. **Ultimately, this research will lead to children with ASD being identified early in development and with educational interventions in place this will give the child the best possible outcome.**