

## **Autism: where has the science got to?**

### **Highlights from the International Meeting for Autism Research (IMFAR) held in San Diego 12<sup>th</sup>-14<sup>th</sup> May 2011**

This year San Diego played host to the International Meeting for Autism Research (IMFAR), an annual gathering of some of the finest minds in medical research from all over the globe. This was a landmark event – the 10<sup>th</sup> anniversary of the first IMFAR conference – and the scale of it gave a clear indication of how much autism research has grown in the intervening decade. Professor Dan Geschwind, Director of Autism Research at the University of California (UCLA) recalling the first conference in 2001 said “Given how small the field was, we wondered if there would be sufficient interest and scientific advancement to warrant a meeting, even every other year. This was an unnecessary worry”. The first conference attracted just 250 scientists. This year there were over 2,000 attendees, and more than a thousand research papers were presented.

The researchers in San Diego represented a broad range of disciplines, including neurology, biochemistry, psychiatry, genetics and statistics. There was a strong sense that in a number of significant areas, research is reaching a new level. For example, autism genetics as usual featured in the scientific presentations but not as prominently as in recent years, instead work was more focused on the links between these genetic variations and the structure and functioning of the brain (so called imaging genetics) and the behavioural phenotype.

A ‘hot topic’ was post-mortem brain research and the neuropathology of autism – and its role in examining the biology of the brain at the cellular and molecular levels. Professor Margaret Bauman (Mass General, Boston) was honoured with a lifetime achievement award for her neuropathology work and her role in the Autism Treatment Network. Dr Eric London in the acceptance of his lifetime Advocate Award referred to the paucity of brains for autism research and to the new brain bank he is setting up in New York County.

Professor Eric Courchesne (University of California San Diego UCSD) in his keynote lecture on the neurodevelopmental biology of autism, reviewed the vast amount of his team’s recent (2009-2011) neuropathology work using post-mortem brain material supplied by the Autism Tissue Programme (of which the UK’s Brain Bank for Autism is a collaborative partner). Professor Courchesne research presentation included captivating 3D images of microscopic brains sections and brought a normally dry topic to life. He explained his findings pointed to alterations in the migratory track of neurones during the development of the brain. He also reviewed a programme of ‘baby well checks’ developed by Dr Karen Pierce through which 18,000 babies were screened, and 420 classified with an ASD. [This type of early screening programme was echoed in a presentation by Professor Amy Wetherby (Tallahassee) regarding the Early Screening for Autism and Communications Disorders (ESAC) and Systemic Observation of Red Flags (SORF)].

Another area of research, pluripotent stem cells, has seen tremendous advances in ways that not so long ago might have been viewed as science fiction rather than scientific creativity. Professor Ricardo Dolmetsch (Stanford) gave a keynote lecture, showing that stem cells (which have the capability of differentiating into any cell in the body) can be obtained from living individuals through small skin biopsies. The stem cells are then grown in culture in the laboratory and encouraged to become neurones and show characteristics of ‘real’ neurones such as growing axons and making synaptic connections. It is thought such differentiated neurones represent the developmental stage of the first or early second trimester of pregnancy (i.e. at a point relating to early brain development and at which post-mortem research has indicated that some neuronal abnormalities arise). Professor Dolmetsch is now at the early stages of drug testing on these neuronal cultures. This work has concentrated on Timothy Syndrome, a monogenic disorder which is an autism risk factor and has yet to be applied to autism. Such research efforts are all

the more remarkable, since Professor Dolmetsch switched the focus of his research career to stem cell research in response to his young son being diagnosed with autism.

There were a number of presentations on research on adults. The research studies were of high quality but the implications of the findings were unsettling and uncomfortable listening:

- Professor Terry Brugha (University of Leicester) reported only 5% adults with autism have a diagnosis.
- Professor Pat Howlin (King's College, London) in her review of 23 published studies on adults with autism and her 40 year follow-up of the 'Maudesley cohort', reported that about 50% of adults diagnosed with autism as a child have poor or very poor outcomes in adulthood, around 30% have fair outcomes and only 20% have good or very good outcomes; 19% live independently, 50% live with parents and 33% in residential care; adults with autism become more isolated with age, losing contact with friends from school and failing to make new friendships and relationships; ASD symptomatology reduces with age as measured using the ADI-R (but still remains within the spectrum), notably there is a reduction in severe behavioural problems with age; for some adults with autism Professor Howlin said there is a drop in IQ over time with a bigger drop being seen in women than men affected.
- Professor Marsha Seltzer (Wisconsin) reported adults who begin drug treatment, are often on medication for life. She reported that the extent that an individual has developed useful language at age of 5-6 years is important indicator for adult outcomes. She also said that the lack of structured life in adults allows the invasion of rituals, and that there is a higher proportion of mental health problems and adults with high ability not served well.

There were a number of reports regarding Autistica funded research, such as from scientists connected with the British Autism Study of Infant Siblings (BASIS, Birkbeck College London) who reported their early findings that in babies aged 10 months there are some patterns of brain activation that may relate to the presence of autistic symptoms at 36 months of age. Professor Jonathan Green (University of Manchester) gave an overview of the iBASIS study – an early intervention study which has started very recently.

*More information about these projects can be found on the Autistica website (see [http://www.autistica.org.uk/research/research\\_we\\_fund.php](http://www.autistica.org.uk/research/research_we_fund.php))*

Professor Sally Rogers (UC Davis) reviewed the challenges in conducting intervention research in very young children – namely stability of the symptoms and diagnoses (stating that less than one third of young children retain an ASD classification as they age from 18-36 months). She highlighted the potential impact this may have on the mis-allocation of resources and recommended interventions be applied to ameliorate the worst of the autistic symptomatology and that investment be made in developing 'efficacious parents'. Professor Alice Carter (Hunter), in the context of stability of symptoms, urged careful consideration of 'message framing' (i.e. how to considerately inform parents of a new diagnosis for their child - when the diagnosis and symptoms may change).

Dr Tom Insel (Director at the US National Institute of Mental Health NIMH) gave his annual update on the activities of the Interagency Autism Co-ordinating Committee which reports to Congress in response to the Combating Autism Act. The IACC has identified some new research objectives including the 'microbiome' (i.e. how the bacteria part of our daily life influence human health and disease), epigenetics (i.e. heritable changes caused by mechanisms other than changes in the underlying DNA sequence) and non-verbal individuals (a neglected group in terms of research). Dr Insel revealed that National Institute of Health (NIH) had invested two thirds of the \$314m spent on autism research in the US in 2009 (the latest year for which figures are available) and \$122m was committed under the American Recovery and Reinvestment Act in response to the economic downturn. In 2010, \$217m was invested, representing a 4-fold increase over 2003 in a '0' inflationary climate. For further details see [www.iacc.hhs.gov](http://www.iacc.hhs.gov)

Overall the atmosphere was positive at IMFAR and the scientific progress strong.

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