

Autism: where has the science got to?

With autism attracting a great deal of attention and funding worldwide, Paul Burden takes a look at the current state of research into this debilitating condition

I recently brought up the issue of autism in conversation with a friend who works in the area of special education. The response of my friend, an educational psychologist, was immediate and emphatic. The SEN business, he told me, was being inundated by a tidal wave of demands from parents seeking diagnoses of autism for children who were having difficulties at school. There was, he said, no evidence that there had been a huge increase in the occurrence of autism in the population. The driving force for more diagnoses was the increasing number of parents wanting a concrete explanation for their child's problems and with it, perhaps, the promise of a statement of special needs and the hope of extra specialist help.

But what reassurance or certainty does a diagnosis of autistic spectrum disorder (ASD) bring? The truth is that the world of autism is a complex and sometimes frightening maze of largely unanswered questions; autism is a disabling neurological disorder which is life-long and for which there is no substantial remedy. However, there is now more reason to be hopeful; over the past ten years, a massive international research effort has built up, bringing with it the prospect, eventually, of new therapies and interventions to relieve the symptoms which cause such distress to families affected by autism.

The scale of that effort was manifested in May this year at a major medical conference in California. San Diego, famous as the home of the US Navy's Pacific fleet, 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 something of a landmark event – the 10th 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) recalled the first conference in 2001. "Given how small the field was," he said, "we wondered if there would be sufficient

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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, and more than a thousand research papers were presented at the conference.

The researchers at San Diego represented a broad range of



Babylab researcher with baby Piers. Photo: Babylab, Birkbeck.

disciplines, including neurology, biochemistry, psychiatry, genetics and statistics, all seeking the answers to two basic questions: what is autism and what can be done about it? The hope was that somewhere in the mass of data presented to the delegates would be found the crucial leads to unlock the mystery which surrounds the condition.



Imitation study with typically developing toddler. Photo: Babylab, Birkbeck.

Among the participants was Dr Jenny Longmore, Director of Research at Autistica, which has channelled over £2 million into a wide variety of research projects being undertaken at British universities. The atmosphere, she reported, was positive. There was a strong sense that in a number of significant areas research is reaching a new level. For example, it has been widely accepted for some time that autism is an inherited condition. There has been considerable progress in identifying specific genes which are associated with autism. Attention is now being targeted on the links between these genetic variations and the functioning of the brain which gives rise to the patterns of social impairment which are characteristic of autism.

Scientists now have the ability, says Dr Longmore, to use MRI scans to focus on small areas of the brain and so detect minute variations which are found specifically in people with autism. The scientists now have a much clearer picture about the stage of development at which these variations occur. Much of the new information which is now emerging is a direct result of improvements in the technology used in brain scans.

Another recent development has been in the area of stem cell research; scientists are now able to develop stem cells from tiny samples of skin so

that they can study in the laboratory the neurones, the key elements in the neurological system which are believed to play an important role in the development of autism.

Meanwhile, the development of new IT systems means that the quantities of

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data emerging from the research of the human genome can be analyzed very much more quickly.

One important factor in the acceleration of autism research has been the provision of large scale funding, notably by the US Government which recently committed some \$314 million to this area. While the US may be the biggest player in the field, it was acknowledged in San Diego that the UK is also playing a significant role.

One of the most important British-funded projects is an ongoing study of the behaviour of very young children at risk of developing autism because the

condition has been diagnosed in other family members. The scientists believe that the best hope of alleviating the impact of autism is to intervene at the earliest possible age. However, the standard method of testing for autism is not effective below the age of three years old. Scientists at Birkbeck College in London have been examining toddlers of as young as six months to

establish whether there are discernible signs of abnormal behaviour at this very young age.

The Holy Grail for researchers is to identify a biological marker – an observable physical distinction in the autistic person which definitively confirms their diagnosis. When they reach that stage, the scientists can start to claim honestly that they have a real understanding of what autism is.

Meanwhile, parents grappling with the problems of raising an autistic child must be resigned to waiting, perhaps for a while yet, before the medical profession can offer them substantial hope. Notwithstanding the progress of the last few years, the researchers hunting for the truth about autism are a little like the mountaineers on an unclimbed peak. They hope that when they reach the next ridge, the summit will be in sight, but it may still be some way off. **SEN**

Further information

Paul Burden is Communications Director at Autistica, a UK charity which raises funds for medical research into autism:
www.autistica.org.uk