

Report from Angelman Syndrome Foundation Scientific Symposium and Biennial Conference, Orlando, Florida, July 28th-August 1st 2009

The Angelman Syndrome Foundation of the USA has established the tradition of holding a scientific meeting for professionals working in the field of AS in the two days before the ASF family conference. This has the advantage of enabling scientists and clinicians who work in the field of Angelman Syndrome to get together and share ideas and then to have the chance of staying on for the family meeting where a variety of professionals including paediatricians, geneticists, psychologists, behaviouralists, teachers and others can give presentations and learn useful tips on managing different aspects of Angelman syndrome. The most useful information is often gained from the presentations by parents and discussion with the families themselves. For those interested in AS, this was a unique opportunity to meet with parents and AS children from over 70 families.

This year's meeting was a momentous one for the ASF as Dr Charles Williams, Chair of the ASF Scientific Advisory Committee announced their intention to establish ATRI, the Angelman Treatment and Research Institute, with the aim of prioritising their increasing amount of research funding for projects which aimed to improve management and find treatments for AS.

The presentations at the scientific meeting, "Angelman Syndrome at the Synapse" were of high quality and reflected the broad range of Angelman research, though with a much greater focus this time on basic science research rather than clinical studies. There were some inspiring presentations particularly by Ben Philpot of North Carolina who has demonstrated that the mouse AS gene, Ube3A is involved in experience dependant maturation of the mouse cortex and from Michael Ehlers from Duke University who has shown that Ube3a localises to the Golgi apparatus may disrupt function of this organelle. Edwin Weeber from the University of South Florida has previously published in Nature Neuroscience on work demonstrating a relationship between CAMKII, Ube3A and symptoms of AS in a mouse model. He presented further work on Neuregulin-1 signalling in the AS mouse model suggesting that it, too had a role in the symptomatology. For me, one of the best and most exciting of the scientific presentations was given by David Segal from the University of California. He talked about the use of artificial transcription factors and their possible role in gene therapy for Angelman Syndrome. The presentation was clear and inspiring and the technology generalisable. Europe was represented by Peter Hammond, Jill Clayton-Smith and Chris Oliver from the UK. Peter Hammond presented work on 3D facial imaging in AS, showing elegantly that 3D imaging had the capacity to distinguish not only between AS syndrome and normal faces, but also between the different classes of Angelman Syndrome. Chris Oliver's team presented further work on social behaviour in Angelman Syndrome. One further presentation worthy of mention was that by Jane Summers, a psychologist from McMaster Childrens' Hospital who has been developing a battery of tests to assess memory, motor performance and motor imitation in children with AS. When you have a condition where verbal skills are impaired so much, the validates tests available for testing normal children just don't work and Dr Summers's video clips demonstrated well the use of the test battery she is developing. She was awarded a well-deserved prize by the ASF for her work on Angelman Syndrome over the year. Dr Joe Wagstaffe who first published the discovery of the UBE3A gene was awarded a research prize posthumously and this was accepted by his sister.

With professionals from so many different fields, all presenters worked hard to make sure their presentations were understandable to a mixed audience. Parents were allowed to attend as observers and several took this opportunity. One fact that is now clear is that Angelman syndrome affects the whole brain, not just the cerebellum and hippocampus as was previously suggested. There is evidence that dendritic branching isn't as good in AS. However, it is not a progressive degenerative disorder and the mouse work offers real prospects for a treatment which, if instigated early enough, might ameliorate the symptoms of AS. No wonder the ASF, a very well informed group, have chosen to direct their efforts into the ATRI.

Moving on to the family meeting, this consisted of plenary sessions with lectures given by invited professionals such as Dr Weeber. Stephen Calculator, Professor of Communication Sciences and Ganesh Gupta, an orthopaedic surgeon with expertise in management of Angelman syndrome, interspersed with workshops on various topics including sleep, behaviour, seizures, provision of services etc. There was a wealth of relevant information for parents of children of all ages. There were moving presentations, too, from parents and a workshop for siblings. Finally, anyone who took the time to read some of the journals and scrapbooks in the conference foyer could learn a great deal about the natural history and evolution of the syndrome from a parents' point of view, and about its impact on families. All presentations were audiotaped and available to purchase from the ASF.

So for all those who think you already know all about Angelman Syndrome, I would recommend that you go to an ASF meeting and you will be amazed how much more you can learn. Although medical care in the US doesn't translate directly to the options available in Europe, this meeting really shows what can be done for families when parents and professionals get together and are organised well and the ASF provides an excellent model for support groups everywhere.

Jill Clayton-Smith, August 2009

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