Dear Families,

Another year has gone by and we are writing again to wish you a Merry Christmas and Happy 2010 New Year and our thanks and appreciation for your support of our research efforts. Our clinical team has remained busy throughout the year seeing children and families and now over 30 more families have joined the genetics research effort. These and many more Irish families who were previously recruited have been included in the genetic studies.

Important Research Findings

Last year when we wrote we told you about the new findings in autism genetics research. You might remember we wrote about ‘copy number variation’. This is where a person can get more (duplication) or less (deletion) genetic material in a particular region than usual. Sometimes duplications or deletions can affect one or more genes. In general, we have two copies of each gene (one from our mother one from our father). One of the most common seen ‘copy number variant’ syndrome in the population is Down’s syndrome – where a child gets an extra copy of a chromosome 21. We know from our AGP study that around 5% of the 2000 affected children that took part in the study have rare deletions or duplications of parts of their chromosomes. Some of the genes that these variations are affecting are involved in guiding connections in the brain between cells. Last year we told you about a region on chromosome 1 (1q21). Now ours and other studies are highlighting regions on chromosome 5, 16, 15 and 22 among others. These findings are helping us to identify some cases of autism that have a known genetic cause. As we identify groups of individuals with similar chromosome variations we can examine the type and course of the individual’s autism and use this information to help predict health and treatment outcomes for others born with similar variation. Tests like this are some way in the future and we still need to do more work to understand more about the genetics of the condition.

You may also be aware of our effort and that of others to identify genes important in autism in the wider population. In the last year a number of studies have been reported in the mainstream press highlighting genes that contain variation that are over- and unrepresented in individuals with autism. These variation affect genes (Cadherins and Semaphorins) involved in the structure and function of the neurone, and how neuronal messages are past. Our researchers are examining how these variations impact on the biology of the neurone.

Trinity researchers are also examining the data from these large reports alongside that of our own as part of a meta-analysis of many thousands of individuals with autism. These meta-analyses will provide further strong evidence to identify genes and variation that can be used to better understand autism.

AGP Phase 3:

We are making plans now and applying for research funding to develop the next phase of the research program across the 50 international sites of genetics researchers. We are at an exciting time in our project. Already we have included >6000 individuals with autism and family members worldwide. We plan to expand on this number more in the next phase.

New Projects:

We also have some other projects that are going on at the current time:

Sequencing project:

We are planning to look at some genes very deeply using ‘sequencing’ which is a way of looking directly at the genetic code. This will tell us if there are errors in the genetic code that cause genes to malfunction. The human genome project cost approximately $10 billion to complete and over 13 years to produce a completed draft sequence of the human genome. Technology in our laboratory can repeat this experiment in weeks for around €10,000. Using this technology we will look at “genes of interest” in many hundreds of people to establish whether there are rare or unique DNA differences in those individuals with autism compared to their unaffected family members.

Investigating rates of autism in 1st generation Irish children:

We have noticed through our clinical experience that a large number of the children we see are born to parents who have just moved to Ireland. We are
interested to investigate this further to see if there are higher rates of autism in these children compared with children born to native Irish families. If so, we are interested to know if there is any difference in the type of autism, more or less severe or if there are other factors that might have influenced the development of autism. This research project is starting in July 2009 through the Adelaide and Meath Hospital (incorporating the National Children’s Hospital) in Tallaght. The research funds were donated by the National Children’s Foundation, Tallaght.

**Neuroimaging studies:**

Last year we told you about the TRACT Study *(Trinity Research in Autism – Connectivity of Tracts):*

The TRACT study uses Magnetic Resonance Imaging (Brain imaging) techniques to investigate brain activity and connections between brain regions in children with Autism/Asperger syndrome. We ask study participants to perform computer-based tasks (like simple computer games) while they are lying in an MRI machine that takes rapid pictures of the brain. To date 31 young people, 17 with Asperger’s syndrome and 16 typically developing individuals, have taken part. We are planning to recruit another 25 or so people in the study. We are really grateful to all those who took part and welcome new recruits.

To take part in the study we have to use quite strict criteria to ensure that we can compare the brain scans and so all participants must be male, right-handed, aged between 14 and 21 and have no metal in their body (this includes braces on teeth).

If you would like more information about this study, please contact Dr Jane McGrath (contact details below). She may not get back to you straight away as she has just had a new baby, but will be back in May ready to get going again.

**Social Reward and Autism:**

We are also developing a new neuroimaging study to investigate if people with autism find social interaction less rewarding than typically developing people. We are still setting up the project but will be looking for new recruits early in 2010.

**Reminder regarding questionnaires**

Thanks to everyone who has completed the questionnaires so far. We understand that there are a lot to fill in, and that it is hard to fit in when things are busy. Each of the questionnaires is important for examining different aspects of autism behaviour, and for linking in this to the genetic analysis. If you have forgotten about the questionnaires or mislaid them don’t worry, we will probably be contacting you about them in the New Year.

As these are ongoing research projects we are continuously looking for new recruits, so if you know of any other families that might be interested in taking part in this study, please feel free to pass on our details so that they can contact us for more information about this project.

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