The research continues…

It is perhaps helpful to think about research as like the slow completion of a jigsaw, at times pieces are inserted that are found to be incorrect and other times the pieces fit and the picture becomes a little more apparent. Over time findings from very different types of studies illuminate specific aspects of our understanding, however, it is not until a critical mass comes together that the full picture can be imagined and it is then that understanding may leap forward and the potential for new treatments are realised.

This is very true for research into Down’s syndrome and dementia as our findings and those from other centres are peer reviewed and eventually published for all to see. Pharmaceutical companies are developing treatments that aim to target particular pathways in the brain believed to be important in the causation of Alzheimer’s disease. In the case of DS a protein called ‘amyloid’ appears particularly important as the gene coding for its precursor is on chromosome 21 and people with DS have three, rather than the normal two copies. It is this protein that with age forms the brain plaques characteristic of Alzheimer’s disease. Through the research you have helped with we have been able to show where in the brain and at what age this amyloid becomes apparent in people with DS. Medications are now available that alter amyloid levels and it is these that are of interest, but they would almost certainly have to be started many years before Alzheimer’s disease became apparent to have a chance of success.

So the ‘jigsaw’ is still incomplete but the picture is becoming clearer thanks to your involvement. As you will read about in this newsletter we have a new study funded by Alzheimer’s Research UK and we are also part of an American funded consortium. Both studies are aiming to explore the role of amyloid using brain imaging and also the role of a protein called tau in the causation and course of Alzheimer’s disease. Please help if you feel you can. Thank you.

If you have a general enquiry about our research, or if you would like to be added to our email list, please contact us: ah937@medschl.cam.ac.uk

Thank you again for your interest and involvement,

Tony Holland, Emeritus Professor of Psychiatry
Cambridge Intellectual and Developmental Disabilities Research Group
We have said goodbye to three of our research colleagues who many of you will have met during the course of their work. Tiina Annus finished her PhD and has gone to work for a pharmaceutical company. Liam Wilson also completed his PhD and is now working as a scientific editor. They presented their work at our PhD celebration dinner and we wish them well in their new careers. They would like to thank all the participants and their families for taking part as well as the rest of the department here for their support.

Paula Castro, who many of you will also have met, has returned to Brazil to continue her medical training.

We have a new person working as the research team administrator and keeping track of our newsletter recipients. Agnes Hoctor joined us in October and replaces Suzie Tall who has returned to her teaching career.

Dr Ruma Raha-Chowdhury, one of our local collaborators, has two new PhD students, James and Deniz, working with her at the John Van Geest Centre for Brain Repair.

We have been carrying out a survey to gain carers’ views about potential medication trials that alter amyloid levels in people with Down’s syndrome. We have received 65 responses so far which have provided us with a lot to consider, thank you to those who returned the surveys that came out with the last newsletter.

Initial results show that carers on the whole have a positive attitude towards trials (see the graph to the right) and the study is providing useful information for researchers on how a trial could be designed to enable people with DS to take part and to benefit from new treatments. The online survey has now closed but if you would like to complete the paper version please contact us and we will post you a copy: Liz Jones at ej268@medschl.cam.ac.uk or Agnes Hoctor on 01223 746 007.
Pulling up support for our new study

Russell Ramsey has taken part in all our studies and is a keen supporter of our research. He is the very first person to sign up to the new study. This means he will be continuing to have scans and tests from time to time over the next 4 years. Here he is enjoying doing some of the puzzles and tests at his home with Dr Concepcion (Conchy) Padilla on his day off from work at Marks and Spencer.

As an actor Russell is not new to media attention having played parts with Ricky Gervais and Daniel Radcliffe in 'Extras', Stephen Mangan in 'Never Better' and in an advertisement produced by the Disability Rights Commission highlighting abuse endured by people with a learning disability. Thank you Russell for your continued participation, without you and others with DS this research would not be possible.

New brain imaging study - we need your help!

This major new international project aims to study the links between Alzheimer’s disease and Down’s syndrome using new brain imaging techniques. We are investigating the roles that specific proteins in the brain, called amyloid and tau, have in causing Alzheimer’s disease. We will be conducting the work at the Wolfson Brain Imaging Centre at Addenbrooke’s which is a major research facility housing state of the art imaging equipment.

We will assess the levels of these proteins in the brains of people with Down’s syndrome and their non-Down’s syndrome siblings (where possible) using Positron Emission Tomography (PET) and Magnetic Resonance Imaging (MRI). These scans will be repeated twice over a period of 4 years, alongside cognitive tests and blood tests to determine the link between protein levels in the brain and blood and changes in memory and other cognitive abilities. To take part in this study you will need to come to Cambridge once a year for 4 years. We will pay for your travel and accommodation during your visits.

If you have taken part in our scanning studies in the last few years we will be contacting you to follow up from your previous scans and if you are interested, we will send you all the information and visit you at home.

If you would like to know more, please contact Dr Conchy Padilla by phone or email using the details below. Also, if you know anyone else with Down’s syndrome who might be interested in taking part in this study, please feel free to pass on this newsletter to them.

More information and a short film about the brain scans are available at www.dementiainds.com.

Dr Conchy Padilla can be contacted by email at cfp31@medschl.cam.ac.uk or telephone: 01223 746 127 (voice mail after office hours).

Thank you Russell for your continued participation, without you and others with DS this research would not be possible.

Conchy Padilla and Russell
Thank you for your support and Season’s Greetings!

None of this important and innovative research would be possible without the dedication and enthusiasm of the people with Down’s syndrome who take part in these studies. It is very clear to us that there is only one way that we as researchers and you the public can find the answers we need, that is through working together.

Earlier this year we made a film, entitled Together, showing why we are doing this work, and how our participants, their families and supporters are helping us. The filming took place at our Open Day in March, which celebrated World Down’s Syndrome Day. The film can be viewed online at https://youtu.be/pB7iqWUXQIM

New research published

We have just had a new piece of work published in the Journal of Neurology. The paper is titled “Alzheimer’s disease in people with Down’s syndrome: the prospects for and the challenges of developing preventative treatments” and was written by Paula Castro, Shahid Zaman and Tony Holland. The paper is available online at http://link.springer.com/article/10.1007/s00415-016-8308-8

Dr Ruma Raha-Chowdhury, who is part of our group, has been working on the link between a protein called TREM2 and the development of Alzheimer’s disease. Previously, researchers had thought that TREM2 only caused damage to brain cells in Alzheimer’s disease by affecting inflammation, which is how brain cells deal with injury of any kind. However, Dr Raha-Chowdhury’s results suggest that TREM2 comes via the blood into the brain and is involved in helping brain cells to survive. As we get older there may be smaller amounts of TREM2 available to help protect the brain and this makes it more vulnerable to Alzheimer’s disease. For more information on this please see the publication in the Journal of Alzheimer’s Disease, “Neuroprotective Effect of TREM-2 in Aging and Alzheimer’s Disease Model” or online at https://www.ncbi.nlm.nih.gov/pubmed/27662313

Conference news

Sally Jennings and Maddie Walpert recently attended the Society for Neuroscience Conference in San Diego, California. This conference is the largest neuroscience gathering in the world and this year over 30,000 people attended. Both Sally and Maddie presented posters on the work they are doing in EEG and imaging of the eyes and talked to scientists and doctors from around the world about this important research.

Sally Jennings presents her work at the Society for Neuroscience conference