As projects come to an end, these in turn lead to new ideas and, with this, knowledge slowly builds up over time. Research is very much an interactive process with occasional false starts and dead ends but also times of new insights and exciting advances.

In this newsletter we briefly highlight some of the findings that are now emerging particularly from the neuro-imaging project. This and other projects have been possible because so many people with Down’s Syndrome and their families, and others who support them, have been willing to help out. For some of you this has involved several visits because the scanner broke or something else happened. You will be pleased to hear that the University has received a very large grant to replace the different brain scanners with new state-of-the-art scanners – this is now in progress.

We are also very pleased that we have received a grant from Alzheimer’s Research UK to continue this work and we are part of an American led consortium (funded by the USA National Institute for Health, NIH) also using neuro-imaging to investigate why people with Down’s Syndrome have such a risk for Alzheimer’s Disease in later life.

As you know, our long term aim is to establish whether dementia can be prevented in people with Down’s Syndrome perhaps by altering levels of the protein amyloid that people with Down’s Syndrome have in excess. Ultimately, as potential treatments are developed, this will require the undertaking of a formal clinical trial. You will see in the Newsletter an invitation to complete a questionnaire about this. This can be on paper or on-line. Details are on the back page. Could I ask for as many of you as possible to complete this — your views are valued.

Enclosed with this newsletter is your invitation to the Down’s Syndrome Research Group’s Open Day to be held on Sunday 20th March 2015 at our base in Douglas House, Cambridge. Please do join us if you are able to do so.

If you have a general enquiry about research, please email Tony Holland ajh1008@medschl.cam.ac.uk. As our research papers become published with open access, we will be sending out PDF copies of them to all of you on our email list so you can see how knowledge is advancing. If you would like to be added to our email list, please contact Suzie Tall st520@medschl.cam.ac.uk

Thank you again,

Tony Holland
Professor of Psychiatry
Cambridge Intellectual and Developmental Disabilities Research Group
Energy in Down’s syndrome study

Mitochondria are the power houses of all the cells in our body. They are especially important to cells that are power hungry, because if they fail to supply fuel called ‘ATP’ the cells start to malfunction. This can lead to problems depending on which cells are affected. If brain cells (neurons) are affected this could explain the memory problems seen in dementia.

Amyloid protein deposits are likely to cause dementia and it could be that in dementia these deposits are more likely to occur if there is mitochondrial malfunctioning. Kate (pictured here) has found evidence of this in volunteers with Down’s syndrome. She found that the worse the ability of mitochondria to supply ATP the greater were the amount of amyloid deposits in the brain (see figure below). This could mean that if we improved mitochondrial functioning, say with exercise or medication, then we could prevent or slow down the onset of dementia by reducing the deposition of amyloid.

Brain activity in Down’s syndrome study

Sally is using a technique called electroencephalography (EEG) to investigate how the brain activity of adults with Down’s syndrome changes with age. Thanks to your help, Sally has recruited 42 people with Down’s Syndrome and 34 control participants into her study and is just finishing off visits before getting down to the analysis. Sally is also interested in whether people’s brain activity can predict their future performance on memory games and puzzles. Sally will be concentrating on this part of the study in the coming year.

Contact Sally: (01223) 746147 srj32@medschl.cam.ac.uk
Dementia in Down’s syndrome study

Thanks to many of you who came to our open day in Cambridge and in London and to our ambassador Glenn Madeley, who was brilliant in sharing his experiences, when taking part in the brain scanning study. He is the lovely gentleman in the middle of the picture with Liam and Tiina (on the right). The 'Dementia in DS study' is coming to an end and Liam and Tiina are looking at the brain pictures to understand how dementia develops in people with DS and what can be done in the future to stop people with DS getting memory problems. They have recently published a research article in a scientific journal called ‘Alzheimer’s & Dementia’: http://www.alzheimersanddementia.com/article/S1552-5260(15)00266-9/fulltext. This article found that the fourth decade is a critical period in amyloid accumulation and development of cognitive decline (Figure 1). It also found that amyloid is first seen in the striatum in people with DS, then in the frontal lobe, then temporal lobe and lastly in the occipital lobe (Figure 2). This information is helpful for future studies that will try to remove amyloid from the brain and stop the development of Alzheimer’s disease and dementia in DS.

Eye changes in Down’s syndrome study

Maddie is looking at changes in the eyes using an optical coherence tomography (OCT) scanner. Recruitment for this study has gone really well over the last 6 months, we now have retinal scans for 44 people with Down’s syndrome and 51 control participants.

The images show some of the results we can get using the OCT scanner. The multi-coloured image on the right shows the volume of the retina and the graph below shows the thickness of the retinal nerve fibre layer surrounding the optic nerve. Next year we are hoping to follow up some of these scans to see if people’s eyes have changed over time.

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Contact Maddie: (01223) 746172
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New! Carer views questionnaire

We are currently seeking the views of family carers and support workers about the acceptability of possible drug trials in the future. We would be really grateful for your help with this by spending a few minutes of your time completing a questionnaire. This can be done on-line https://www.surveymonkey.co.uk/r/8DY5J6S or you can request a paper copy from Suzie Tall: st520@medschl.cam.ac.uk Tel: 01223 746007.

Thank you for your support

Many thanks to all of you who have already taken part in one or more of our studies, we really appreciate your time and effort helping us with this research. It is especially valuable if you are able to help us with different studies — this work is only possible with your help.

We would also like to thank everyone who has helped us complete this research including many people with DS and their families and many people who provided support. We would particularly like to acknowledge the Down’s Syndrome Association; Hft; SEPT NHS Trust; Homerton University NHS Trust; Mersey Care; Jenny Gurney at the Fynvola Foundation; Dorothy Pritchard from Warrington Mencap; The Quemby family from Down’s Syndrome OK; VOX, the staff at the Wellcome Trust Clinical Research Facility; the staff at the Wolfson Brain Imaging Centre; and those at the Herchel Smith Building. We are grateful to the MRC, Baily Thomas Charitable Trust, the NIHR Cambridge Biomedical Research Centre, the Health Foundation, the DSA, Stevenage Biomedical Catalyst, Alzheimer’s Research UK, and Addenbrooke’s Charitable Trust for financial support.

We are also part of an international study funded by NIH in the USA and we are grateful for this support which, together with additional support from the DSA and AR-UK, is enabling this research to continue.