Welcome to our first newsletter!

We are a group of researchers based at the University of Cambridge with a special interest in developmental and intellectual disabilities with a particular focus on Prader-Willi Syndrome.

In this newsletter we want to update you on current and future research taking place here at the University. We may be contacting some of you in the near future about taking part in some of these projects. Thank you in advance for your help and support.

Research provides the understanding that can lead to advances and to new treatments and to improvements in policy and practice. For research to be successful it is about having the right partnerships in place between people with PWS, their families, others who support them, and the clinicians and researchers of various disciplines developing the ideas and undertaking the studies.

The UK Prader-Willi Syndrome Association (PWSA) has been central over many years in helping with research. Without the PWSA’s support and without people with PWS being willing to take part and answering yet more questions, having various blood tests and lying in brain scans, no advances could be made. Funding for research is also critical and we are grateful to the Wellcome Trust and Dunhill Medical Trust for funding our earlier research and for recent donations from Sam’s PW Research Foundation (set up by the Gambi Family, whose son Sam has PWS), Addenbrooke’s Charitable Trust, and the Foundation for Prader-Willi Research (FPWR).

Please let us know if have any comments about this newsletter and let us know how we can best keep you informed in the future. Do share this newsletter with any friends or family who might be interested. If they would like to receive their own copy by email or post please ask them to contact us using the details on the back page.
An overview of current research

Professor Tony Holland provides some background to the nature of research into PWS:

Here at the Cambridge Intellectual and Developmental Disabilities Research Group we have been working on PWS research for twenty years. Our early research in PWS led by Joyce Whittington and myself initially focussed on developing a better understanding of the characteristics of the cognitive strengths and weaknesses observed in people with PWS, the over-eating and other behaviours, and the mental health problems associated with having PWS, and how they might differ according to the genetic sub-type of PWS. These findings were published in various research journals and as a book.

We used brain scanning to show that the response of the brain after food intake was atypical in people with PWS and this supported the idea that people with PWS did not sense ‘fullness’ or ‘loss of hunger’ after eating, in the same way as people without PWS. We also made the striking observation that the risk of developing serious mental illness differed between the genetic sub-types of PWS with those individuals with UPD being at greater risk. The various observations made in the first phase of research have led directly to the second. This new phase is about answering questions about the cause and underlying mechanisms in the brain that result in these problems. The hope is that by understanding these mechanisms some of these problems might be prevented and better treated.

In partnership with the PWSA (UK) we recently recognised the need for a more systematic approach to identifying and recruiting people with PWS for research and with this in mind the PWSAs sent out a letter to families and others who support people with PWS to ask if they would help in contacting people with PWS as and when a research project is developed. Having information on the number of people who could be contacted, and the age, gender and genetic sub-type of those with PWS is immensely helpful as it can be used to support a grant application for research funding. Thank you to all of you who have responded. We ask that you also mention this to other families who have a member with PWS who may not have heard of this initiative and if they are interested they can contact us by email or call us, details are on the back page.

Research into PWS is challenging, as it is a rare and a complex genetically determined syndrome with different systems of the body being affected and with striking differences between infancy and later life. Not only ourselves, but research groups in the USA, France, Australia and elsewhere in the world are working on different aspects of the syndrome. Recently the French group made some very important observations on the use of the hormone oxytocin in young infants. Its use in adults remains contentious with differences in findings between the French and Australian groups.

As research findings from groups across the world are published in peer-reviewed journals, so knowledge increases and the pieces of the jigsaw come together and with time a more comprehensive picture emerges and advances in treatment are made. Please read on for more information about the projects currently underway here in Cambridge.

Research database project

To support research and help us obtain research funding we have worked with the UK PWSA to establish a confidential database of the names of families or others who are supporting people with PWS and who have agreed to be contacted about any new research project. They would then speak to the person with PWS they support to see if he or she would be willing to meet with the researchers and to decide whether or not to take part. As PWS is a rare disorder it is very important to have some idea of the ages and genetic profiles of people with PWS who may be willing to help. If you know of others who may be interested in research please pass on our details which are on the back page of this newsletter.

Maintaining mental health in PWS project

We are working on a new study to help us better understand why people with PWS (particularly those with the UPD form of PWS) have an increased risk of developing serious mental health problems. We are submitting a grant application and we are also planning a small pilot study of six people with PWS to help determine the best methods to use. Continued on the next page.
Vagus nerve stimulation (VNS) project

We are trying to understand if a device called a vagus nerve stimulator can help people with PWS who sometimes find it difficult to control their behaviour. This device sends small electrical impulses which activate a nerve called the vagus nerve. This nerve connects the brain and the organs in the upper body. We think that this device can help people with PWS to control their temper.

To see whether it works, we have asked seven people with PWS to try this device and their carers are reporting episodes of difficult behaviour. We are currently collecting data, and will keep you informed about the result of this trial. The result of this study could have a great impact on the lives of people with PWS who have behaviour problems, as well as their families and carers.

Understanding early development and mother infant attachment

In collaboration with the ‘baby lab’ led by Dr Vicky Leong in the Department of Psychology here at the University of Cambridge, we are developing a project that will explore in much greater detail the early development of infants with PWS.

This research has taken on a much greater significance since the findings from the studies of the French group led by Professor Tauber, highlighted earlier in this newsletter, on the use of oxytocin in infancy to improve the feeding behaviour and optimise early social development. Dr Leong and her group have developed methodologies for, not only observing infant/mother interactions in detail, but also how to study how different parts of the brain respond depending on what is happening at the time. This is done using special caps worn by the infant and mother that continuously record the electrical activity across different brain regions in a non invasive and acceptable manner.

Dr Leong is planning a pilot study in the first instance and is applying for permission from the ethics committee. Once this is obtained we will be contacting the parents of children with PWS between the ages of two and five years. The long-term aim of this research will be to understand the underlying brain mechanisms that may be faulty in infants with PWS and how that relates to their early development and to the switch from under to over-eating.
Thank you

Please spread the word about these projects. None of them would be possible without the help of you all and particularly people with PWS.

Thank you also to all those who have funded this work. Research is truly a collaborative and international exercise. If you would like further paper copies or to receive an electronic version of this newsletter to pass on to others do let us know.

About us

Emeritus Professor Tony Holland leads our research team. He is an expert in developmental disorders and is Chair of Learning Disabilities at the University of Cambridge. He has worked with people with PWS for many years both as a clinician and as a researcher and has published many journal articles in this field. In addition to working on the projects listed in this newsletter, Tony is also working with Elizabeth Fistein (Adviser on Medical Ethics and the Law, University of Cambridge Clinical School) and with James O’Brien (Chair of the Australian PWSA) and Nicky Davis (Academic Lawyer, Queensland, Australia) on developing guidelines for controlling access to food when supporting adults with PWS. Tony is Patron of the PWSA UK (www.pwsa.co.uk) and President of the International Prader-Willi Syndrome Organisation (www.ipwso.org).

Dr Joyce Whittington joined Tony Holland in 1997 to carry out research into PWS. Their first project was to find out how many people are born every year with PWS in the UK, and to describe all the differences that make people with PWS special. This was a huge amount of work, and they published nine papers and a book to let researchers and other interested people know what they had found. After this study, Joyce worked on many others studies concerning the needs of people with PWS: on mental health, ageing, eating habits, and the IQ of people with PWS and their siblings. Since reaching the University retirement age, Joyce has been writing articles on PWS, reviewing articles on PWS for academic journals, and collecting abstracts of new findings on PWS every three months for the Scientific Committee of the International Prader-Willi Syndrome Organisation (IPWSO www.ipwso.org). Her main interest is in cognition in PWS, and she has recently been appointed an IPWSO adviser. Joyce and Tony have recently published a review of studies into cognition in people with PWS, if you would like a copy of the paper, which is titled Cognition in people with Prader-Willi syndrome: Insights into genetic influences on cognitive and social development, please let us know using the details below.

Dr Katie Manning has been working on PWS for 6 years. She completed her PhD on brain imaging in PWS last year, and she is now working here as a research associate. She is currently working on three main projects. First she is leading the vagus nerve stimulation study (VNS), trying to understand if the new device can help people with PWS and behaviour problems. Secondly she is working on a new study monitoring mental health in PWS and thirdly she is analysing brain imaging data to understand how the brains of people with PWS work. Katie’s review of brain imaging in PWS was published in the journal Diseases and if you would like a copy please contact us using the details below. Katie is also working with Dr Tony Goldstone and brain imaging experts James Cole and Adriana Azor at the Hammersmith Hospital using an advanced analytical technique to examine differences in brain development in people with PWS.

Lucie Aman is a research assistant. She helps Katie with the VNS study, and the mental health study. She is also working to build a database with names and contact details of people with PWS and their families who want to take part in research. If you want to know more about it email us using the details below.

Contact us

Our website: www.ciddrg.org.uk
Email: ah937@medschl.cam.ac.uk
Phone: 01223 465 216
Write: Agnes Hoctor, Team Administrator
PWS Research Group
Douglas House
18b Trumpington Road
Cambridge CB2 8AL

You are receiving this newsletter because you gave us your contact details to be informed about or to participate in our research. If you would prefer not to receive the newsletter in future please let us know.

Thank you