From Bench to Bedside

Words By ALEX JACKSON
Photos by IAIN GARRETT

A RIDGEWAY psychiatrist has played a key role in a major Oxford health initiative to research and find genes that cause brain abnormalities.

Dr Valerie Murphy has worked as a member of the scientific team behind the project that looks at the genetics of brain malformations and the biology of these diseases, with the aim to develop novel diagnostic tools to aid their diagnosis.

The diverse group of scientists and medical professionals from across Oxfordshire first embarked on the molecular genetics research project almost two years ago with the Wellcome Trust Centre for Human Genetics. The project is funded by the Biomedical Research Centre (BRC) in Oxford, which is ultimately funded by the National Institute for Health Research (NIHR).

"The main aim was that the research carried out would translate directly from work completed in the laboratory (from the bench) at the Churchill Hospital to clinical care (to the bedside)," said Dr Murphy.

"We wanted our studies to lead to the identification of new genes that cause brain abnormalities such as Lissencephaly and Celebellar Hyoplasias. The information is important to the families of those affected by such a condition and all too often a genetic cause is never found."

Working with the Clinical Genetics Department at the Churchill Hospital in Oxford, the team has collected DNA from around 200 people from all over the UK and some from further afield such as Austria, Republic of Ireland and the USA. The DNA of these people was examined using the latest developments in DNA sequencing and a molecular genetics 'chip' containing 'probes'.

"The project has really taken off and through word of mouth at various conferences or in different laboratories across the world, samples have started flooding in.

"In the DNA examples we examined, we found a number of changes in the DNA which are responsible for causing the brain abnormality in these families."

The Oxford Radcliffe Hospitals NHS Trust (ORH) has now adopted the 'chip' that investigates DNA samples as a result of the group's findings. The ORH Trust offer this as part of the investigations staff carry out for families when a child is first presented to the Clinical Genetics Department with a brain abnormality.

Dr Murphy added: "I've been involved in science for many years, but never seen it directly translate into clinical practice with such a clear benefit.

"This in my mind is a direct example of how Ridgeway is supporting clinically relevant research which is making a difference to the lives of those with Learning Disabilities."

Scientific Team

Professor Jonathan Flint is the head of the Psychiatric Genetics Group at the Wellcome Trust Centre for Human Genetics Oxford.
Dr David Keays completed his doctorate at the University of Oxford under Professor Flint. He has just established his own laboratory at the Institute of Molecular Pathology in Vienna, and continues to investigate genes that cause neurodevelopmental disorders.
Dr Valerie Murphy is an academic clinical fellow in Learning disability psychiatry in Oxford.

The Clinical Genetics Team

Dr Usha Kini is a Consultant Clinical Geneticist working at the Regional Genetics Department, The Churchill Hospital, Oxford.
Dr Helen Stewart is a consultant clinical geneticist in Oxford and provides a clinical service for people with learning and development difficulties, as well as structural brain abnormalities.

The Neuropathology Team

Dr Alexander Jeans is an Academic Clinical Lecturer based in the Department of Clinical Neurology, University of Oxford.

For more information , please visit ***www.brainabnormalities.org.uk***

Dr Valerie Murphy