New Zealand Brain Research Institute

Huntington's Newsletter

Autumn 2014

Winter is nearly upon us, but here is our long overdue Autumn newsletter. Our previous newsletter mentioned the screening of the film "The Inheritance" (by Jeff McDonald), a movie that relates the life of a Wellington family with Huntington's Disease. For sixty of us, the screening was an opportunity to get together, ask questions or simply reflect.

Getting in touch with other people and families affected by HD can be a powerful way to find suppor t and help. Check below for two ways to stay connected: one via Facebook, and the second one by reading and leaving your comments on the beautiful blog of young Lucy in Gore .- this newsletter has been compiled by Laura, Research Coordinator at the NZBRI.

One small needle, a huge gain for Huntington's research !

As all our trial and research participants know, blood and/or urine collection is one of the key assessments of a study visit. Over the last few years, during our involvement with the CREST-E trial, we have sent more than 400 blood samples and 150 urine samples to Massachusetts General Hospital in the USA. Those samples, linked to the anonymous clinical information and data entered in the study database, were stored in the Canterbury Health Laboratory freezer at minus 80 C for a few months before being shipped twice a year by courier to the USA.

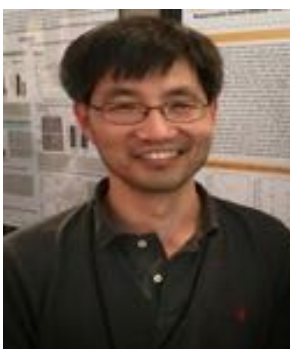
Some blood and urine samples from our Creatine trial participants. They will be carefully packaged and shipped on dry ice to USA.

It may not look like much, but the collection of blood/ DNA/ urine samples and medical information is super important for research.

By making a donation of your blood and clinical information, researchers can look a variety factors and pathological mechanisms that may or may not play a part in Huntington's Disease. They can determine if some chemicals found in the blood might be a marker of progression or resistance; they will also have a better understanding of genetic factors which influence the clinical features of HD (see below). This will ultimately improve the diagnosis and treatment of HD.

Disease modifiers.

I recently listened to a presentation by Jong-Min Lee from Massachusetts General Hospital on genetic variations and HD.
He shared the results of the Genome Wide Association Study 2008 and 2012, two very large studies looking at the roles that other genes play in the onset of HD.



Jong-Lee Ming, Massachusetts General Hospital

While the rate of pathological changes in the brain is vastly determined by the length of the CAG repeat on the Huntington gene (or "the number of repeats", as we sometimes call it), the variations in symptoms presentation strongly suggest that other factors also play a role. Some of those factors are called "**disease modifiers**" because they delay or accelarate the onset of symptoms.

The research team looked at DNA samples of more than 4000 people with Huntington's Disease and have identified genes on three chromosomes that have a significant effect on the disease onset. For instance, chromosome 15 contains both good and bad modifiers as it may delay the onset of HD by up to 6 years or hasten the onset by 18 months.

The next step could be to identify the good gene on chromosome 15 and see how it works, to ultimately make up a pharmaceutical compound that replicates the positive effect of this chromosome.

"If we manage to identify and use all the disease modifiers, we might be able to delay the onset of HD by 20-25 years" – Jong-Min Lee.

This discovery has only been possible when the team was able to access a large bank of blood samples that allowed for strong statistical evidence.

Brain compensation, or the brain's clever Plan B .

The TRACKOn-HD Study has followed 240 European pre-manifest participants (participants who are not showing any signs of HD yet) between 2012-2014. It is lead by Dr Sarah Tabrizi (UCL Queens Square, London). It is looking at the way the brain regions affected by HD communicate with each other.

Imaging of the brain (MRI) of the participants has shown that changes in the brain are not necessarily associated with a difference in thinking abilites. The researchers noticed that people who are more resilient to change use their brain differently –though unknowingly; some particular communication paths used by the brain are more active in those patients.

The researchers were then able to create a program that trains patients to modify their brain activity, while capturing this on the MRI machine. This led to an improvement on a simple motor task. While this is certainly not a treatment, it shows that the brain is able to cope and compensate for some defects.

Let's start connecting !

If you would like to access the Facebook page set up for our local HD community, please email Amanda at akhampton2@gmail.com. She will then invite you to the group. The group's name is HD Members and Support Group Christchurch. Since this is a private group, no posting is viewable by the public. Once you are a member of that group, you will be able to accept other members.

In her blog, Lucy shares her "unexpected journey" through Huntington's. Please check it at the following address: http://huntingtonshobbitshealingandhope.weebly.com/blogs and feel free to leave a comment. It is a truly eloquent and beautiful blog that will resonate with all of us.

Your suggestions and questions are welcome, please contact Laura at 03 378 662 or laurap@cdhb.health.nz