

Oz Buzz Updates: Day 3

Day 3 of our coverage of the Huntington's disease World Congress 2011 in Melbourne

By Dr Jeff Carroll September 14, 2011 Edited by Professor Ed Wild

ur final daily report from the Huntington's disease World Congress brings together all the live updates from our twitter feed. Video of both Oz Buzz sessions - with news, interviews and features - is on YouTube now and will be available to watch at HDBuzz.net later this week.

Wednesday, September 14, 2011

8:33 - Jeff and Ed are now live from day 3 of the World Congress on HD!



Robi Blumenstein of CHDI, HD World Congress, Melbourne 2011

8:40 - Jeff: HD may be much more common than we thought - Michael Hayden

8:47 - **Jeff:** HD is an increasing burden on the elderly, who in the past may not have lived long enough to have symptoms - Hayden

9:02 - Ed: More on increased 'prevalence' of HD - how it's more common than we thought

9:07 - **Ed:** studies in sperm helping to predict whether a CAG repeat in the 'gray area' will cause problems for next generation

9:13 - We need to move away from talking about 'onset' in Huntington's - symptoms begin gradually over years - Dr Mark Guttman

9:32 - Huntington's disease causes a wide variety of symptoms so doctors should always bear it in mind when seeing patients- Elizabeth McKusker

10:36 - Ed's reporting from the 'Science: omics' session - Jeff's in 'International models of care'.

10:37 - Ed: 'omics' means measuring loads of things at once. Like genomics (looking at tons of genes)

10:45 - **Ed:** Metabolomics is measuring metabolites - small molecules in blood. Wayne Matson's done it in HD & finds interesting changes

10:54 - **Ed:** Levels of a chemical I3PA seem to be reduced in blood of HD patients and HD mice. Not yet clear why but could be useful - Matson

11:02 - **Jeff:** HD patient care in Australia is difficult because of complex health care administration - Andrew Churchyard

11:00 - **Jeff:** Significant numbers of HD families don't interact with the medical system, and we don't know why - Churchyard

11:11 - **Ed:** Brain immune cells called microglia are abnormal in HD mice - Dr Blair Leavitt - are they helping or harming?

11:13 - **Ed:** There are also changes in the mice's brain blood vessels that could increase the cross-talk between brain & body - Leavitt

11:26 - **Jeff:** Access to medical services for HD families in South Africa is limited - Amanda Krause

11:34 - **Jeff:** In black Africans a disease called 'HD like 2' looks very much like HD, but is caused by a different mutation - Krause

11:38 - Ed: Ruth Luthi-Carter examines which genes are more and less activated in different mouse models of HD & compares them to humans

11:51 - **Ed:** These gene activation changes could help us to understand the disease and develop and test new drugs - Luthi-Carter

12:06 - **Ed:** Proteins do stuff by sticking to each other. Mutant and normal huntingtin stick to different groups of partners - Chris Ross

12:11 - **Ed:** An international consortium of researchers is using stem cell models to help understand Huntington's disease

12:18 - Jeff: Francisco Cardoso - new Latin american HD network active at rlah.org

"The HORIZON study recruited rapidly and was run efficiently: good news for future trials of upcoming treatments - Bernhard Landwehrmeyer "

12:20 - **Ed:** The consortium has figured out the recipe for turning stem cells into the neurons most affected by HD. Very valuable research tool

12:27 - Red Latino-Americana de Huntington: investigación de tratamientos efectivos para la Enfermedad de Huntington

12:43 - **Ed:** Comparing gene switching and behavior across mouse models shows that different models mimic different aspects of HD Lesley Jones

12:44 - **Ed:** One of the mouse models called Q150 actually produces less huntingtin protein overall - weird but important to know about - Jones

12:45 - **Ed:** An HDBuzz article on the different mice used in Huntington's disease research is coming soon

13:47 - Ed and Jeff are now reporting from the final science session - late breaking hot topics. Jeff is the first speaker in the session!

13:56 - Jeff studies metabolites in several tissues in HD mice. Tissues like brain, fat and liver are all changed differently by the mutation.

14:00 - Metabolic changes Jeff found in HD mouse blood mirror the changes in the brain - could be useful for studying human patients

14:19 - Dimebon showed no benefit for HD in big trial- HORIZON. The quest for treatments for thinking probs continues.

14:21 - However, HORIZON study recruited rapidly and was run efficiently - good news for future trials of upcoming treatments - Bernhard Landwehrmeyer

14:42 - Chemical 'tattoos' are added to DNA by enzymes. HD messes this up. HDAC inhibitor drugs should help - a trial's underway - Larry Marsh

14:50 - There are many different DNA 'tattoos' = many ways to try to improve things with drugs. Animal studies will identify best ones - Marsh

15:10 - **Jeff:** Clare van Eyk uses fruit flies to try and understand how the mutated huntingtin gene kills brain cells

15:12 - 'RNA', as well as protein might contribute to brain cells dying - Clare

15:13 - Jennifer Thompson is studying the psychiatric symptoms of HD, like apathy and depression, which can be devastating

15:16 - Apathy is incredibly common in HD, and worsens over time - Thompson

15:18 - Interestingly, depression is also common in HD, but doesn't seem to change much over time - Thompson

15:36 - Robi Blumenstein of CHDI - like a chess game, we need to think far ahead if we're

going to beat Huntington's disease

15:40 - Success is a three-legged stool: (1) an effective treatment (2) the ability to test it and (3) enough trial volunteers - Blumenstein

16:00 - Lots of HD family members needed for trials, now & future. Find out how to get involved at Enroll-HD.org

The authors have no conflicts of interest to declare. <u>For more information about our</u> <u>disclosure policy see our FAQ...</u>

GLOSSARY

huntingtin protein The protein produced by the HD gene.

Metabolomics the simultaneous measurement of many metabolites in a sample
Metabolite a chemical produced by cells as they break down fuels for energy
CAG repeat The stretch of DNA at the beginning of the HD gene, which contains the sequence CAG repeated many times, and is abnormally long in people who will develop HD

prevalence A figure estimating how many people there are in a particular population who have a certain medical condition.

stem cells Cells that can divide into cells of different types

microglia the brain's immune cells

neuron Brain cells that store and transmit information

HDAC histone de-acetylases (HDACs) are machines that remove acetyl tags from histones, causing them to release the DNA they're attached to

RNA the chemical, similar to DNA, that makes up the 'message' molecules that cells use as working copies of genes, when manufacturing proteins.

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