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'Buzzilia' from the Huntington's Disease World Congress: day 2

#Buzzilia, day 2: Jeff and Ed's roundup of happenings from the second day of the HD World Congress in Rio de Janeiro



By [Dr Jeff Carroll](#) September 16, 2013 Edited by [Dr Ed Wild](#)

Our second daily report from the World Congress on Huntington's Disease in Rio de Janeiro, Brazil.

08:06 - Good morning from Rio for day 2 of the World Congress on Huntington's Disease

08:07 - We begin the day with a session on 'biomarkers' - things we can measure in people that may help us develop and test drugs.

Roberto Weiser fills us in on the situation in Venezuela where HD is unusually common

08:08 - **Alexandra Durr** of Institut Marie Curie, Paris, begins the session with a talk about TRACK-HD.

08:08 - TRACK-HD was led by Prof Sarah Tabrizi in London. Prof Durr leads the Paris site and has a special interest in metabolic changes. TRACK-HD completed this year and has produced a toolkit of measures we can use to run clinical trials.

08:11 - **Durr:** HD has a long presymptomatic phase before symptoms begin, and people can get tested to identify they are at risk. This enables us to study & understand the early stages of the disease and offers the opportunity to treat early to prevent onset. This is a big advantage over other [neurodegenerative](#) diseases, where early diagnosis is not possible. The premanifest phase brings challenges, too. It's very difficult to detect things that are different in premanifest HD.

08:14 - **Durr:** TRACK-HD ran across 4 sites - London, Paris, Leiden (NL) and Vancouver. TRACK-HD was run like a [clinical trial](#) without a drug - emphasis was on collecting the best data and consistency between sites. [MRI](#) scans were used to measure brain atrophy or shrinkage. Everyone's brain is shrinking all the time! In HD it happens a bit faster. The quality of imaging data from TRACK-HD enables many detailed analyses revealing new aspects of HD. The TRACK-HD toolkit enables us to calculate how many people we need for each [clinical trial](#) that's planned

08:28 - **Durr:** despite detectable brain changes, premanifest HD people continue to function normally. The brain is good at compensating. TRACK-HD tells us there is a lot to SAVE in the HD brain. The TrackOn-HD study is focusing on discovering these compensation mechanisms so that we can perhaps enhance them. Several clinical trials are now being planned using discoveries from TRACK-HD to help design them.

08:48 - Next, **Karl Kieburtz** (University of Rochester) reflects on what studies like TRACK-HD and PREDICT-HD have done to help clinical trials. Drugs get approved to treat illnesses, not brain shrinkage or other biomarkers. PREDICT-HD involved a large number of presymptomatic HD mutation carriers across 32 sites. What are the differences between TRACK-HD and PREDICT-HD? TRACK-HD had a broader range of entry criteria, including people with symptomatic HD. Both studies found reliable measures of change across the spectrum of HD. Both studies found brain imaging techniques to be powerful ways of measuring the progression of HD. Simple cognitive (thinking) tests are also good measures of progression.

09:01 - **Kieburtz:** so... how can these studies help us get new drugs tested and approved for HD? Regulatory agencies like the FDA are very cautious with very strict criteria for approving drugs for human use. The FDA has recently indicated it is willing to relax the criteria for Alzheimer's disease. The FDA now appears willing to consider biomarkers in deciding whether to approve a drug for very early brain diseases. Only one drug (tetrabenazine / Xenazine) is specifically licensed for treating [chorea](#) caused by HD in the USA.

09:41 - **Ralf Reilmann** - clinicians working in HD need to give basic scientists good measurements they can use in drug trials

09:43 - **Reilmann:** Are the changes observed in the brains of HD mutation carriers useful as outcome measures in clinical trials? Reilmann is a movement disorders specialist, and argues that movement changes are a very good measure of HD disease progression. The tools used to measure movement changes in HD patients don't work very well in HD mutation carriers that aren't yet sick. Reilmann builds custom machines that are used to measure much more subtle movement changes in HD mutation carriers. Very subtle movement changes can be detected in HD mutation carriers, even 10-20 years before they're expected to be sick. Some of Reilmann's machines require people to push a lever with their tongue, trying to hold steady pressure. HD patients have troubles sticking out their tongue and holding it steady - something noticed by Dr. Huntington in 1872. Reilmann believes that some of the movement changes they've described could be used as measures in trials of HD drugs. Reilmann believes that these early movement changes in mutation carriers don't mean that people with these changes are 'sick'. Reilmann's group has tested their new movement tests in a trial of HD patients, and the tasks worked well, with low '[placebo](#) effects'.

10:04 - **Julie Stout**, of Monash University, studies how HD mutation carriers thinking ability, or 'cognition', changes over life. The specific kinds of problems with cognition vary from HD patient to HD patient. Stout is working to compare different types of questionnaires and tests to measure peoples cognitive difficulties. Even HD mutation carriers that aren't yet sick show subtle changes in cognition - though the measured changes are very small. Measured changes in cognition in HD mutation carriers are slow and subtle over time. Specific tests are pretty good at predicting when people will show symptoms of HD, including a simple circle-tracing task. Stout and others have noticed that changes in brain shape occur long before there are signs of disrupted thinking or movement in HD. The brain has a surprising ability to cope with damage in HD, performing normally for a long time before becoming dysfunctional. Scans suggest that HD mutation carriers might be using their brains differently to solve complex tasks. There is some data that suggests having a more 'passive' lifestyle might lead to earlier symptoms of HD - we should all stay active! **Stout** shows data which suggests stress might be bad for the memory performance of HD patients. Stout summarizes by suggesting there's good evidence that cognitive problems in HD are subject to modification by the environment.

10:33 - **Rachael Scahill**, UCL, uses high powered brain scanners to study changes in the shape of brains in HD mutation carriers

10:36 - **Scahill:** brain shape changes really only matter if they cause symptoms in HD mutation carriers. Data from the TRACK-HD suggests that having faster brain shrinkage is linked to being more likely to progress to overt HD. The good news about all these brain changes is that we might be able to use them in trials, looking for improvement after treatment. We need to use both brain imaging and measures of peoples function to get a good picture of HD progression.

A great feature of this HD World Congress is most scientists are presenting twice - once for fellow boffins + again for family members. We thoroughly endorse the notion of scientists communicating directly and clearly with the people their research directly affects!

12:37 - **Anna Wickenberg**, of Teva pharmaceuticals, is describing the efforts of this pharmaceutical company in HD. Teva has two drugs under development for treating HD, the first attempts to reduce 'inflammation' in the brain. Immune cells, including immune cells in the brain, are hyperactive in HD. Maybe calming them down will help? Teva has a drug, Laquinimod, that calms down brain immune cells. They're interested to test this in HD to see if it's beneficial. Teva's other HD drug is "Pridopidine", formerly called "ACR-16". This drug tested by drug company Neurosearch, now Teva has bought it. Pridopidine was tested in two trials of HD patients - called MermaiHD (Europe) and HART (US + Canada). Pridopidine had beneficial effects on movement symptoms of HD, especially at high doses, but the trials weren't convincing enough

12:49 - Teva and collaborators are planning a new trial in HD patients 2014, results to be published in 2015, trying to find the right dose. This new trial will focus on a large range of movement tests to see if higher doses of pridopidine are helpful. Just in case you're confused - Pridopidine is also called "Huntexil"!

13:00 - Bernhard Landwehrmeyer asks the question - when testing drugs in HD, do we care about making people looking better, or function better? Agencies that approve drugs have suggested methods for finding meaningful measures of symptom improvement in HD patients

13:04 - **Landwehrmeyer:** Finding good measurements of HD symptoms is difficult, because these symptoms change over the course of the disease. Proving that a treatment is beneficial for patients lives is a difficult, but worthwhile, challenge.

The crowd gather for Buzzilia: videos will be up at hdbuzz.net after the conference

13:09 - **Ralf Reilmann** has opened the George Huntington Institute in Germany <http://george-huntington-institut.de/>

13:10 - Reilmann asks the question - are we able to recruit sufficient numbers of HD patients for upcoming clinical trials? More than 12,000 subjects in 18 countries in Europe have joined the REGISTRY study, suggesting patients want to get involved. An [observational](#) study of HD, called Enroll, already has over 1,000 patients in the Americas, Australia and NZ.

13:14 - **Reilmann:** Both the Huntington Study Group and the European HD Network have run good clinical trials in HD - proving it's possible. Based on his experience in HD drug trials, Reilmann believes quick recruitment for future trials is possible.

14:01 - **Landwehrmeyer** opens an afternoon session on what he calls one of the most important skills of people affected by HD - coping. Aam Hubers from Leiden is studying the phenomenon of suicide in HD - what do we know about it?. Published studies suggest that suicide is 2-8 times more common in HD than the general population. HD patients thoughts of suicide seem to occur at a stage when they are losing their independence and becoming more dependent on care. Because existing information was sparse, Hubers set out to study a large number of HD patients across time to understand suicide in HD. Hubers wonders - what factors lead to HD mutation carriers thinking of, or attempting, suicide? About 20% of HD mutation carriers studied by Hubers, whether or not they have symptoms of HD, thought of suicide in the last month. The studies Hubers is conducting suggest that HD patients who report being depressed seem to have higher risk of suicide. Hubers recommends HD patients with depression should be carefully watched for signs of thinking of suicide, based on increased risk

14:26 - **Ken Serbin** - blogger '**Gene Veritas**' - addresses the WCHD about coping. His blog about HD can be found at - <http://curehd.blogspot.com>

14:27 - **Serbin** has deep connections to Brazil, studying Brazilian history by profession. Serbin shares his families struggles with HD, only learning about the disease when his mother began to show signs. Serbin shares with the congress his coping strategies for living in an HD family. First among Serbin's coping strategies is becoming educated about the disease, which he calls the 'basis for advocacy and hope'. Serbin describes how he actively engages with HD by exercising, eating well and taking supplements, hoping they could help. Serbin feels that writing for his blog has been an enormous help in emotionally coping with living in an HD family. Serbin, to HD families: It's time to be an advocate, tell your story!

14:51 - **Serbin:** 'We are living in a new era of hope, as gene carriers'

14:58 - Chales Sabine addresses the WCHD. You can read about his struggle with HD here - <http://moreintelligentlife.com/content/laura-spinney/health-different-battle>

15:02 - Sabine, after testing positive for HD, was told 'There is nothing you can about this disease, just live your life as best you can'. Sabine has shown this was completely incorrect, since becoming actively engaged in the struggle against HD

15:04 - **Sabine:** part of the darkness of HD is the feeling of isolation experienced by HD families. Social media allows young people from HD families to connect, helping to fight the sense of isolation. "No one is going to come along with a magic wand that will immediately cure HD". Instead, he believes that a cure will come in the form of increasingly effective treatments that are developed over time. Developing each of these individual treatments requires the involvement of an engaged HD community. Sabine, to HD researchers: 'Even small advances in the laboratory empower the spirit to go on'

15:17 - **Sabine**, on reasons to have hope: 'The very best of humanity surrounds HD'

16:18 - **Roberto Weiser** describes the experience of HD patients in Venezuela - families whose DNA contribute to the discovery of the HD gene

16:20 - **Peg Nopoulos** addresses the WCHD on the topic of [juvenile HD](#), a subject her team studies at the University of Iowa. Nopoulos notes that the appearance of [juvenile HD](#) can be quite different than the symptoms that occur in adult patients. Nopoulos is interested in changes that occur very early in people carrying the HD mutation, even during childhood and adolescence. Nopoulos is studying brain structure in kids from HD families, to try and understand if there are changes in their early development.

16:44 - **Carlos Cosentino**, an HD physician from Lima Peru, is interested in whether the presentation of HD symptoms varies across countries. The earliest published report of HD in Peru were in 1950. Canete, a region is southern Peru, is a region with a much higher than average incidence of HD. Cosentino believes that HD patients in Peru experience very similar symptoms compared to other HD patients around the world. Francisco Cardoso, from Belo Horizonte Brazil, is talking about 'differential diagnosis' of HD - what if it looks like HD but isn't?

17:03 - **Cardoso:** most cases of suspected HD are HD on genetic testing, but in around 1% of cases the HD test is negative - they don't have HD. These cases are called HD-like disorders, or HD lookalikes. It's difficult to figure out the frequency of these HD-like cases because it varies a lot with geography and docs' expertise. A disease called HD-like-2 or HDL2 is quite common in Brazil because it tends to affect people with African ancestors. Another HD mimic is a condition called SCA17. (These conditions are all rare, remember. In general, if it looks like HD, it's probably HD, especially in a known HD family)

Sunset conclusions

Today highlighted great progress over the past decade in developing biomarkers that will help us make trials - and hence drugs - a reality. We also heard about some specifics of forthcoming trials. There's plenty more to come in the remaining day and a half, so stay tuned!

The authors have no conflicts of interest to declare. [For more information about our disclosure policy see our FAQ...](#)



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- **Glossary**
- **neurodegenerative** A disease caused by progressive malfunctioning and death of brain cells (neurons)
- **clinical trial** Very carefully planned experiments designed to answer specific questions about how a drug affects human beings
- **observational** A study in which measurements are made in human volunteers but no experimental drug or treatment is given
- **inflammation** Activation of the immune system, thought to be involved in the HD disease process
- **juvenile HD** Huntington's disease where symptoms begin before the age of 20.
- **placebo** A placebo is a dummy medicine containing no active ingredients. The placebo effect is a psychological effect that causes people to feel better even if they're taking a pill that doesn't work.
- **chorea** Involuntary, irregular 'fidgety' movements that are common in HD
- **magnetic resonance** A technique using powerful magnetic fields to produce detailed images of the brain in living humans and animals
- [Read more definitions in the glossary](#)

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