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CHDI's chief scientist: 'Not a time for the Huntington's community to rest its laurels'

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At Risk for Huntington's Disease

HD is a genetically caused brain disorder that causes uncontrollable bodily movements and robs people's ability to walk, talk, eat, and think. The final result is a slow, ugly death. Children of parents with HD have a 50-50 chance of inheriting the disease. There is no cure or treatment.

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THURSDAY, FEBRUARY 28, 2019

CHDI's chief scientist: 'Not a time for the Huntington's community to rest on its laurels'

Given the unprecedented interest from biopharmaceutical firms and broadening range of approaches to attacking the disease, the need for the Huntington's disease community to participate in research studies and clinical trials is "exploding," CHDI's head scientist said at the organization's 14th Annual HD Therapeutics Conference yesterday.

"This is not a time to rest on our laurels," said Robert Pacifici, Ph.D., the chief scientific officer for CHDI Foundation, Inc., the nonprofit virtual biotech dedicated to finding HD treatments. "Trials won't go forward without people willing to take the risk of taking a drug never taken before."

Dr. Pacifici's comments came in an interview with me at the conference venue, the Parker Palm Springs hotel in Palm Springs, CA. The gathering of scientists, pharma executives, and advocates ends today.

Critical new research studies

The conference comes just after news of the start of Roche's historic Phase 3 gene-silencing clinical trial and other clinical trial programs (click here to read more).

"It's exciting times," Dr. Pacifici observed. "Now more than ever we need to make sure that these channels of communication between patients and caregivers and researchers remain open and clear so that everybody knows what's going on."

With the advent of more trials, both the pace and number of research projects requiring volunteers is growing rapidly, he explained. One involves a forthcoming brain imaging study (using a PET-ligand).

To help understand the disease and measure the effect of potential drugs in clinical trials, scientists need to find more biomarkers (signals of the disease) in humans. For years, they have collected samples of blood. More recently, they started examining the cerebrospinal fluid and saliva.

Now CHDI wants to collect semen, Dr. Pacifici said. "There are certain things that happen only in those cell types that are going tell us what happens to Huntington's DNA, and that's going to be really critical for our efforts as well," he explained.

CHDI will also establish a "Brains for HD" initiative to request donations of that organ from deceased individuals, Dr. Pacifici added. "Invaluable," he said of the importance of brains in HD research. "Especially the rare young brain, if somebody happens to pass away from something else, and we can harvest the brain before it experiences the ravages of Huntington's disease."

You can watch my interview with Dr. Pacifici in the video below.

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Not a time for the Huntington's community to rest on its laurels

from **Gene Veritas**

23:22

Not a time for the Huntington's community to rest on its laurels from Gene Veritas on Vimeo.

'Huntington's homework'

Dr. Pacifici outlined the conference's three major areas of focus.

He described the first as "Huntington's homework."

"We might not like it, and it might be tough, and it requires some dedication, but we still have basic research to do to understand the underlying pathophysiology of Huntington's," he explained. "There are still some fundamental questions about what's broken in HD that we need to understand, because until you understand what's broken, it's hard to fix it."

To that end, this year's conference talks have included topics such as HD's hampering of the brain's ability to repair itself and the loss of white matter in presymptomatic gene carriers like me.

Modifier genes: a key discovery

A second area involves the discovery of so-called modifier genes – a major advance only made possible with the participation in the research of 9,000 HD family members.

"We now know there are other genetic factors that influence, for any given CAG [the length of the genetic coding for the huntingtin gene], whether you'll get the disease sooner or later," Dr. Pacifici noted.

People with same gene length can get the disease decades earlier or later, he said. "That's a really big range, and what we've found is that the propensity to get it earlier or later is also heritable."

The modifier genes affect onset by as much as a dozen years or more, he added.

"Imagine if we could make a drug that did the same thing – or did it even better," Dr. Pacifici said. CHDI and HD scientists are at work on this task.

'A wonderful time for HD research'

A third major aspect of the conference concerns "genetically identified targets" that are changed in people because of HD. This type of lab research is still in the early stages, Dr. Pacifici observed. However, these targets might also serve as models for drug design.

Dr. Pacifici noted that in the relatively mature field of "huntingtin lowering" – the approach of the Roche drug and of several other companies present at the conference – something "unthinkable" five years ago has occurred: the possibility of a "small molecule" drug. Such a drug would be taken orally, thus avoiding the spinal taps and other highly invasive procedures currently envisioned for these drugs.

On February 26, Anuradha Bhattacharyya, Ph.D., of Ireland- and U.S.-based PTC Therapeutics presented a paper on the company's efforts to develop such a drug.

As a nonprofit, CHDI's job is to provide all firms access to the necessary biotechnological tools they need and to potential volunteers for studies and trials, Dr. Pacifici said. CHDI has spent hundreds of millions of dollars in the effort to treat HD, including grants to scientists and companies.

He concluded: "It's a wonderful time to be a researcher in the area of Huntington's drug discovery and it will soon be an opportune time to engage the broader patient community, because we really need your help."

(Future articles will discuss other aspects of the meeting. For additional coverage, visit HDBuzz.net.)

Posted by Gene Veritas at 9:37 AM

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