





**Dina De Sousa**



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***CONTRIBUTING TO RESEARCH IN  
HD***

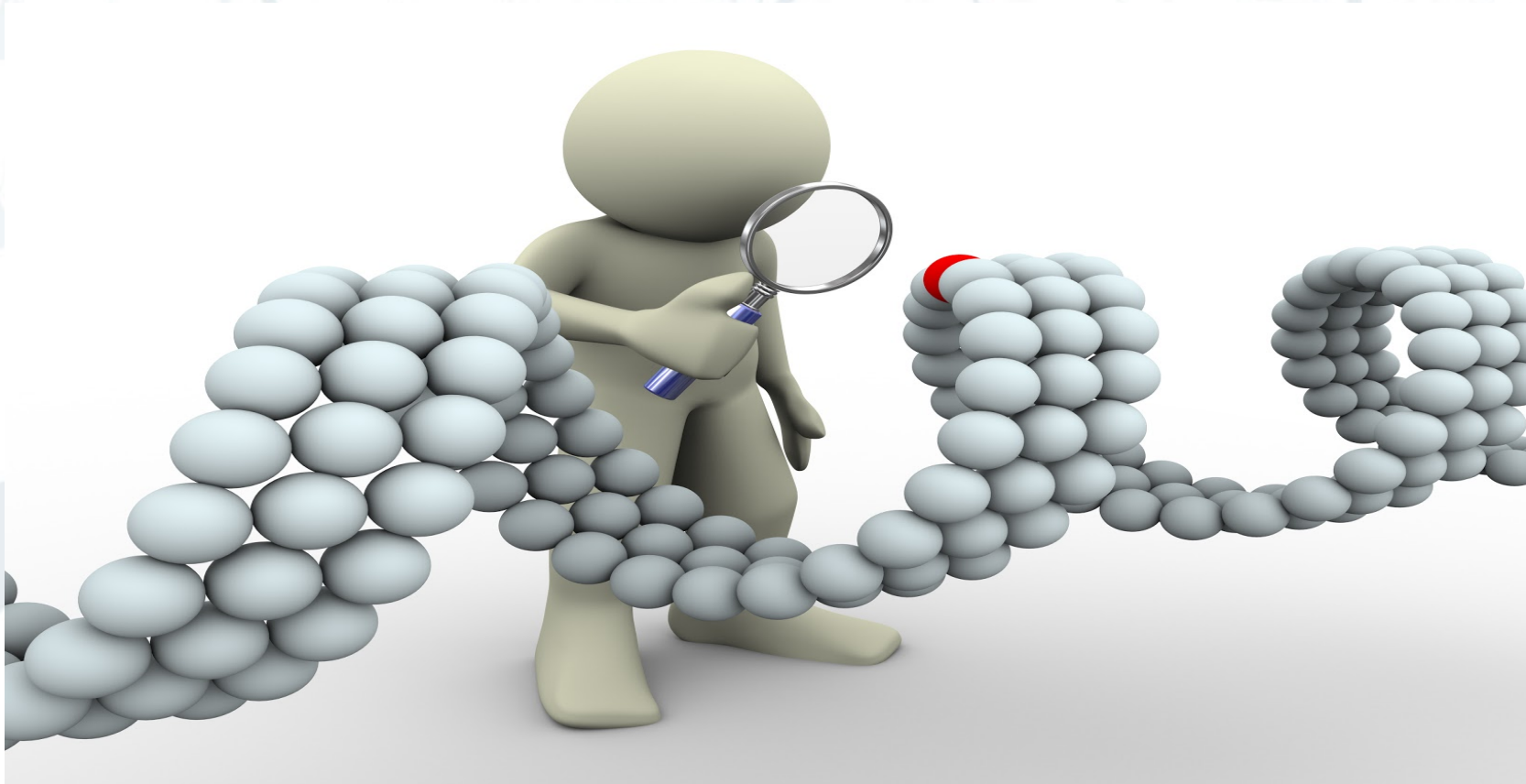
***WHY IT'S IMPORTANT***

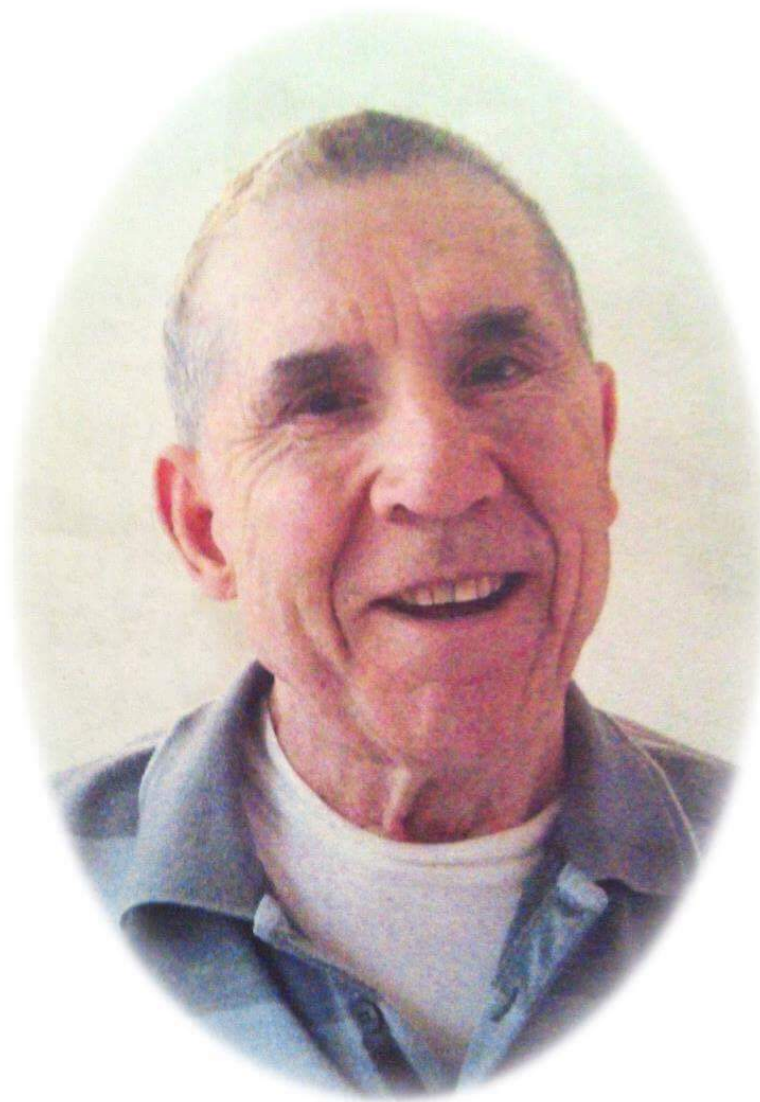
***MY PERSPECTIVE***





# PREDICTIVE TESTING







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# EHDN

EUROPEAN  
HUNTINGTON'S  
DISEASE  
NETWORK

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*Advancing Research, Conducting Trials, Improving Care*

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# WARSAW 2015





[www.eurohuntington.org](http://www.eurohuntington.org)







# scottish huntington's association



coalition for patient engagement



Observational/  
Information Gathering

TrackOn>HD

Identifying Predictive Measures



Predictive Biomarker Detection



Neural Feedback Training



**FOR HIRE: FREE**



A **INTERVENTIONAL** trial tests either a new drug or a non-drug therapy (exercise or diet) to see if it can improve symptoms or stop the disease.

It is set so some volunteers get the drug, others get an inactive pill (placebo).

Both groups are monitored to see who does better.

An **OBSERVATIONAL** study like usually just takes measurements to explore how a disease changes over time and what affects the health of people with a particular disease.

There is no drug being tested.







# The New York Times

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NEW YORK, WEDNESDAY

## Team Pinpoints Genetic Cause Of Huntington's

### 10-Year Quest Produces Key to Brain Disease

By NATALIE ANGIER

After 10 backbreaking years in a research purgatory of false leads, failed experiments and long stretches of mordant despair, an international team of scientists says it has discovered the most coveted treasure in molecular biology, the gene behind Huntington's disease.

Now that they have the gene in hand, researchers say they can begin making headway in understanding the disorder, a neurodegenerative illness that usually strikes a person in the 30's or 40's, insidiously destroys body and sanity alike, and kills within 10 to 20 years.

Huntington's disease afflicts about 30,000 Americans, and as many as 150,000 are at risk of developing it. The best-known victim was the folk singer Woody Guthrie.

#### Holy Grail of Genetics

The first clues to the gene's location came in the early 1980's, at the dawn of the contemporary era of molecular genetics. But researchers soon ran into a succession of snags that transformed the search into an irresistible if irritating quest that seduced some of the biggest names in biology.

Of particular interest to scientists was the mutation that causes the disease, one they have lately seen in genes that cause other illnesses, a sort of molecular accordion effect in which a segment of the gene is abnormally expanded and repeated over and over.

#### Key to the Puzzle

Researchers emphasized today that much work needed to be done before they could use the mutation as any sort of precise prognostic tool. Nor does finding the gene mean that a treatment for the disease is imminent. But the discovery is essential to cracking the puzzle of Huntington's.

The finding will be reported Friday in the journal *Cell*, crediting as author the Huntington's Disease Collaborative Research Group. This reflects a rare instance of sustained scientific cooperation in which six laboratories in the United States, England and Wales shared their data and ideas.

Biologists everywhere, even those who were competing against the victorious consortium, greeted the news with uninhibited joy and deep relief.

"This is a landmark event," said Rick Myers of the University of California at San Francisco, who had independently been seeking to isolate the gene. "I can hardly believe that it's finally here after all these years." David L. Nelson, a molecular geneticist at Baylor College of Medicine in Houston, said: "I think this is fantastic. It's taken so long to find this gene, and there's been such bizarre speculation about why people couldn't get it, that I'm relieved and thrilled to see the search has ended." Dr. Nelson had worked on another disorder, called fragile X syndrome, which is caused by a similar abnormal gene expansion.

glia, a region of the brain that controls movement and possibly cognition.

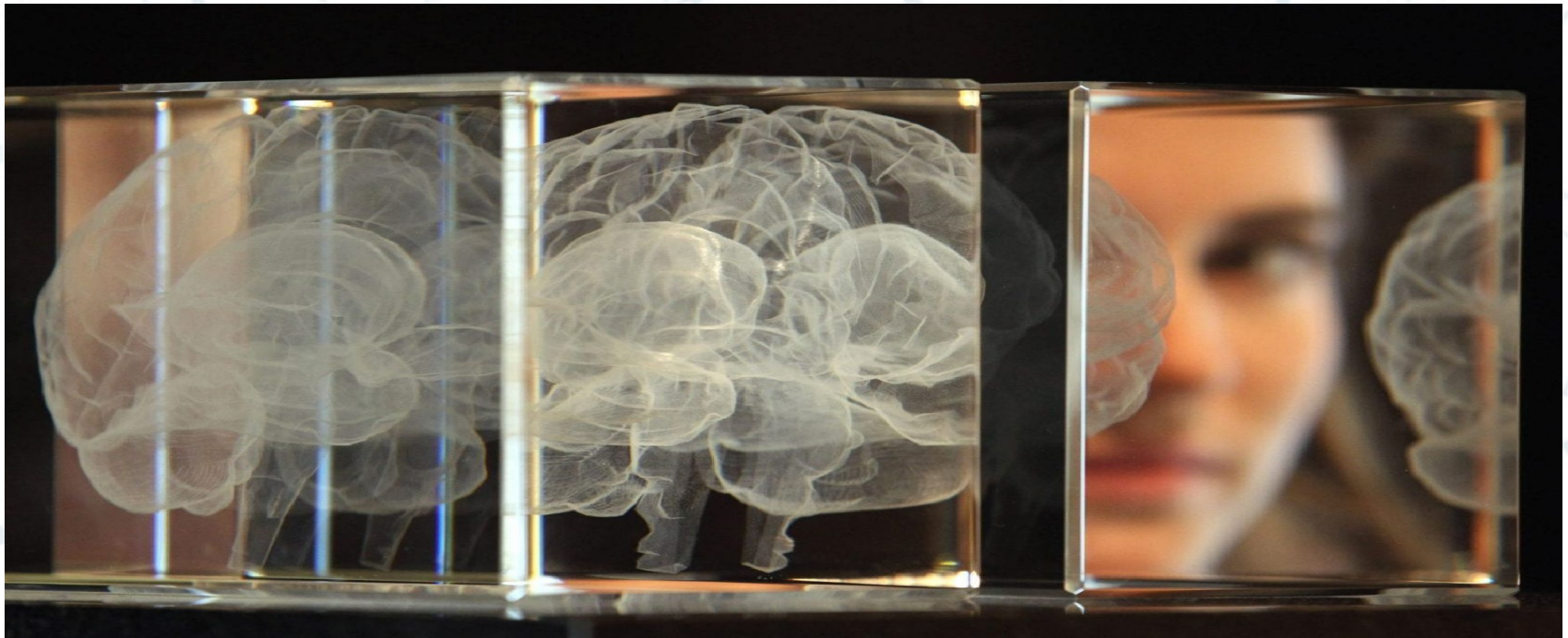
chemical initials CAG. In normal people, the gene has from 11 to 34 of these







# Huntington's disease: scientists hail 'biggest breakthrough in 50 years'



Huntington's disease, also known as Huntington's chorea, is an inherited disorder that results in death of brain cells. (Photo by Dan Kitwood/Getty Images)





Roche announces de  
'pivotal' huntingtin-

The GENERATION-HD1 trial will test whether  
- slows the progression of Huntington's d



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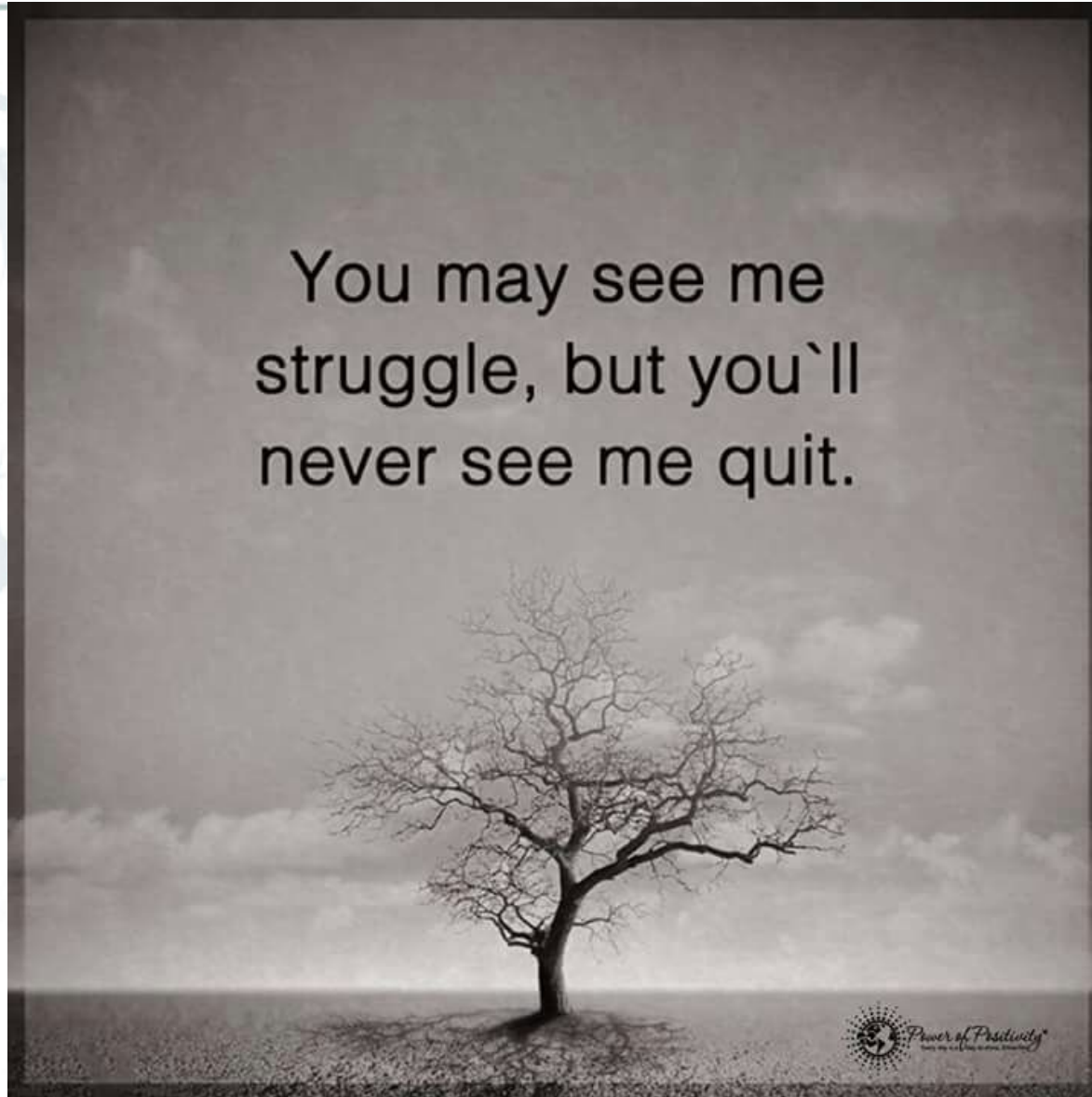








You may see me  
struggle, but you`ll  
never see me quit.





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<http://eurohuntington.org/how-can-you-contribute-2/>





<https://foxtrialfinder.michaeljfox.org/understanding-clinical-trials/Why-Do-Trials-Need-Volunteers/>



**THANK YOU**