HDdennomore
MAI PIÙ NASCOSTA • OCULTA NUNCA MÁS

POPE FRANCIS' SPECIAL AUDIENCE WITH THE HUNTINGTON'S DISEASE COMMUNITY IN SOLIDARITY WITH SOUTH AMERICA
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ABOUT HUNTINGTON'S DISEASE

FAST FACTS

- Huntington's disease (HD) is a fatal genetic disorder. It has no cure.
- It affects an estimated one million people globally. The world's highest concentration of HD is found in pockets of South America.[1]
- HD manifests as a triad of motor, cognitive, and psychiatric symptoms which progress over many years until death. Symptoms generally present between ages 30 and 50.
- Stigma around HD has forced those affected by the disease to hide in shame for generations, hindering the availability of services to improve their daily lives. Tragically, suicide is common.
- Every child of a parent with HD has a 50/50 chance of carrying the faulty gene that causes HD.
- Treatments to manage symptoms are currently available and potential new medicines that could help to slow its progression are currently being tested.

WHAT IS THE GENETIC CAUSE OF HD?

Researchers identified the gene that causes HD in 1993 by studying families in Lake Maracaibo, Venezuela. HD is caused by an expanded CAG repeat length on the HTT gene, which codes for an important protein in brain cells called huntingtin. This gene expansion causes cells in parts of the brain to die.

As the brain cells die, a person with Huntington's becomes less able to control movements, recall events, make decisions and control emotions. The disease leads to incapacitation and, eventually, death (generally due to other health complications, like pneumonia).

What are the symptoms of HD?

- Personality changes, mood swings & depression
- Forgetfulness & impaired judgment
- Unsteady gait & involuntary movements (chorea)
- Slurred speech, difficulty in swallowing & significant weight loss
- Stiff or awkward walking, increased clumsiness or changes in speech for JHD

When do HD symptoms typically start?

- Symptoms of Huntington's disease usually develop between ages 30 and 50 but can appear as early as age 2 or as late as 80.
- In approximately 10% of cases, HD affects children or adolescents. Juvenile HD (JHD) typically progresses more rapidly than adult onset HD.

WHY ARE FAMILIES WITH HD STIGMATIZED?

People with symptomatic HD have uncontrolled, irregular, rapid, jerky movements, called chorea, and cognitive challenges such as loss of speed and flexibility in thinking and memory loss.

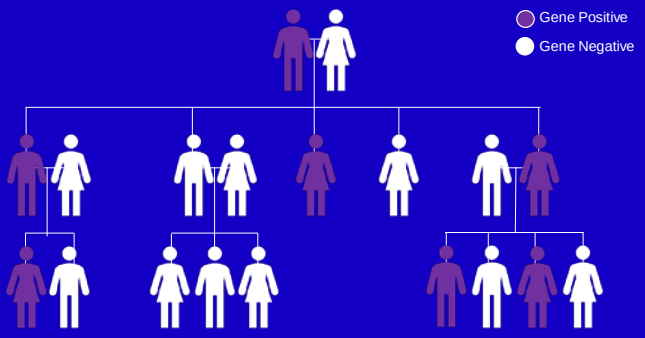
They also have psychiatric disorders including depression and anxiety. In some areas of the world, society has shunned these families, leading those affected to fear public criticism, discrimination and unfair treatment.

WHO IS MOST AT RISK?

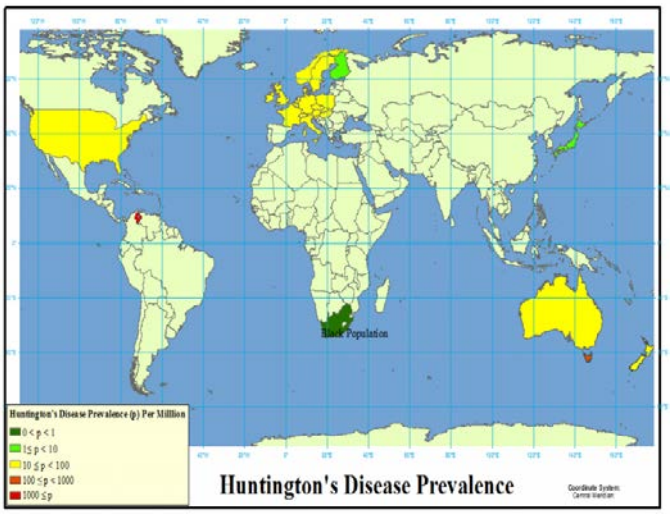
EVERY CHILD OF A PARENT WITH HD HAS A 50/50 CHANCE OF INHERITING THE EXPANDED GENE THAT CAUSES THE DISEASE.

If the child has not inherited this expanded gene, he or she will not develop the disease and cannot pass it on to their children.

Diagnostic genetic testing is available for symptomatic individuals. Predictive genetic testing is available for those with a family history of HD.



WHAT IS THE PREVALENCE OF HD GLOBALLY?



HD is currently found across different countries and ethnic groups, with the highest frequencies found in South America.

In Western countries, it's estimated that about five to seven people per 100,000 are affected by HD.

The world's highest concentration of HD has also been found in pockets of South America where the prevalence can be as high as about 700 per 100,000.[2]

In Africa, the Middle East and Asia, the prevalence of HD is low.

1 https://ghr.nlm.nih.gov/condition/huntington-disease
2 https://www.huntingtonsnsw.org.au/information/hd-facts/how-common