Unveiling the Causes of Fatal Huntington’s Disease

Description

BOSTON — Researchers are making strides in unraveling the enigma behind Huntington's disease, a debilitating and fatal hereditary condition that impacts people in their prime. This disorder causes the degeneration and death of nerve cells in specific brain regions.

Although the genetic mutation responsible for Huntington's has been known for some time, scientists have long puzzled over why individuals with the mutation from birth often exhibit no symptoms until later in life.

Recent studies reveal that the mutation remains benign for decades. However, it gradually enlarges until it surpasses a critical threshold, at which point it begins to produce harmful proteins, leading to the death of affected brain cells.

"The puzzle in our field has been why a genetic disorder manifests later in life despite being present from conception," remarked Dr. Mark Mehler, director at the Institute for Brain Disorders and Neural Regeneration at the Albert Einstein College of Medicine. He praised the research as a "landmark" study, addressing persistent questions in the field.

As brain cells die, individuals experience difficulties with movement, cognition, and behavior. Symptoms like involuntary movements, unstable gait, personality shifts, and poor judgment typically manifest between ages 30 and 50, progressively deteriorating over 10 to 25 years.

The study, conducted by scientists from the Broad Institute of MIT and Harvard, among others, examined brain tissue from people with Huntington's and those without. They scrutinized a specific gene with a DNA segment where the sequence CAG repeats at least 40 times in individuals with the disease, compared to 15-35 times in those without. Over time, these repeats can expand significantly, reaching a tipping point where neurons begin to die.

The findings indicate that these DNA stretches grow slowly in the first two decades but accelerate once they reach 80 CAGs. Researchers aspire that their discoveries might pave the way for new strategies to delay or even prevent Huntington's, offering hope to the 41,000 Americans affected.

Efforts to stop the DNA repeat expansion could potentially be more effective than current treatments, which only manage symptoms. While success isn't guaranteed, McCarroll notes that many companies are now investing in these promising avenues.

Vocabulary List:

1. **Degeneration** /ˌdɛdʒ.əˈnɛr.eɪ.ʃən/ (noun): The process of losing functional abilities or strength.
2. **Mutation** /mjuːˈteɪ.ʃən/ (noun): A change in the DNA sequence of an organism.

3. **Manifest** /ˈmænɪfɛst/ (verb): To display or show (a quality or feeling) by one’s acts or appearance.
4. **Cognition** /kɒgˈnɪʃ.ən/ (noun): The mental action or process of acquiring knowledge and understanding through thought experience and the senses.
5. **Neurons** /ˈnʊrɒnz/ (noun): Cells in the nervous system that transmit information.
6. **Strategies** /ˈstrætɪdʒiz/ (noun): Plans of action designed to achieve a long-term or overall aim.

Comprehension Questions

Multiple Choice

1. What is the primary focus of researchers in relation to Huntington's disease?

   Option: Treatment strategies
   Option: Genetic mutation analysis
   Option: Symptom management
   Option: Brain tissue examination

2. When do symptoms of Huntington's disease typically manifest in individuals?

   Option: At birth
   Option: During childhood
   Option: Between ages 30 and 50
   Option: After the age of 60

3. What is the critical threshold that triggers the production of harmful proteins in Huntington's disease?

   Option: 20 CAGs
   Option: 40 CAGs
   Option: 60 CAGs
   Option: 80 CAGs

4. Which institute was involved in conducting the study on Huntington's disease?

   Option: Albert Einstein College of Medicine
   Option: Johns Hopkins University
   Option: Broad Institute of MIT and Harvard
   Option: Mayo Clinic

5. What type of cells degenerate and die in specific brain regions in Huntington's disease?

   Option: Blood cells
   Option: Nerve cells

Option: Muscle cells
Option: Skin cells

6. At what point do symptoms of Huntington's disease start to progressively deteriorate?

Option: After 5 years
Option: Over 20 years
Option: Within 1 year
Option: Over 10 to 25 years

True-False

7. Huntington's disease is a contagious condition.

8. Symptoms like involuntary movements and poor judgment are associated with Huntington's disease.

9. Dr. Mark Mehler is the director at the Institute for Brain Disorders and Neural Regeneration at Johns Hopkins University.

10. Efforts to stop DNA repeat expansion in Huntington's disease may be more effective than current symptom management treatments.

11. Researchers aim to find new strategies to delay or prevent Huntington's disease.

12. The DNA repeats in Huntington's disease grow rapidly in the first two decades.

Gap-Fill

13. The sequence CAG repeats at least _____ times in individuals with Huntington's disease.

14. Symptoms of Huntington's disease typically manifest between ages 30 and _____.

15. The DNA repeats in Huntington's disease accelerate once they reach _____ CAGs.

16. Efforts are being made to pave the way for new strategies to delay or even prevent Huntington's disease, providing hope for the _____ Americans affected.

17. Dr. Mark Mehler praised the research on Huntington's disease as a "_____" study.

18. Huntington's disease causes difficulties in movement, cognition, and _____.

Answer

Multiple Choice: 1. Treatment strategies 2. Between ages 30 and 50 3. 80 CAGs 4. Broad Institute of MIT and Harvard 5. Nerve cells 6. Over 10 to 25 years
True-False: 7. False 8. True 9. False 10. True 11. True 12. False
Gap-Fill: 13. 40 14. 50 15. 80 16. 41,000 17. landmark 18. behavior

Answer

CATEGORY

1. Health - LEVEL4

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