What causes Huntington's disease (HD)?

HD is an illness of the nervous system caused by a defective gene and has a broad impact on a person's functional abilities[1], usually impacting how a person:

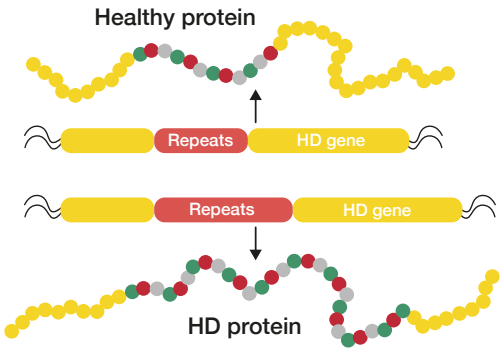
Thinks

Behaves

Moves

In HD, the gene that determines the structure of the huntingtin protein is affected[1–3]

The DNA of this gene is faulty, with an expanded repeating genetic sequence of three nucleotides, cytosine-adenine-guanine (**CAG**)[1–3]



The mutant huntingtin protein (mHTT) created from the faulty gene has an abnormally long structure compared with the healthy, wild-type protein (wtHTT), and is toxic to the brain and nervous system[1,3,4]

The more repetitions of CAG in the huntingtin gene, the earlier the clinical onset of HD[1]

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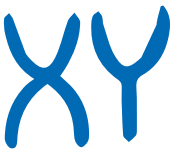


Everyone inherits two copies of the huntingtin gene from their parents – one on each chromosome – but only one chromosome needs to have a mutated huntingtin gene to cause HD[1]

Each child of an affected parent has a

50:50

chance of inheriting the genetic mutation that causes HD[1]

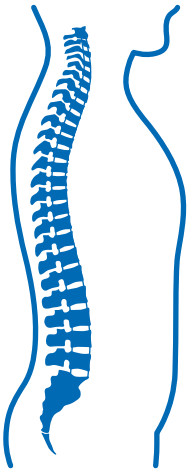
Men and women are equally likely to inherit the mutation[1]

The mHTT protein is toxic to the nervous system, which causes abnormalities in the processes that nerve cells use to stay healthy[4–6]

This may result in changes in the communication between nerve cells, or even cause them to die[1,5–8]

The severity of a patient's HD symptoms is dependent on the amount of mHTT protein[9]

The concentration of this mutant protein can be measured in cerebrospinal fluid (CSF)[10]



Reductions in mHTT protein levels in CSF may be associated with therapeutic benefits in people with HD[10]

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