Huntington's Disease

Huntington’s disease, also referred to as Huntington’s Chorea, is a neurodegenerative disorder affecting the caudate of the basal ganglia resulting in characteristic movements as well as cognition and mood changes. The disease displays an autosomal dominant inheritance pattern and is caused by a trinucleotide repeat. The excessive accumulation of nucleotides in the DNA results in the overexcitation of neurons from glutamate and excitatory neurotransmitter overactivity. As CAG repeats accumulate with each generation, the disease gets more severe and earlier in onset. This pattern of worsening severity is a phenomenon called anticipation. Patients usually present in adulthood with depression, aggressive tendencies, and jerky movements referred to as choreiform movements. As the disease progresses, the movements become more pronounced and patients often develop an early onset dementia.



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Pathophysiology

Autosomal Dominant
Domino
This disease is inherited in an autosomal dominant fashion.

Trinucleotide Repeat
DNA-strand with Repeats
Huntington's Disease is caused by a trinucleotide repeat expansion. This repeat expansion in the Huntingtin gene results in a mutant form of protein, which eventually causes damage to certain neurons.

CAG Repeats
(C) Cat, (A) Apple, (G) Glue Repeats
The CAG triplet is the genetic code for glutamine. The CAG expansion causes a repeating glutamine chain that alters the protein. This mutant form of protein causes damage to certain neurons.

Chromosome 4
Chromosome (4) Fork
The repeat of CAG occurs in chromosome 4.

Anticipation
Progression from Grandpa to Child
Anticipation is the genetic term that describes a phenomenon in which the symptoms of a genetic disorder become apparent at earlier ages, as it is passed from one generation to the next. In Huntington's disease, anticipation is thought to occur as the trinucleotide repeat expands, as it is passed from generation to generation.

Caudate
Cod-dates
The caudate is part of the basal ganglia involved in higher-order motor control, learning, memory, and emotions. It is characteristically affected in Huntington's disease.

Decrease of ACh
Down-arrow A-seagull-cola
In Huntington's Disease, the caudate loses the neurotransmitter, acetylcholine.

Decrease of GABA
Down-arrow GABA-goose
In Huntington's Disease, the caudate loses the neurotransmitter GABA, an inhibitory neurotransmitter.

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Signs and Symptoms

Chorea

Korean-flag

Chorea is described as jerky, random uncontrollable movements, and these are often characteristic of Huntington's disease.

Dementia

Demented-D-man

Patients often display cognitive dysfunction, including dementia.

Depression

Depressed-emo

Patients often display neuropsychiatric dysfunction, including depression.

Glutamate Toxicity

Glue-tomatoes

Glutamate is an excitatory neurotransmitter that is believed to be involved in the death of nerve cells of people with Huntington's Disease, due to over-excitation, which eventually leads to cell death.

Neuronal Death via NMDA Binding

NMDA-roots

Glutamate binds to NMDA receptors, resulting in over-excitation and eventually cell death.

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