# JUVENILE HUNTINGTON'S DISEASE (JHD)

## What it is:

Huntington Disease Brain  Normal Brain

Courtesy of Dr. Jean Paul Vonsattel Columbia University

Huntington's Disease (HD) is the breakdown of nerves cells in the brain. When HD appears in someone under the age of 20, it is recognized as JHD. It is characterized by loss of control over:

- Movement
- Emotion
- Thinking

## Behaviour

- A child or youth with JHD will experience the loss of abilities, competencies and independence. This can cause feelings of frustration, anger, sadness, fear and grief. It can become difficult for the child or youth to regulate stimuli. Obsessive thoughts or irrational fears can add additional stressors, and at times, may cause them to express negative emotions through aggressive behaviour.

## Impact on Thinking

- JHD is a gradually progressing disease and symptoms can occur slowly – possibly a number of years before an official diagnosis can be made. Children may begin to struggle performing tasks they had mastered before (e.g. writing, reading and counting). It can become increasingly difficult for a child or youth with JHD to learn new information and form new memories.

## Diagnosis: Genetic Testing

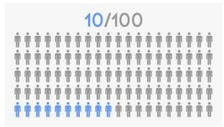
Physicians can seek confirmation of a suspected neurological disorder through Diagnostic Genetic Testing. In JHD, the CAG repeat expansion – which is the genetic mutation that leads to HD – involves a larger number of repeats than those found in adult HD.

## First Symptoms

The early signs of juvenile Huntington disease will represent a noticeable change from the past behaviour of the child. These symptoms may include:

- Rigidity
- Slowness and stiffness
- Awkwardness in walking
- Diminished coordination
- Personality changes
- Changes in behaviour and poor judgment
- Slowness in responding
- Variable/poor school performance
- Difficulty in learning new information
- Unable to do things previously learned

**Less than 10%** of people get JHD

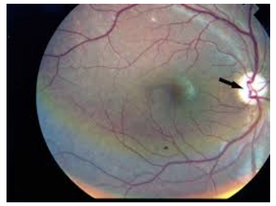



www.reallygreatsite.com

# 5 things to know about WOLFRAM SYNDROME

## 1 What is it?

**Wolfram syndrome is a progressive, neurodegenerative disorder that damages your brain and other tissues in your body.**

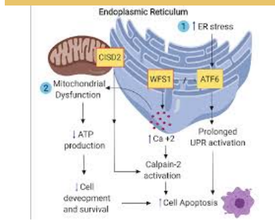



## 2 Symptoms:

The primary symptoms of Wolfram syndrome (diabetes mellitus, optic atrophy, diabetes insipidus, hearing loss and nervous system dysfunction) can emerge at different ages and change at different rates. Diabetes and vision changes before age 15 are usually the first symptoms. Eventually, impaired brain function can lead to early death.

## 3 Testing

For the diagnosis of Wolfram syndrome, genetic testing has been proven to be useful to confirm the diagnosis.

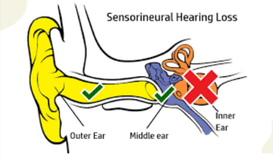



## 4 Types

There are two types of Wolfram Syndrome: Wolfram syndrome 1 (WFS1) and Wolfram syndrome 2 (WFS2)

## 5

Dominant mutations in the WFS1 gene are a common cause of low-frequency sensorineural hearing loss




reallygreatsite.com