Kennedy's Disease

FACT SHEET | WWW.ALS.CA

Kennedy's Disease is a rare genetic disease with symptoms that are similar to ALS. It is also called spinal and bulbar muscular atrophy.

Cells in the human body have 23 chromosome pairs containing our DNA. One of these pairs is the sex chromosomes, which determine if someone is male or female.

The Kennedy's Disease mutation is found on the X chromosome, which means it is an X-linked disorder. X-linked disorders mainly affect men. This is because men have just one X chromosome, while women have two. Women are almost always unaffected, since their other X chromosome can typically compensate.

Women with the mutation are carriers, meaning their sons have a 50% chance of inheriting the condition, depending on which X chromosome is passed on.

The mutation that causes Kennedy's Disease occurs in the androgen receptor gene, which encodes the androgen receptor protein. Androgen receptors respond to the male sex hormones testosterone and DHT. A person with Kennedy's Disease has normal levels of these hormones, but abnormal receptors.

Fragments of the affected receptor proteins build up on the motor neurons, leading to the gradual loss of motor neurons.

Estimates of the prevalence of Kennedy's Disease range between 1 in 50,000 to 1 in 350,000 men. Symptoms tend to occur between ages 18-64, and typically begin in a man's 30s or 40s.

SYMPTOMS

Kennedy's Disease has similar symptoms to ALS, although the symptoms have a different cause. Because the symptoms are similar, and because Kennedy's Disease is so rare, it is sometimes misdiagnosed as ALS.

A genetic test can confirm a diagnosis of Kennedy's Disease. A person with Kennedy's Disease will have more than a piece of the androgen receptor gene. These repeats prevent the normal function of the androgen receptor.

The main symptom of Kennedy's Disease is progressive, adult-onset muscle weakness and wasting in the limbs. Later in the course of the disease, a person may experience weakness in the muscles of the face and tongue, causing difficulty with swallowing and speaking.

Androgen receptors are found in high concentrations in the motor neurons, which are involved in the control of voluntary muscles. Like in ALS, when these motor neurons are damaged, people with Kennedy's Disease experience weakness and wasting of the muscles.

Unlike ALS, Kennedy's Disease has a much slower disease progression. People with Kennedy's Disease typically have a nearly normal lifespan.

Other symptoms of the disease include infertility, testicular atrophy, enlarged breasts (gynecomastia), low sperm count, and erectile dysfunction, all of which have to do with the androgen receptors.

There is no treatment that can cure Kennedy's Disease. Treatment involves managing the symptoms. This may involve physiotherapy, assistive devices such as braces and walkers, gynecomastia surgery when necessary, prevention of pneumonia and asphyxia, as well as exercise.

RESEARCH & EXPERIMENTAL TREATMENTS

Researchers have been working to understand and develop treatments for Kennedy's Disease for many decades.

This work has resulted in a number of clinical trials and interventions such as exercise and leuprorelin acetate are promising potential treatments that may have an effect on disease progression.

SUMMARY

- Kennedy's Disease is a genetic disorder affecting the male sex hormone receptors. It is an X-linked disorder affecting mainly men.
- The symptoms of Kennedy's Disease are similar to those of ALS.
- Symptoms tend to begin in a man's 30s and 40s, and almost always between the ages of 18-64.
- Symptoms include muscle weakness, infertility, enlarged breasts, testicular atrophy, low sperm count, and erectile dysfunction.
- There is no cure for Kennedy's Disease, but research has revealed potential promising treatments.

People with Kennedy's Disease and their families can find more information from the Kennedy's Disease Association, https://www.kennedysdisease.org/.

ADDITIONAL RESOURCES

Kennedy Disease
About Kennedy's Disease
X-linked spinal and bulbar muscular atrophy (Kennedy's Disease): The first case described in the Brazilian Amazon
Beyond motor neurons: Expanding the clinical spectrum in Kennedy's Disease
Kennedy's Disease

KNOW THAT WE ARE HERE TO HELP I For people and families living with ALS in Ontario, ALS Canada can assist in connecting you to support services, equipment, and ALS clinics. Whether you are a person living with ALS, a family member or a caregiver, we will strive to support you along this journey. If you live outside of Ontario, please contact your provincial ALS Society for information on support available in your region. Learn more at www.als.ca.

Special thanks to everyone who helped write and review this fact sheet.

* Last updated 10/2020