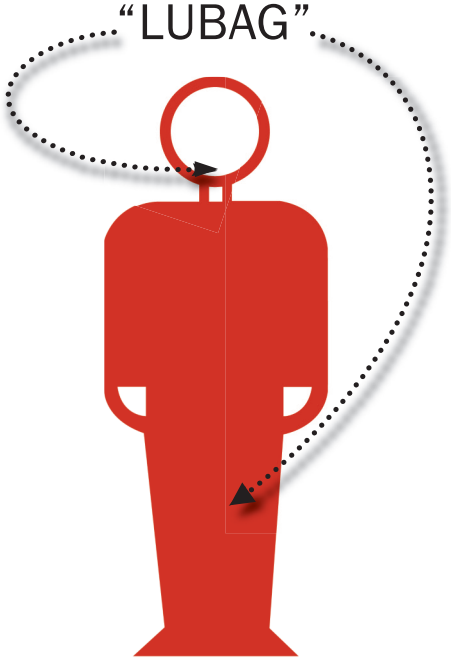
X-Linked Dystonia-Parkinsonism



X-linked dystonia-parkinsonism (XDP) is a genetic form of dystonia that affects men of Filipino descent almost exclusively. The disorder is also known as lubag based on the Ilonggo term referring to muscle twisting and spasms. It is a progressive, neurodegenerative disorder. Although there is no cure at this time, treatments are available and researchers are actively pursuing new and improved therapies.

DYSTONIA MEDICAL RESEARCH FOUNDATION

serving all dystonia-affected persons

Symptoms

- XDP affects primarily Filipino men and, very rarely, women.
- Symptoms may occur as early as adolescence or as late as the mid-70s.
- The earliest symptoms are usually parkinsonian, including resting tremor, bradykinesia (slowness of movement), rigidity, poor balance, and a shuffling gait.
- Symptoms of dystonia develop later in the limbs, tongue, pharynx, and/or larynx, with the most common being jaw dystonia that progresses to cervical dystonia (affecting the neck and shoulders).
- Neuroimaging studies may reveal neurodegenerative changes associated with parkinsonism in an area of the brain called the basal ganglia.

Genetics

- XDP is a genetic disorder inherited in an x-linked manner.
- XDP is associated with mutations in and around the DYT3/TAF1 gene.
- Males with XDP pass the disease-causing gene mutations to all of their daughters but not to their sons.
- Women who are carriers have a 50% chance of transmitting the mutation in each pregnancy.
- Males who inherit the mutation from their mothers will develop XDP; females who inherit the mutation will be carriers and only rarely have symptoms.
- Genetic counseling is recommended for affected individuals and families to provide guidance for genetic testing and identify family members who may be at risk of developing XDP.

Diagnosis

- XDP is diagnosed by observance of dystonia and parkinsonism symptoms, family history with x-linked mode of inheritance, genetic testing, and ancestral roots from the Panay Island in the Philippines.

Treatment

- Treatment for XDP involves using medications to address both dystonia and parkinsonism.
- Anticholinergic medications, benzodiazepines, and sometimes neuroleptics are used in the early stages of dystonia. Zolpidem and tetrabenazine may be used to treat more advanced dystonia symptoms.
- Botulinum neurotoxin injections may improve dystonia in the neck and shoulders, eyelids and brow, tongue dystonia, or jaw dystonia but may worsen swallowing in individuals with dysphagia (difficulty swallowing).

- Parkinsonism symptoms may be treated with levodopa and dopamine agonist medications to control tremor.
- Deep brain stimulation (DBS) surgery may be an important option in individuals with severe, advanced XDP responding poorly to less invasive treatments.

Prevention of Complications

- Symptoms such as difficulty swallowing and dystonia affecting the pharynx make certain individuals with XDP vulnerable to serious complications such as aspiration pneumonia and recurrent infections. Part of treatment for these individuals should include swallowing therapy and diet modification.
- Physical therapy may help preserve mobility and delay disability associated with movement symptoms.

Support

Individuals and families affected by XDP are strongly encouraged to:

- Seek out the best medical care, including a multi-disciplinary movement disorders team and genetic counselor.
- Learn as much as possible about XDP and treatment options.
- Use a multi-layered support system of support groups, online resources, friends, family, and mental health professionals, if needed.
- Investigate complementary therapies.
- Participate in the XDP and dystonia community.

MORE INFORMATION:

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