



MACHADO/JOSEPH DISEASE.. WHAT IS IT ???

ONE THING FOR SURE....IT'S REAL AND IT IS KILLING OUR PEOPLE....

Description:

Machado/Joseph Disease (MJD) is a fatal autosomal dominant genetic disorder of the nervous system that cripples and paralyzes while leaving the intellect intact. The disease is characterized by weakness in the arms and legs and a general loss of motor control that eventually confines the patient to a wheelchair. Symptoms appear when a defective gene causes a breakdown and loss of cells in specific areas of the brain known as the striatum, the cerebellum, and the substantia nigra, but what sets this process in motion is still unknown. MJD, first documented in the 1970's, is named for Antone Joseph, a Portuguese sailor with the defective gene who came to California in 1845. The disease occurs primarily in people of Portuguese ancestry, but it has also been found in other ethnic groups, nationalities, and races.

At least two Brazilian families originated in mainland Portugal, and another one claims to descend from Portuguese Jews in Amsterdam. Another affected family migrated directly from northeastern Portugal to the United States.

Taking into account the family names and traditional professions, physical phenotype, and places of residence of the affected families in mainland Portugal and of the Bastiana (Joseph) family, it is suggested that the original MJD mutation may have arisen among the settlements of **Sefardic Jews** in northeastern Portugal. The **Sefardic Jews**, having arrived on the Iberian peninsula mainly with the Moorish invasions, later came to Portugal seeking refuge from imminent expulsion from Spain in 1492, and they settled mainly along the Spanish-Portuguese border, subject to varying periods of tolerance and repression. When forced conversion or expulsion was decreed also in Portugal in 1496, most of the Jews chose to stay and had to practice their faith in secret or assimilate. Mixed religious practices and cultures can still be found in those areas, *although local Sefardic communities can no longer be well identified*. Interestingly, many of the Portuguese with MJD, both in Portugal and the United States, "still bear family names traditionally attributed to the **Sefardim**".

Symptoms:

Some symptoms of MJD resemble those of other neurological disorders such as Multiple Sclerosis and Parkinson's disease. A careful diagnosis is therefore important and should be made by a physician with expertise in neurology. Symptoms of MJD include:

- Weakness in the arms and legs
- Spasticity, especially in the legs
- Awkward body movements
- Staggering, lurching gait-easily mistaken for drunkenness
- Difficulty with speech and swallowing
- Involuntary eye movements
- double vision
- Bulging appearance of the eyes
- Frequent urination

MJD is diagnosed by identifying the typical symptoms in a family in which the disease occurs. Characteristic features include progressive difficulty in walking and speech beginning in the late teen years or in the 20's through the 50's. The gait is abnormal due to spasticity and speech is slurred because of spastic weakness in the throat muscles. The MJD patient may be unable to look upward or inward, and the eyes may oscillate from side to side.

Late-onset MJD, the type that begins when a patient is 70 or older, is characterized by an uncoordinated gait that may cause the patient to stumble or fall, the slurring of speech and the loss of muscle in the arms and legs. Neurologists have classified MJD into three types, depending on age of onset and characteristic symptoms. But it is uncertain whether the three types are subtypes of the same disease or three separate diseases. Because the three types have at times borne different names, the plural term Joseph diseases has been used. The disease progresses relentlessly and death occurs from 6 to 29 years after onset.

Incidence and Carriers:

MJD is an autosomal dominant disorder. This means that each child of an affected parent has a 50 percent chance of inheriting the defective gene. MJD does not skip generations, but people at risk who escape the disease will not pass it on to their children or future generations. People at risk must decide whether to have children without knowing for sure whether they might pass the gene on. As with any inherited disorder, MJD is not contagious and cannot be "caught" by people who are not at risk.

Treatment:

Currently there is no cure for MJD, but some symptoms of the disease can be treated. Spasticity as well as sleep disturbances can be treated with medications. Physical therapy can help patients with gait disturbances.

Three forms of MJD are recognized:

Types MJD-I, MJD-II, and MJD-III. The differences in the types of MJD relate to the age of onset and severity. Earlier onset usually produces more severe symptoms.

Sources: Mazor Guide, National Institute of Neurological Disorders and Stroke, WebMD.

MANY LATINOS BEING MIS-DIAGNOSED WORLD WIDE:

Throughout the Caribbean, Latin America and even in the USA, many Latinos are being mis-diagnosed with either Parkinson's, Multiple Sclerosis OR Muscular Dystrophy because of the incredible similarities that MJD has with these 3 famous diseases. Then, their Doctors begin treating them for these diseases and after several months of treatments, the Doctors discover to their dismay, that it's not what they thought at all ! The worst part is, that *"they don't know what it is"*. MJD is virtually an *"unknown"* in the medical industry and a cure *"must"* be found. Many medical professionals, Neurologists and geneticists have stated, that if a cure can be found for MJD....it could very well lead to...or be the cure for the other famous 3.

WE NEED YOUR HELP....

The goal of the **Aliyah Sefarad International & the Sefardic DNA Research Center** is to offer Latinos everywhere, the opportunity to give a DNA sample, that can help

- 1) find out if they are of Sefardic Descent and if so,
- 2) utilize that DNA sample to determine if they are a carrier of the genetic marker of **Machado/Joseph Disease**.
- 3) if the marker is found, we will log it and use every means at our disposal, to help find a cure for this debilitating killer.

Currently Aliyah Sefarad International has an exclusive agreement in place with the prestigious National Institute for Biotechnology in the Negev (NIBN) at the famous Ben Gurion University in Israel to gather the funding necessary to find a cure for MJD and according to these experts *within a 2 year period!* **Please help in any way you can. Call us, email text us... give to this noble endeavor**