# Machado-Joseph Disease

Machado-Joseph disease (MJD), also called spinocerebellar ataxia type 3, is a rare, hereditary (passed from parents) ataxia. (Ataxia is a general term meaning lack of muscle control.) The disease is characterized by clumsiness and weakness in the arms and legs, spasticity, a staggering lurching gait easily mistaken for drunkenness, difficulty with speech and swallowing, involuntary eye movements, double vision, and frequent urination. Some patients have dystonia (sustained muscle contractions that cause twisting of the body and limbs, repetitive movements, abnormal postures, and/or rigidity) or symptoms (problems) similar to those of Parkinson's disease. Others have twitching of the face or tongue, or peculiar bulging eyes. In Machado-Joseph disease and other spinocerebellar ataxias, degeneration of cells in an area of the brain called the hindbrain leads to deficits in movement. The hindbrain includes the cerebellum, the brainstem, and the upper part of the spinal cord. MJD is an inherited, autosomal dominant disease, meaning that if a child inherits one copy of the defective gene from either parent, the child will develop symptoms of the disease. People with a defective gene have a 50 percent chance of passing the mutation on to their children.

## TREATMENT

MJD is incurable, but some symptoms of the disease can be treated. For those patients who show parkinsonian features, levodopa therapy can help for many years. Treatment with antispasmodic drugs, such as baclofen, can help reduce spasticity. Physiotherapy can help patients cope with disability associated with gait problems, and physical aids, such as walkers and wheelchairs, can assist the patient with everyday activities. Other problems, such as sleep disturbances, cramps, and urinary dysfunction, can be treated with medications and medical care.

## WHAT WILL HAPPEN

The severity of the disease is related to the age of onset, with earlier onset associated with a more severe form of the disease. Symptoms can begin any time between early adolescence and about 70 years of age. MJD is also a progressive disease, meaning that symptoms get worse with time. Life expectancy ranges from the mid-thirties for those with severe forms of MJD to a normal life expectancy for those with mild forms. For those who die early from the disease, the cause of death is often aspiration pneumonia.

## RESEARCH BEING DONE

**Research is continually being done to learn more about this problem.**

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