

Machado-Joseph Disease (MJD-III), also called spinocerebellar ataxia type III, is a rare, inherited, ataxia (lack of muscular control) affecting the central nervous system and characterized by the slow degeneration of particular areas of the brain called the hindbrain. Patients with MJD may eventually become crippled and/or paralyzed but their intellect remains intact. The onset of symptoms of MJD varies from early teens to late adulthood. MJD is a rare inherited neurological disorder that disproportionately affects individuals of Portuguese descent, especially those from the Azores, an island colonized by Portuguese people. MJD appears to affect slightly more males than females. While a family history and physical examination help in the diagnosis, the gold standard of diagnostic tests that detects 100% of the cases is the direct determination of the number of suspect CAG triplets in a patient's DNA. This may be readily done at a specialized genetic clinical laboratory.