

The term olivopontocerebellar atrophy (OPCA) has historically been used to describe a group of disorders that affect the central nervous system and are termed neurodegenerative diseases because they result in a progressive deterioration of nerve cells in certain parts of the brain. These conditions are characterized by progressive balance problems (disequilibrium), progressive impairment of the ability to coordinate voluntary movements (cerebellar ataxia), and difficulty speaking or slurred speech (dysarthria). OPCA is a group of rare disorders that affects males and females in equal numbers. Because of confusion regarding the naming and classification of these disorders, determining their frequency in the general population is difficult. The frequency of all forms of OPCA has been estimated to be 3-5/100,000 in the United States. A diagnosis of OPCA is a preliminary diagnosis that is made by a thorough clinical examination and identification of characteristic symptoms. Hereditary OPCA can be diagnosed based on a family history of the same condition or by molecular genetic testing for gene mutations known to be associated with the condition. Molecular genetic testing is available for several of the SCAs. A diagnosis of sporadic OPCA is made if hereditary OPCAs and other conditions associated with symptoms of OPCA are ruled out. Testing may include blood work to rule out other conditions, MRI scans of the brain to look for degenerative changes in the brainstem, EMG testing to look at the electrical testing of muscles and nerves and sometimes examination of spinal fluid.