Olivopontocerebellar Atrophy

What is Olivopontocerebellar Atrophy?
Olivopontocerebellar atrophy (OPCA) is a term that describes the degeneration of neurons in specific areas of the brain such as the cerebellum, pons, and inferior olives. OPCA is present in several neurodegenerative syndromes, including inherited and non-inherited forms of ataxia (such as the hereditary spinocerebellar ataxia known as Machado-Joseph disease) and multiple system atrophy (MSA), with which it is primarily associated. OPCA may also be found in the brains of individuals with prion disorders and inherited metabolic diseases. The characteristic areas of brain damage that indicate OPCA can be seen by imaging the brain using CT scans or MRI studies.

Alternative Names
OPCA; Olivopontocerebellar degeneration; Multiple system atrophy C cerebellar predominance; MSA-C

Causes, incidence, and risk factors:
Olivopontocerebellar atrophy can be passed down through families (inherited form), or it may affect people without a known family history (sporadic form). Researchers have identified certain genes that are involved in the inherited form of this condition. The cause of olivopontocerebellar atrophy in those without a history of the disease is not known. The disease slowly gets worse (is progressive). This disease is slightly more common in men than in women. The average age of onset is 54 years old.

Classification
There are at least five forms of OPCA. All are inherited in an autosomal manner. Both autosomal dominant and autosomal recessive types are known. There is considerable complexity and some confusion about the names for the different forms of OPCA, which is also known as spinocerebellar ataxia or SPA:
OPCA I (or SPA 1) -- Autosomal dominant. Onset of symptoms usually in the third or fourth decade of life, most often around age 30. Due to expansion of a CAG trinucleotide sequence in the ataxin-1 gene (ATX1) on chromosome 6p23.
OPCA II -- Autosomal recessive. Called the Fickler-Winkler type of OPCA. Differs from OPCA I in a lack of involuntary movements and of sensory changes. Gene not known.
OPCA II (or SCA 2) -- Autosomal dominant. Called the Cuban type of OPCA. Due to expansion of a CAG trinucleotide sequence in the ataxin-2 gene (ATX2) on chromosome 12q24.
OPCA III (or SCA 7) -- Autosomal dominant. OPCA with macular degeneration and ophthalmoplegia. Due to expansion of a trinucleotide sequence in the ataxin-7 gene (SCA7) on chromosome 3p.
OPCA IV -- Autosomal dominant. Similar clinically to OPCA I but with spastic paraplegia. May be due to a different mutation in the ataxin-1 gene (ATX1) and so be allelic to OPCA I.
OPCA V -- Autosomal dominant. OPCA with progressive dementia and extrapyramidal neurologic signs. The gene responsible for this disease has not been identified.