Clinical features and molecular genetics of autosomal recessive cerebellar ataxias.

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Source

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Abstract

Among the hereditary ataxias, autosomal recessive spinocerebellar ataxias comprise a diverse group of neurodegenerative disorders. Clinical phenotypes vary from predominantly cerebellar syndromes to sensorimotor neuropathy, ophthalmological disturbances, involuntary movements, seizures, cognitive dysfunction, skeletal anomalies, and cutaneous disorders, among others. Molecular pathogenesis also ranges from disorders of mitochondrial or cellular metabolism to impairments of DNA repair or RNA processing functions. Diagnosis can be improved by a systematic approach to the categorisation of these disorders, which is used to direct further, more specific, biochemical and genetic investigations. In this Review, we discuss the clinical characteristics and molecular genetics of the more common autosomal recessive ataxias and provide a framework for assessment and differential diagnosis of patients with these disorders.

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