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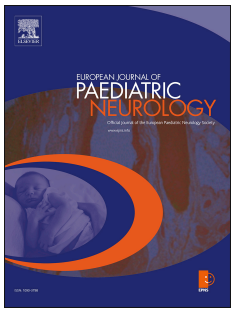

ATP1A3-related disorders: An update

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Abstract

Alternating Hemiplegia of Childhood (AHC), Rapid-onset Dystonia Parkinsonism (RDP) and CAPOS syndrome (cerebellar ataxia, areflexia, pes cavus, optic atrophy, and sensorineural hearing loss) are three distinct, yet partially overlapping clinical syndromes that have long been thought to be allelic disorders. From 2004 to 2012, both autosomal dominant and *de novo* mutations in *ATP1A3* have been detected in patients affected by these three conditions. Growing evidence suggests that AHC, RDP and CAPOS syndrome are part of a large and continuously expanding clinical spectrum and share some recurrent clinical features, such as abrupt-onset, asymmetric anatomical distribution and the presence of triggering factors, which are highly suggestive of *ATP1A3* mutations. In this review, we will highlight the main clinical and genetic features of *ATP1A3*-related disorders focusing on shared and distinct features that can be helpful in clinical practice to individuate mutation carriers.