MEF2C haploinsufficiency syndrome

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Introduction:

MEF2C haploinsufficiency syndrome is characterized by severe intellectual disability, epilepsy, stereotypic movements, minor dysmorphisms and brain abnormalities. We report the case of a patient with a new MEF2C mutation, comparing his clinical and imaging features to those previously reported in the literature.

Case report:

A 10 year-old boy first came to pediatric neurology clinic at the age of 11 months because of severe psychomotor delay, without regression. He presented generalized hypotonia, poor eye contact, hand-mouth stereotypies, strabismus and minor facial dimorphisms. Epileptic seizures started at 26 months of age and were refractory. Brain MRI showed a slight increase in periventricular white matter signal and globally enlarged CSF spaces. Molecular analysis revealed a de novo, pathogenic and causative MEF2C mutation.

Discussion:

MEF2C haploinsufficiency syndrome was recently recognized as a neurodevelopmental disorder. Severe intellectual disability with inability to speak and epilepsy are universal features in patients with MEF2C mutations, although mild cognitive and speech disorders have been reported to occur in patients with duplications. Epilepsy might be absent in patients with partial deletions. Abnormal movement patterns are very common in patients with MEF2C haploinsufficiency. Delayed myelination seems to be more commonly observed in patients with MEF2C mutations, while malformations of cortical development were only reported in patients with microdeletions. Although MEF2C haploinsufficiency prevalence is yet to be determined, it should be considered in the differential diagnosis of patients with severe intellectual disability and Rett-like features.