MEF2C haploinsufficiency syndrome

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Mutations in the

MEF2C

(

myocyte enhancer factor 2

) gene have been established as a cause for an intellectual disability syndrome presenting with seizures, absence of speech, stereotypic movements, hypotonia, and limited ambulation. Phenotypic overlap with Rett's and Angelman's syndromes has been noted. Following the first reports of 5q14.3q15 microdeletions encompassing the

MEF2C

gene, further cases with point mutations and partial gene deletions of the

MEF2C

gene have been described. We present the clinical phenotype of our cohort of six patients with MEF2C mutations and compare our findings with previously reported patients as well as with a growing number of genetic conditions presenting with a severe neurodevelopmental, Rett-like, phenotype. We aim to add to the current knowledge of the natural history of the "MEF2C haploinsufficiency syndrome" as well as of the differential diagnosis, clinical management, and genetic counseling in this diagnostically challenging group of patients.