Muscular, Ocular and Brain Involvement Associated with a De Novo 11q13.2q14.1 Duplication: Contribution to the Differential Diagnosis of Muscle-Eye-Brain Congenital Muscular Dystrophy.

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Abstract

Muscular weakness and hypotonia may be associated with multisystem involvement giving rise to complex phenotypes, many of which are uncharacterized. We report a patient presenting with congenital hypotonia and severe ocular and brain abnormalities, evoking a Muscle Eye Brain disease (MEB). She had global muscular weakness, hypotonia and amyotrophy, joint hyperlaxity, kyphoscoliosis, respiratory insufficiency, dysmorphic features and severe intellectual disability. Brain MRI showed cortical atrophy and hypoplasia of the corpus callosum. Normal CK levels, non-progressive course and absence of dystrophic features or α-dystroglycan abnormalities on the muscle biopsy were not typical of MEB. CGH array identified a large de novo duplication in chromosome 11, including regions partially duplicated in three other patients with common clinical
features. This report adds to the differential diagnosis of complex phenotypes characterized by muscular, ocular and CNS involvement and highlights the potential contribution of still unrecognized chromosomal abnormalities to these phenotypes.

**KEYWORDS:** CNV; Myopathy; congenital muscular dystrophy; dystroglycanopathy; muscle-eye-brain; ocular involvement

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