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Format: Abstract

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Clinical characteristics of megaconial congenital muscular dystrophy due to choline kinase beta gene defects in a series of 15 patients.

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Abstract

A new form of congenital muscular dystrophy (CMD) with multisystem involvement and characteristic mitochondrial structural changes, due to choline kinase beta (CHKB) gene defects has been characterized by intellectual disability, autistic features, ichthyosis-like skin changes, and dilated cardiomyopathy. We define the clinical characteristics in 15 patients, from 14 unrelated families with so-called 'megaconial CMD', all having mutations in CHKB. Core clinical phenotype included global developmental delay prominent in gross-motor and language domains, severe intellectual disability (ID), and/or muscle weakness in all cases. Muscle biopsies were equivocally 'megaconial' in all. Other peculiarities were: ichthyosis-like skin changes (n = 11), increased serum CK levels (n = 12), microcephaly (n = 6), dysmorphic facial features (n = 7), neonatal hypotonia (n = 3), seizures (n = 3), epileptiform activity without clinically overt seizures (n = 2), dilated cardiomyopathy (n = 2), decreased left ventricular systolic function (n = 2), congenital heart defects (n = 3), sensorineural (n = 1), and conductive hearing loss (n = 1). Ten patients had cranial neuroimaging (MRI-MRS) study, which was notably normal in all, other than one patient having a decreased choline: creatine peak. Intra-familial variability in clinical expression of the disease is

noted in four families. Two siblings from the same family, one presenting with global developmental delay and dilated cardiomyopathy, and the other with ichthyosis, ID and proximal weakness without cardiomyopathy died at the ages of 2 years 1 month, and 7 years 4 months respectively. Evolution was progressive (n = 13) and static (n = 2).

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