Muscular Dystrophy, Congenital with Severe Central Nervous System Atrophy and Absence of Large Myelinated Fibers

**WHO International Classification of Diseases**
Diseases of the nervous system

**OMIM Number**
601170

**Mode of Inheritance**
Autosomal recessive

**Description**
Congenital muscular dystrophy is a heterogeneous group of disorders characterized by muscular hypotonia of prenatal onset and the histologic features of muscular dystrophy. It can also be associated with involvement of the central nervous system.

**Epidemiology in the Arab World**

**United Arab Emirates**
Sztriha et al. (1999) described two female infants born to consanguineous parents in an inbred Arab family of United Arab Emirates origin with a syndrome of micrencephaly with simplified gyral pattern, abnormal myelin formation, and arthrogryposis. In the first case, the female infant was born at 35 weeks gestation by Cesarean section as a result of fetal distress and breech presentation. The patient as a neonate did not have spontaneous respiration after birth and needed mechanical ventilation for two months. The first MR imaging of the brain and spinal cord was performed when the baby was seven days old. The size of the vermis and cerebellar hemispheres was moderately reduced and the fourth ventricle and cisterna magna were enlarged. The second infant had multiple joint deformities and a small brain with a wide space between the cerebral surface and the bones. In both cases, increased variation of fiber size was seen in the muscle biopsy; creatine kinase, however, was normal. Large areas of muscle were replaced by adipofibrous tissue. Both infants had dysmorphic features consistent with the fetal akinesia/hypokinesia sequence.

**References**

**Contributors**
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