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Megaconial Congenital Muscular Dystrophy: A Case Report

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Abstract **Case Report**

Megaconial congenital muscular dystrophy (MCM) is a rare autosomal recessive mitochondrial myopathy caused by mutations in the CHKB gene, affecting lipid metabolism and mitochondrial function. It is characterized by enlarged mitochondria at the periphery of muscle fibers ("megaconia"). Clinical features include early-onset hypotonia, progressive muscle weakness, global developmental delay with intellectual disability, and cardiomyopathy. Some patients may present congenital heart defects, autistic traits, or ichthyosiform skin lesions. Diagnosis relies on elevated serum CK, electromyography showing a myogenic pattern, muscle biopsy revealing megaconia, and genetic confirmation of CHKB mutations. Management is multidisciplinary, involving physiotherapy, orthotic support, respiratory care, and symptomatic treatment. We report a 10-year-old child with MCM. Due to its multisystem involvement and broad clinical spectrum, differential diagnosis includes other neurometabolic and mitochondrial disorders. Prognosis depends on symptom severity and progression, particularly regarding cardiac and respiratory function.

Keywords: Muscular Dystrophy, Megaconial, Ck, Myogenic Involvement.

INTRODUCTION

Megaconial congenital muscular dystrophy is a rare genetic disorder of skeletal muscle, characterized by early-onset hypotonia, muscle weakness, global developmental delay with intellectual disability, and cardiomyopathy. Congenital structural cardiac anomalies and ichthyosiform skin lesions have also been reported. Muscle biopsy reveals a characteristic enlargement of mitochondria located at the periphery of muscle fibers.

This report concerns a 10-year-old patient treated at a pediatric hospital, who was diagnosed with megaconial congenital muscular dystrophy.

CASE REPORT

A 10-year-old male child, born to first-degree consanguineous parents, with a family history of an undiagnosed psychomotor delay in his sister who died at age 11, presented with neonatal hypotonia, psychomotor delay, sleep disturbances, and hyperactivity. Clinical examination revealed dry skin, oral aphthae, flaccid muscular hypotonia, absent deep tendon reflexes, and mild muscle atrophy. Cardiopulmonary examination was unremarkable.

Laboratory tests showed elevated creatine kinase (CK), AST, pyruvate, and lactate levels. Electromyography (EMG) revealed a myogenic pattern, while brain MRI was normal. Genetic testing via whole exome sequencing confirmed the diagnosis by identifying a homozygous pathogenic variant in the CHKB gene. Cardiac evaluation was unremarkable. The child was started on coenzyme Q10 and L-carnitine supplementation.

DISCUSSION

Megaconial congenital muscular dystrophy is a rare myopathy, generally inherited in an autosomal

recessive manner, characterized by early-onset muscle weakness, hypotonia in early childhood, and proximal muscle involvement. It is caused by mutations in the CHKB (choline kinase beta) gene, leading to defective biosynthesis of membrane phospholipids, particularly phosphatidylcholine.

Histologically, muscle biopsy shows fibers containing giant mitochondria, mainly located at the periphery, hence the term *"megaconial"*. Clinically, affected children present with motor delay, unstable gait, symmetric proximal muscle weakness, and often elevated muscle enzymes (CK). Cardiac involvement (dilated cardiomyopathy) and cognitive impairment may also occur in some cases, worsening prognosis.

Diagnosis relies on a combination of clinical, biochemical, histological, and molecular data. To date, no curative treatment exists, and management remains primarily symptomatic and multidisciplinary, focusing on physiotherapy, cardiac and respiratory follow-up, and nutritional support. Genetic testing is crucial for confirming the diagnosis, guiding genetic counseling, and potentially enabling future targeted therapeutic approaches.

CONCLUSION

This case illustrates the characteristic clinical and histological features of megaconial congenital muscular dystrophy, a rare but recognizable entity when early-onset muscle weakness is associated with giant mitochondria on biopsy. It emphasizes the importance of considering this disorder in the differential diagnosis of congenital myopathies to rapidly confirm the etiology through genetic analysis. Although no specific treatment is currently available, early diagnosis allows for appropriate multidisciplinary care and genetic counseling for the family.

REFERENCES

- Magri F, Antognozzi S, Ripolone M, *et al., Megaconial congenital muscular dystrophy due to novel CHKB variants: a case report and literature review. Skeletal Muscle*. 2022 ;12 :23. (Rapport de cas décrivant un enfant de 6 ans avec des mutations nouvelles du gène CHKB, et revue de la littérature sur la pathologie.) SpringerLink
- Zemorshidi F, Nafissi S, Boostani R, *et al., Megaconial congenital muscular dystrophy due to CHKB gene variants: the first report of thirteen Iranian patients. Neuromuscul Disord*. 2023 ;33(7):589–595. (La première série iranienne de 13 patients, décrivant les aspects cliniques, biopsiques et les variantes génétiques identifiées.) PubMedinstitut-myologie.org
- Bardhan M, Polavarapu K, Bevinahalli NN, *et al., Megaconial congenital muscular dystrophy secondary to novel CHKB mutations resemble atypical Rett syndrome. J Hum Genet*. 2021 ;66(8):813–823. PubMed
- Haliloglu G, Talim B, Sel CG, Topaloglu H. *Clinical characteristics of megaconial congenital muscular dystrophy due to choline kinase beta gene defects in a series of 15 patients. J Inherit Metab Dis*. 2015 ;38(6):1053–1061institut-myologie.org+1
- Castro-Gago M, Dacruz-Alvarez D, Pintos-Martínez E, *et al., Congenital neurogenic muscular atrophy in megaconial myopathy due to a mutation in CHKB gene. Eur J Paediatr Neurol*. 2014 ;18(6):796–800.