Congenital Myotonic Dystrophy: A Retrospective Study of a Single Center

Duygu Yılmaz[1], Gökçe Eser[1], Haluk Topaloğlu[2]

1. Yeditepe University Hospital, Department of Pediatric Heath and Diseases
2. Yeditepe University Hospital, Division of Pediatric Neurology

ICNC 2022
17th INTERNATIONAL CHILD NEUROLOGY CONGRESS
ANTALYA, TURKEY | OCTOBER 3-7, 2022

INTRODUCTION

Congenital myotonic dystrophy (CMD) is an autosomal dominant genetic disorder caused by trinucleotide repeat expansion of CTG (cytosine-thymine-guanine) in the 3' untranslated region (UTR) of the DMPK (dystrophia myotonica protein kinase) gene.

The incidence of CMD is 1 in 47619 live births. It is a multisystemic disease characterized by hypotonia, feeding difficulties, and respiratory distress. In addition, it can cause cataracts, cardiac conduction abnormalities, insulin resistance, and developmental disabilities. Disease severity increases, and age of onset decreases in successive generations, known as anticipation.Given the anticipation, diagnosing the disease and providing genetic counseling to families is essential.

OBJECTIVES

CMD, which has similar clinical features with other neuromuscular diseases, should be considered in the differential diagnosis.Therefore, we reviewed the CMD patients diagnosed in our clinic and compiled the criteria that helped us to diagnose.

MATERIALS & METHODS

Seventeen patients diagnosed with CDM by molecular genetic testing in our clinic were retrospectively analyzed in prenatal history, family history, mental development, laboratory findings, neurological, gastrointestinal, and respiratory system findings.

RESULTS

Seventeen patients were diagnosed with CMD in our clinic, eight were male, and nine were female. The age at diagnosis was between 1 month and 16 years. The most common complaint of patients younger than 1 year of age was hypotonia and respiratory distress. Children over the age of 10 were unable to open their hands. In addition, there was consanguinity between the parents of 4 patients. 11 patients had a family history of similar disease. Seven patients were followed up in the neonatal intensive care unit, 5 patients were hypotonic, and 5 patients had respiratory distress in the neonatal period. The mental development of 5 patients was normal. 9 patients had mild, 2 had moderate, and 1 had severe mental retardation. The walking age ranged from 14 months to 4 years. Muscle hypertrophy was not observed in any patients. 12 patients had proximal muscle weakness. Seven patients were hypotonic on examination. Kyphoscoliosis was not observed in any of the patients. Deep tendon reflexes were decreased in 2 patients, and they were normal in other patients. 4 patients had contractures in their joints. A high-arched palate was observed in 14 patients. 1 patient had dysphagia. Three patients had complaints of constipation. Creatine kinase values ranged from 88-269 u/l. 12 patients had a partial response, 2 had a complete response and 3 had no response to physical therapy.

CONCLUSIONS

CMD should be considered in the differential diagnosis of newborns with a history of hospitalization in the neonatal intensive care unit, hypotonia, respiratory distress, high-arched palate, proximal muscle weakness, and mental development delay. In addition, newborns with a family history should be evaluated for CMD, and genetic counseling should be provided to families.

REFERENCES

1. Lanni S, Pearson CE. Molecular genetics of congenital myotonic dystrophy. Neurobiol Dis. 2019 Dec;132:104533. doi: 10.1016/j.nbd.2019.104533. Epub 2019 Jul 19. PMID: 31326502.
2. Hayes HA, Dibella D, Crockett R, Dixon M, Butterfield RJ, Johnson NE. Stepping Activity in Children With Congenital Myotonic Dystrophy. Pediatr Phys Ther. 2018 Oct;30(4):335-339. doi: 10.1097/PEP.0000000000000537. PMID: 30277969.
3. Omura J, Chen M, Haviland M, Apkon S. Respiratory outcomes in children with congenital myotonic dystrophy. J Pediatr Rehabil Med. 2022;15(2):275-279. doi: 10.3233/PRM-200726. PMID: 34806628.

CONTACT

E mail: md.duyguyilmaz@gmail.com
Phone: +905343703073