A Multicenter Retrospective Study in Turkish Children with Myotonic Dystrophy Type 1

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OBJECTIVES

Myotonic dystrophy type 1 (DM1) is a multisystem disorder with a broad spectrum of severity caused by autosomal dominant inherited unstable expanded CTG repeat in the *DMPK* gene. Disease onset is highly variable, ranging from newborn to late adult. The phenotype has been well described in adults, data in children remain scant. This study aimed to investigate the genetic and clinical features of pediatric-onset cases, which are less common.

METHODS

Demographic, genetic and clinical data of the patients younger than 18 years with a genetically confirmed diagnosis, from nine centers in different geographical regions (Figure I) of Türkiye were retrospectively investigated.

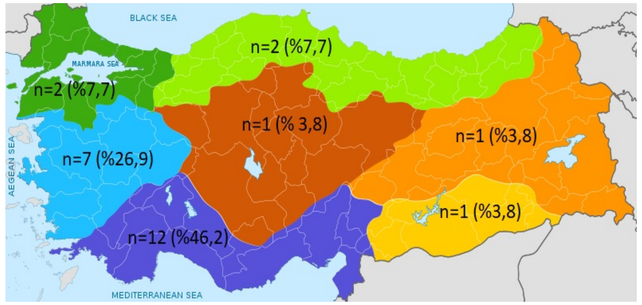

Figure I. Distribution of cases by geographical regions of Türkiye

RESULTS

Twenty-six patients (mean age 11.9 years, 50% females, with 30.8% congenital, 53.8% pediatric, 15.4% juvenile form) were included (Figure II). While the mean age of symptom onset was 5,6 (min=0,1, max=15), the mean age of diagnosis was 7,8 years (min=0,5, max=16) .

Antenatal history of polyhydramnios (7.7%), reduced fetal movements (26.9 %), low birth weight (15.3%), and prematurity (23.1%) were evident.

Six patients needed mechanical ventilator care in the neonatal period.

22 patients had a family history of DM1 (13 maternal/ 9 paternal). The initial clinical manifestations were muscle weakness (53.9%), hypotonia (38.4%), myotonia(26.9%), developmental delay (23%), and abnormal gait (7.6%). Various findings showing multisystem involvement (3/26 cardiac dysrhythmia, 2/26 type 1 diabetes mellitus, 1/26 polycystic ovary syndrome, 12/26 psychiatric comorbidities (6/12 attention deficiency and hyperactivity), 6/25 gastrointestinal system findings, 3/25 abnormality in cranial magnetic resonance imaging) were detected.

15 patients were receiving carbamazepine and two mexiletine. 13 of them had partial treatment response.

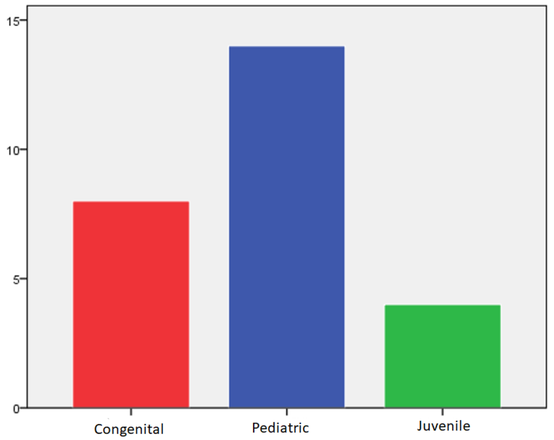

Figure II. Distribution of cases by disease onset

CONCLUSION

The genetic and clinical findings of pediatric DM1 patients are diverse, which requires a multi-faceted approach and management.

REFERENCES

1. Ho G, Carey KA, Cardamone M, Farrar MA. Myotonic dystrophy type 1: clinical manifestations in children and adolescents. Arch Dis Child. 2019 Jan;104(1):48-52.
2. Lagrue E, Dogan C, De Antonio M, Audic F, Bach N, Barnerias C,et al. A large multicenter study of pediatric myotonic dystrophy type 1 for evidence-based management. Neurology. 2019 Feb 19;92(8):e852-e865.

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