



Oro-facial manifestations in myotonic syndrome

Manifestações bucais na síndrome miotônica

Mubeen Khan^[a], Rai Puja^[b]

^[a] Professor and head, Department of Oral Medicine and Radiology, Government Dental College and Research Institute, Bangalore - India.

^[b] Postgraduate student, Department of Oral Medicine and Radiology Government Dental College and Research Institute, Bangalore - India, e-mail: pujas.rai@gmail.com

Abstract

Objective: To present a case of myotonic syndrome in a 35-year-old male patient and to review and discuss the literature. **Discussion and conclusion:** The myotonic syndrome is a steadily progressive, familial, distal myopathy with associated weakness of the muscles of face, jaw, neck and elevators of the eyelids, a tendency for myotonic persistence of contraction in the affected parts and testicular atrophy. The patient was referred to the Department with complaint of recurrent temporomandibular joint dislocation, presenting with the characteristic oro-facial manifestations of myotonic syndrome.

Keywords: Myotonic dystrophy. Steinert disease. Myopathic facies.

Resumo

Objetivo: Apresentar um caso de síndrome miotônica em um paciente adulto, 35 anos, sexo masculino, bem como rever e discutir a literatura pertinente. **Discussão e conclusão:** A síndrome miotônica é uma miopatia distal de progressão contínua, familiar, e associada à fraqueza dos músculos da face, mandíbula, pescoço e músculos elevadores das pálpebras, com tendência de persistência de contratura miotônica nas partes afetadas, bem como atrofia testicular. O paciente se apresentou ao departamento com queixa de deslocamento recorrente da articulação temporomandibular, apresentando as características de manifestações orofaciais de síndrome miotônica.

Palavras-chave: Distrofia miotônica. Doença de Steinert. Fácies miopática.

Introduction

Myotonic dystrophy, or *dystrophia myotonica* (DM), or Steinert disease is a hereditary neuromuscular multisystem, autosomal dominant disease with varying clinical expression and severity. The genetic locus for DM is localized on the long arm of chromosome 19 (1). The mutation is caused by an increased number of CTG trinucleotide repeats in the 3' untranslated region of the DM protein kinase gene (or myotoninkinase) (2, 3). The number of CTG trinucleotides in normal chromosomes ranges from 3 to 30 in normal individuals, from 50 to 80 in mildly affected persons, and 2000 or more copies in severely affected individuals (2). The size of the fragment is related both to age at onset of the disease and to the severity of the phenotype and increases through successive generations in parallel with increasing severity of the disease (anticipation phenomenon) (3). It is the most common form of adult muscular dystrophy. DM usually begins to manifest during the second and third decades of life.

Muscular involvement is characterized by myotonia, progressive muscular weakness and atrophy, mainly of the facial, neck and the distal muscles of the limbs (4, 5). Temporomandibular dislocations, recurrent locking of the jaw and frequent clicks are common complaints (4). Myotonia is defined as an inability of the skeletal muscles to relax after voluntary muscle contraction and usually occurs after the third year of life, being the first sign of the disease.

Report or case

A 35-year-old male patient reported to the Department with a complaint of recurrent temporomandibular joint dislocation from 3 years. Medical history revealed weakness of both upper and lower limbs from past few years, progressively worsening to present state. There was increased fatigability and the patient was unable to carry out his normal day-to-day activities. Patient presented hypersomnia; excessive yawning lead to recurrent temporomandibular joint dislocation. Weak, monotonous, nasal type of voice and complaint of subsequent dysphagia were present. On general examination, patient had an abnormal gait, short stature and poor built. Narrowing

of lower third of face with ptosis and generalized weakness of facial musculature, gave the patient a characteristic “myopathic facies” and “swan neck” appearance (Figure 1).

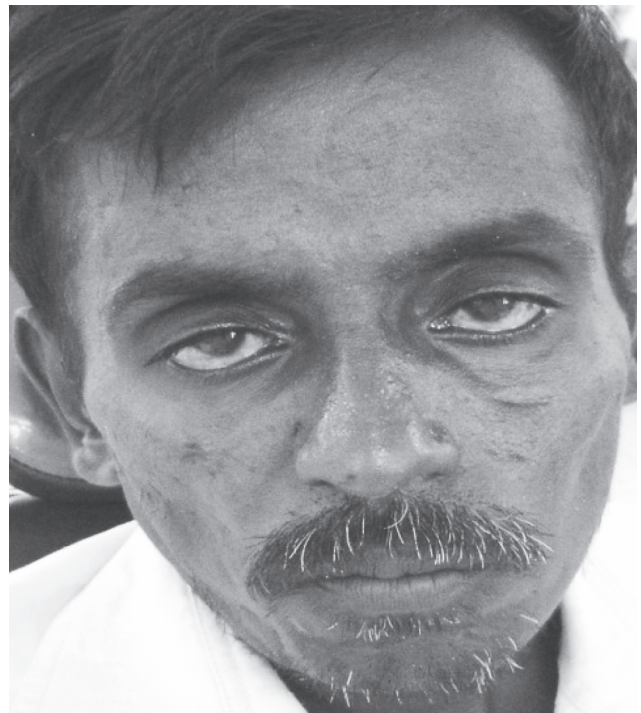


Figure 1 - “Myopathic face”; eyelid ptosis; narrowed lower third of face

Moderate pallor was present. Vital signs were within normal range and patient was well oriented to time, place, and person. Cardiovascular examination revealed S_1S_2 split sounds with occasional missed beats. Central nervous system examination revealed normal higher mental functions and no other cranial nerves obvious abnormalities noted. Motor function test revealed slow relaxation of extensors in hand grip (Table 1). Weakness of neck and trunk muscles present. No sensory involvement noted. Reflexes of the patient were found to be affected.

Patient was subjected for complete blood examination, liver function and thyroid function test, which came out to be within normal range, except for a significant increase in the level of creatinine phosphokinase 528.62IU/L (normal value-10-50IU/L). ECG revealed poor progression of ‘R’ wave. ECHO cardiogram revealed normal chambers, valves, and

flows. Concentric needle EMG was performed which showed myotonic discharges. Clinical findings, medical history, elevated creatinine phosphokinase level and concentric needle EMG were suggestive of myotonic syndrome.

Table 1 - Results of the motor function test

S. No.		Power	
		Right	Left
1.	Shoulder	5/5	5/5
2.	Elbow	5/5	5/5
3.	Hand grip	3/5	3/5
4.	Hip	5/5	4/5
5.	Knee	4/5	4/5

Discussion

Four types of DM can be discerned, with varying symptoms, as follows:

- *mild*, with age of onset > 50 yr and very slight problems;
- *adult-onset* or *classical*, which is the commonest type, with symptoms presenting between age 10 and 50 yr;
- *childhood-onset*, with symptoms starting between age 1 and 10 yr; and
- *congenital type*, with symptoms at birth.

Myotonia and progressive muscular weakness are the hallmarks of the disease. In childhood and congenital types of DM, learning and speech difficulties, and neuropsychiatric disorders, are common (6). Patients also present with a characteristic feature of facial diplegia with a tented upper lip and sagging jaw (1).

Reduced bite force and slower chewing cycle have been shown in adult DM patients and temporomandibular dislocation, recurrent locking of the jaw and frequent clicks are common complaints (7, 8). The prevalence of DM worldwide is 5 to 19 per 100,000, but there are regions with a much higher prevalence (e.g. the Saguenay province in Quebec,

Canada, and the northern parts of Sweden) (4). In a recent study from western Sweden, the prevalence of DM in childhood was reported to be 5 per 100,000 (9).

Facial examination of a patient with DM may show premature frontal scalp balding and facial narrowing (mask-like faces). Muscle wasting of the masticators (ie, temporalis, masseter, pterygoids), sternocleidomastoids, muscles of the tongue, and pharynx is frequently present (10, 11). This may manifest as a triangular mouth opening, protruding mandible, lingual myotonia, velopharyngeal incompetence and dysphagia.

Dysphagia and delayed gastric emptying predispose this group of patients for high risk of aspiration (12, 13). Dentofacial findings include increased interocclusal space, increased facial height, high palatal arch, transverse maxillary deficiency, dental crowding, and anterior open bite (10, 12, 14). Facial weakness, one of the earliest and most constant features of DM, leading to the typical expressionless face, wasting of the temporalis muscles (a prominent feature of myotonic dystrophy), temporo-mandibular dislocation (well documented in DM), recurrent locking of the jaw, difficulty in chewing and frequent clicks along with weakness of the palate contributing to nasal, indistinct speech, were important diagnostic orofacial manifestations in our patient. Systemic manifestations include weakness of the limbs, characterized by distal to proximal paresis, a very common finding in DM patients. The patient's inability to release their hand after a handshake is a classic sign (10). Ocular manifestation includes cataracts, macular degeneration and ptosis of the eyelids. Mild to moderate degree of mental retardation is often present (15). Cardiac manifestations occur in the form of dilated myopathy, conduction defects and mitral valve prolapse (16). Approximately 73% of the patients exhibit abnormal electric cardiogram (ECG) findings (17).

Weakness of the diaphragmatic and intercostal muscle generally leads to decreased lung volumes (vital capacity) and reduction in the maximum expiratory pressure (18). Hypersomnolence is noted frequently in these patients. It is likely a direct result of alveolar hypoventilation and may be worsened by administration of respiratory depression agents such as benzodiazepines and narcotic (19). Patients with DM also may be afflicted with endocrine abnormalities such as diabetes mellitus, diseases of the thyroid, testicular and ovarian dysfunction.

Informed consent

The patient signed an informed consent, agreeing with presentation of the full face image in this article. The document is in the archives of the Institution.

Conflict of interest

The authors declared no conflict of interest in the present article.

References

1. Shaw DJ, Harper PS. Myotonic dystrophy: developments in molecular genetics. *Br Med Bull.* 1989;45(3):745-59.
2. Brook JD, McCurrach ME, Harley HG, Buckler AJ, Church D, Aburatani H, et al. Molecular basis of myotonic dystrophy: expansion of a trinucleotide (CTG) repeat at the 3-prime end of a transcript encoding a protein kinase family member. *Cell.* 1992;68(4):799-808.
3. Harley HG, Rundle SA, Mac Millan JC, Myring J, Brook JD, Crow S, et al. Size of the unstable CTG repeat sequence in relation to phenotype and parental transmission in myotonic dystrophy. *Am J Hum Genet.* 1993;52(6):1164-74.
4. Harper PS, editor. *Myotonic dystrophy.* 2nd ed. London: WB Saunders Co; 1989.
5. Harper PS, Rudel R. Myotonic dystrophy. In: Engel AG, Franzini-Armstrong C, editors. *Myology: basic and clinical.* 2nd ed. New York: Mc Graw-Hill; 1994. p. 1192-219.
6. De Die Smulders C. Long-term clinical and genetic studies in myotonic dystrophy. Thesis, Maastricht, the Netherlands: University of Maastricht; 2000.
7. Kiliaridis S, Meijersjo C, Thilander B. Muscle function and craniofacial morphology: a clinical study in patients with myotonic dystrophy. *Eur J Orthod.* 1989;11(2):131-8.
8. Odman C, Kiliaridis S. Masticatory muscle activity in myotonic dystrophy. *J Oral Rehabil.* 1996;23(1):5-10.
9. Darin N, Tulinius M. Neuromuscular disorders in childhood: a descriptive epidemiological study from western Sweden. *Neuromuscul Disord.* 2000;10(1):1-9.
10. Kaufman J, Friedman JM, Sadowsky D, Harris J. Myotonic dystrophy: surgical and anesthetic considerations during orthognathic surgery. *J Oral Maxillofac Surg.* 1983;41(10):667-71.
11. White RJ, Bass SP. Myotonic dystrophy and paediatric anaesthesia. *Paediatr Anaesth.* 2003;13(2):94-102.
12. Zanoteli E, Yamashita HK, Suzuki H, Oliveira AS, Gabbai AA. Temporomandibular joint and masticatory muscle involvement in myotonic dystrophy: a study by magnetic resonance imaging. *O Surg O Med O Pathol O Radiol Endod.* 2000;94(2):262-71.
13. Casey EB, Aminoff MJ. Dystrophic myotonia presenting with dysphagia. *Br Med J.* 1971;2(5759):443.
14. Chiappetta AL, Oda AL, Zanoteli E, Guilherme A, Oliveira ASL. Oropharyngeal dysphagia in the myotonic dystrophy: phono audiological evaluation and nasofibrolaryngoscopic analysis. *Arq Neuropsiquiatr.* 2001;59(2-B):394-400.
15. Crenshaw J. Myotonic dystrophy. *J Am Dental Assoc.* 1996;72(8):1405-10.
16. Roig M, Balliu PR, Navarro C, Brugera R, Losada M. Presentation, clinical course, and outcome of the congenital form of myotonic dystrophy. *Pediatr Neurol.* 1994;11(3):208-13.
17. Fragola PV, Luzi M, Calò L, Antonini G, Borzi M, Frongillo D, et al. Cardiac involvement in myotonic dystrophy. *Am J Cardiol.* 1994;74(10):1070-72.
18. Dalal FY, Bennett EJ, Raj PP. Dystrophia myotonica: a multisystem disease. *Can Anaesth Soc.* 1972;19(4):436-44.
19. Florek RC, Triffon DW, Mann DE, Ringel SP, Reiter MJ. ECG abnormalities in patients with myotonic dystrophy. *West J Med.* 1990;153(1):24-7.

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