Myotonic Muscular Dystrophy May Be Labelled As Mental Retardation: A Case Report

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Myotonic muscular dystrophy is a multisystem disease inherited as an autosomal dominant trait characterized by progressive muscle weakness, atrophy and myotonia. Mental retardation may be a symptom of the disorder, but not always in this way. We an 11-year-old boy erroneously labelled as mentally retarded due to the presence of myopathic facial appearance. We emphasise the importance of considering an underlying myopathic condition in the differential diagnosis of mental retardation.

Key Words: Myotonic Muscular Dystrophy, Mental Retardation, Facial Expression

Myotonic muscular dystrophy (MD), or Steinert disease, is a multisystem disorder inherited as an autosomal dominant trait and has a variety of manifestations in addition to muscle weakness and myotonia. 1 MD is commonly associated with mental retardation (MR), but not always in this way2. We present an 11-year-old boy erroneously labelled as mentally retarded due to the presence of myopathic facial appearance.

CASE REPORT

Our patient was an 11-year-old boy who had a 4-year history of difficulty in writing, slurred speech and attention deficit. Past medical history of the perinatal and early infantile periods was unremarkable. He had a normal birth weight of 3200 g, length of 50 cm and head circumference of 36.5 cm. He was the first child of unconsanguineous parents. He had no another sibling. The father had frontal baldness and azoospermia as well as his son’s symptoms and signs. Neurologic examination of his grandmother (father’s mother) was normal except cataract. Besides these, the family history was unremarkable.

Physical examination was unremarkable. Neurological examination revealed facial weakness, inverted V- shaped upper lip (Fig. 1), slurred speech, steppage gate, distal muscle wasting in upper extremities, the flattening of the thenar and hypothenar eminences and clinical myotonia which was demonstrated when the patient was asked to tighten his fist and then to open it quickly. He also had a percussion myotonia in his tongue (Fig. 2). His IQ was 83 %. His neurological examination was otherwise normal. No cataract finding was found on ophthalmologic examination.
The results of the laboratory tests were as follows: routine complete blood count, biochemical and thyroid function tests, starvation blood glucose, creatinin kinase, adrenocorticotropic hormone, cortisol and IgG were all normal. The electromyographic evaluation performed on abductor muscle of right thumb revealed myotonic discharges without myopathic or neurogenic involvement. His genetic study revealed 600 repetitions of the cytosine-thymine-guanine (CTG) trinucleotide. The numbers of CTG triplet of his father and grandmother were 500. The electromyographic evaluation and the genetic study of this patient and his examination by a specialist led to the diagnosis.

DISCUSSION

In this patient, the diagnosis of MD was performed by presence of myopathic facial appearance, articulation disorder, distal muscle weakness (especially in hands) and clinical myotonia. The typical discharges in electromyogram supported the clinical diagnosis. Diagnosis was further confirmed by genetic analysis revealing the 600 repeats of the CTG codon in the myotonic dystrophy gene.

Myopathic face and muscle weakness are also seen in other neuromuscular disorders. Unlike other neuromuscular disorders, MD has myotonia and distal weakness\(^1\). Although most patients with myotonia have MD, it is not specific for the disorder. Myotonia is also seen in several rare disorders including sodium-channel myotonias such as paramyotonia congenita and hyperkalemic periodic paralysis with myotonia, chloride channel myotonias such as myotonia congenita (Thomsen disease) and recessive generalized myotonia (Becker disease), and myotonic chondrodystrophy (Schwartz-Jampel disease).\(^3,4\) Unlike Thomsen disease, the myotonia in MD is less prominent. In addition, there are fixed weakness and characteristic systemic features. Furthermore, patients with Thomsen disease have a 'herculoid' appearance. Patients with Schwartz-Jampel disease have a typical physical appearance (dwarfism, blepharophimosis and joint contractures) and have distinctive myotonic discharges on electromyography. Distinguishing, clinically, chloride channelopathies from sodium channelopathies may be difficult, since myotonia is prominent and muscle hypertrophy is common in these two disorders. However, while sodium channelopathies produce increased myotonia, chloride channelopathies produce decreased myotonia by exercise. In addition, paramyotonia is a temperature-related myotonia that is aggravated by cold.\(^3\)

MD is a slowly progressing hereditary disease and causes dysfunction in multiorgan systems. It is the second most common muscular dystrophy in childhood, with an incidence of 1 in 8000 to 18000\(^1\). This disease has three forms: congenital, classic and mild forms. Each of these forms may begin at the earlier ages\(^4\). MD is a neuromuscular disorder exhibiting 'the phenomenon anticipation' in which each consecutive generation shows more severely clinic symptoms in terms of a previous generation.\(^5,6\) The congenital form of the disorder is a typical example of 'the phenomenon' in MD. The minimally mother affected may give birth to a baby with severe congenital MD\(^4\).

In severe congenital form, the arthropathies ranging from club foot to severe joint contractures are seen. This form can also exhibit severe generalized hypotonia, facial weakness and feeding and respiratory problems. The majority of the patients in the form are lost within the first year of life.
Myotonia may not be present both clinically and electromyographically by the 5th year. The classic form is characterized by distal muscle weakness, myotonia, testicular atrophy, slurred speech, cataract, disrhythms, frontal baldness and restrictive pulmonary disease, whereas the major symptom of the mildest form is cataracts with minimal or no muscle weakness. Clinic symptoms and signs of our case and his father were consistent with typical classic MD, but grandmother mildest form.

The genetic defect in MD is an expanded CTG trinucleotide repeat in the 3′-untranslated region of a protein kinase gene on chromosome 19q13. The degree of the clinical symptoms in the disorder correlates with the number of CTG repeats. Although normal persons have 35-40 repeats, the mildest, classic and congenital forms of the disease are associated with 50-80, 100-1000 and >1000 repeats, respectively. Our case and his father had 600 repeats being consistent with classic MD. However, it was interesting that clinic manifestations of his grandmother was consistent with the mildest form and that the number of CTG repeats showed classic pattern is unknown. Tanaka et al. proposed that maternal inheritance may be a possible intrauterine factor affecting the brain developing. Even though the patients with MD do not show any mark of MR, they may be labelled as MR due to probably their peculiar facial expression caused by the atrophy of the facial and masticatory muscles. Although our case had no signs of MR, he had been labelled as mentally retarded at school and even at the hospital by pediatricians. Since there were serious negative effects of an incorrect label of MR on both child’s and parents’ lives, such misdiagnoses must be avoided. We, therefore, would like to stress that pure MR should not be diagnosed before it is carried out by a proper psychometric evaluation as well as a thorough neurologic assessment. In addition, the early diagnosis of MD in a family is very important for genetic counselling and prenatal diagnosis and cannot be omitted.

REFERENCES


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