Case Report

Myotonic Dystrophy presenting as chronic diarrhoea and anal incontinence: A Case Report

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Abstract

Myotonic dystrophy (MD) or Steinert's disease is a rare cause of chronic diarrhoea and anal incontinence. We present the case of a 37 year old man who presented with chronic diarrhoea and faecal incontinence as the initial symptoms to our outpatient clinic and was later diagnosed with MD. In the presence of chronic diarrhoea and faecal incontinence with muscle weakness, neuromuscular disorders such as myotonic dystrophy should be considered in the differential diagnosis.

Keywords: Myotonic dystrophy, Steinert's disease, Chronic diarrhoea , Anal incontinence

Introduction

Steinert's disease, also called myotonic dystrophy (MD), is an autosomal dominant inherited muscular disorder which usually presents in early adulthood, is characterized by progressive muscular weakness, myotonia, frontal baldness, cataract and testicular atrophy. Gastrointestinal involvement is frequently observed in MD. Abnormalities of smooth muscle lead to gastrointestinal motility disorders with chronic diarrhoea and rarely incontinence.

Case report

A 37year old man from rural areas of West Bengal presented with diarrhoea three to four times per day, without pus or blood, for the last 9 months to the outpatient department. Diarrhoea was frequently combined with faecal incontinence which had a marked negative impact on his social life.

He also described muscle weakness especially in the distal parts of his upper extremities that was present for last 7years. He was having difficulty with holding objects and performing other activities of routine daily living.

His family history revealed that several members of his family were suffering from similar problems. Examination showed that his vital signs were normal, mild pallor was present but no icterus or lymphadenopathy was noted. Facies was notable for atrophy of bilateral masseter, temporalis and sternocleidomastoid with frontal baldness giving a characteristic 'hatchetfacies' appearence. Cardiovascular and respiratory system examinations were normal. Abdominal examination showed no organomegaly or tenderness. Neurological revealed weakness and atrophy in the neck, face, and distal extremity muscles in both upper and lower limbs, Deep tendon reflexes were intact. Percussion myotonia was elicited. Bilateral cataract was found on ocular examination.

Laboratory tests showed normal whole blood count, serum levels of glucose, urea, creatinine, calcium, sodium, potassium were within normal limits. Creatine kinase (CK) was normal. Stool examination for ova and parasites was negative. Microscopic evaluation of faeces did not reveal any blood or leucocytes. No bacteria or parasitic growth was observed on stool cultures. Colonoscopy demonstrated no abnormality with normal appearing mucosa.

Histopathological examinations of the gastrointestinal system endoscopic biopsy samples detected no inflammation or parasite.

Anorectal manometric study showed that internal and external sphincters were weak with reduced resting and squeezing anal pressures but rectal sensation was within normal limits. Internal anal sphincter relaxed normally on rectal distension. External anal sphincter electromyography (EMG) showed myopathic features and polyphasic high-amplitude motor units. EMG showed signs of a myopathic process in the distal muscles of his extremities.

Typical myotonic discharges were recorded. These results were compatible with MD. Additional tests were done to exclude comorbid illnesses of MD. His electrocardiogram demonstrated normal sinus rhythm with no signs of ischemia. No abnormality was detected in echocardiography. Hormonal studies including free T4,

TSH, morning cortisol, LH, FSH, total testosterone, free testosterone showed no abnormal results. The final diagnosis of MD was confirmed with genetic evaluation. A course of Norfloxacin partially alleviated his diarrhoea. Patient was demonstrated special exercises to increase his pelvic muscle tone. He was counselled about the nature of the illness and advised family screening.

Figure 1: showing characteristic facies-frontal baldness with atrophy of masseter and temporalis muscle (typical hatchet facies)

Discussion

Myotonic muscular dystrophy, also called Steinert's syndrome is a neuromuscular disease characterized by myotonia or difficulty in muscular relaxation, atrophy and weakness of skeletal muscle [1].

Gastrointestinal involvement is frequently observed in MD patients, and digestive complaints may be the first sign of the disease [2].

Diarrhoea, usually accompanied with malabsorption, steatorrhea, and crampy abdominal pain, is a frequent complaint in MD patients [3].

Diarrhoea and malabsorption has been attributed to reduced peristaltic activity, leading to bacterial overgrowth [4].

For the diagnosis of bacterial overgrowth, hydrogen and methane breath tests are the most important diagnostic methods [5].

Norfloxacin is often used for this symptom. But role of prebiotics and probiotics is not validated by studies.

Our patient was prescribed norfloxacin twice daily which helped control his diarrhoea.

Diarrhoea may be accompanied with faecal incontinence as in our index patient.

Anal manometric studies usually report a decrease in both the resting and squeezing pressure [6].

Treatment of defecation disorders involves electro-anal stimulation and biofeedback which was not possible in our hospital.

Also surgical management of anal incontinence is unsatisfactory in the long term [2]. Thus patient was treated conservatively and advised against surgery.

Conclusion

In clinical practice, the persistence of diarrhoea and faecal incontinence with muscle weakness should lead the physician to perform an anal manometric study and EMG as MD becomes a strong possibility in such situations.

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