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Case Report

A case report of Becker's variety of myotonia congenita

Nurul Amin Khan[1], Shaheen Wadud[2], Torikul Islam[3], Liton Chandra Ghosh[4]

[1]Associate Professor, Department of Neuromedicine, Dhaka National Medical College, [2]Associate Professor, Department of Neuromedicine, Dhaka National Medical College, [3]Registrar, Department of Neuromedicine, Dhaka National Medical College, [4]Associate Professor, Department of Nephrology, Dhaka National Medical College.

Abstract:

Myotonia congenita is a disease of skeletal muscle. It usually begins at childhood and problem in muscle relaxation after contraction of muscles. It involves lower limb muscles more than any other group of muscles. It causes muscle stiffness and interfere with movement like walking, running and other daily activities. Our case was a thirteen years old Muslim male presented with history of stiffness and transient weakness. On examination we found that myotonia in grip, tongue and other muscles with hypertrophy of the proximal muscle and myotonia was more marked on lower limbs. CPK was mildly raised, electro-physiological test showed typical dive bomber's myotonic discharge and muscle biopsy showed the myopathy. On later presentation, more severe disease and marked hypertrophy of proximal muscle established the recessive Becker type of myotonia. Explanation of the nature of disease with symptomatic treatment by phenytoin, improved the patient symptoms. Familial counselling about this disease had also been carried out.

Introduction

The non-dystrophic myotonia are skeletal muscle ion channel disorder traditionally considered to be distinct from myotonic dystrophy because of the absence of progressive weakness and systemic features. The non-dystrophic myotonia are now known to be caused by the dysfunction of key skeletal muscle ion channels and includes myotonia congenita, para-myotonia congenita and sodium channel myotonia.

The worldwide prevalence of non-dystrophic myotonia has been estimated to be 1 in 100000 (Emry, 1991). Prevalence may vary considerably between geographical regions. Myotonia congenita alone was estimated to have a prevalence of between 07 in 10 in 100000 in Scavadinavia.[1,2] Myotonia congenita is basically a chloride channel myopathy CLCN-1, the gene encoding the major skeletal muscle chloride channel, is localized to chromosome 7q35 locus. The mutation of this gene can cause either dominant Thomsen or recessive becker's type myotonia.[3] Myotonia; a tonic spasm of muscle after forceful voluntary contraction stands as cardinal feature of myotonia and repeated contractions wear it out and marked profound after inactivity.[4] Myotonia affects the face, jaw, tongue, pharynx, arm and legs and is more severe than that in myotonic dystrophy.[5]

Figure–I: Figure showed Herculean appearance and difficulty from sitting to standing position.

We reported the case as because, early detection and treatment of this variety may improve the patient outcome and genetic counseling helps the further emergence of disease in this family. Myotonia congenita due to chloride channel defects can be distinguished from the sodium channel myotonia (Hyperkalemic PP, para-myotonia congenita, sodium channel myotonia) by the rather striking muscle hypertrophy and by DNA mutational screening.[6]

Case report

A thirteen years old Muslim male, gave history of consanguinity marriage, was admitted with the complaints of stiffness of both lower limbs and difficulty in walking with problem in initiating his gait. He also developed hardening of calf muscle intermittently. He also noticed recurrent fall during walking and impairment of recent memory. Patient complained difficulty from sitting to standing posture and failure to relax his hand grip easily. His oral cavity remained open during contraction of forehead. Stiffness markedly increased with exercise after initial improvement. He also stated that tongue movement abnormality after any pressure over tongue. Patient also developed weakness after playing and running. Fine works and writing could not able to do like his friends or mates. Patient attendant did not give any history of birth injury or delayed milestone and head or spine injury. He gave no history of substance abuse or use of traditional medicine. This patient's parent denied such problem in their family. There was no involvement of bladder and bowel. On examination – patient's body built was average and he was cooperative and oriented. Pulse 80/m, BP- 100/65 mmHg, Temp- Normal, Jaundice and Cyanosis – Absent, Thyroid and skin condition – revealed no abnormality and rest of the general examinations appear to be normal. On systemic examination, most of the system revealed no abnormality except locomotor and nervous system. Proximal and calf muscle were hypertrophied. There was evidence of the myotonia during grip and over tongue. No myotonia on eyelids. Myotonia improved just after exercise but patient felt weakness after heavy exercise. Speech was normal, muscle power was normal, all deep reflexes were mildly diminished and planter bilaterally flexor. Other cranial nerves examination revealed no abnormalities and sensory examination were also normal. Relevant investigations showed CBC – normal, RBS – 5.3 mmol/l, S. Creatinine – 0.9 mg/dl, S. Bilirubin – 0.2 mg/dl, S. SGPT – 60 u/l, Na – 142 meq/l, K – 3.7 meq/l, Chloride – 103 meq/l, S- Calcium – 9 mg/dl, PBF – non-specific abnormality, S. CPK – 269 u/l, Thyroid function test – normal, USG of W/A – normal, HB-electrophoresis – normal, HBsAg – Negative, X-ray chest P/A view – normal, Urine R/E – normal. ECG within normal limit and Echocardiography also normal, muscle biopsy showed the evidence of myopathy and lastly electrophysiological study showed normal nerve conduction, but EMG showed myotonic discharge – typically repetitive discharge of varying amplitude and frequency along with classical dive bomber's sound. Genetic evaluation e.g. CTG repeat mutation in DNA is not possible in our country so these test were excluded. Considering the autosomal recessive pattern inheritance and clinical picture and the diagnosis of Becker's variety of myotonia congenita was entertained. We started the treatment by anticonvulsants, clonazepam and patient was improved. Patient referred for genetic counseling and rehabilitation. During follow up, patient did not develop further deterioration.

Discussion

Myotonia congenita typically two types – Type -1, autosomal dominant myotonia congenita and type- 2, Becker's type recessive myotonia congenita. Myotonia congenita first described by the Thomsen in 1876 in his own family. Myotonia is persistent contraction of skeletal muscle following stimulation leading to generalized muscular hypertrophy and herculean appearance. In fig-I showed the herculean appearance and difficulty to stand from sitting position. Myotonic muscle stiffness is painless and exacerbated by anxiety, cold and fatigue. The autosomal dominant disease should be differentiated from the recessive generalized myotonia which is similar but has a later age of onset, a more severe in nature and manifest transient muscle weakness during muscle exertion after rest.[7] In general, patient with recessive disease experience transitory bout of weakness after period of disuse and may developed progressive myopathy; in than the dominant form and becker's myotonia is more common than Thomsen disease.[8] The age of onset is in Thomsen variety usually within first decade but recessive variety at 10 to 14 years.[8] Our patient presented at the age of thirteen years. Some clinical finding is more common in recessive than the dominant form but considerable overlap exit, and recessive form more severe, is frequently associated with muscle hypertrophy and with diminished deep reflexes.[9] This case manifested as hypertrophy of proximal and calf muscle and associated with diminished deep reflexes, but our case did not show any evidence of cranial nerve involvement except tongue that is usually predominantly involved in myotonic disorder.[10] The brunt of the Becker's disease fall on the lower limbs, probably as a result of work hypertrophy, since the quadricep's and other muscles are in continual state of contraction.[10] This case showed the marked myotonia in lower limb. Positive family history, early appearance and lack of progression of myotonia and generalized muscular hypertrophy

distinguish the myotonia congenita form myotonic dystrophy. Para-myotonia congenita is a myotonia that is precipitated by cold.[11] Our patient did not show any cold sensitivity. The testicular atrophy, cardiac abnormalities, frontal baldness, and cataract- the features that characterize myotonic dystrophy are conspicuously absent.[4] Our patient did not show any systemic features but lower limb involvement was more marked than upper limbs. On relevant investigation – The CPK was mildly raised, electro-physiological test was typically showed myotonic discharge and muscle biopsy showed evidence of myopathy. Becker's type myotonia showed the features of progressive myopahty.[8] In one study form Portugal, patients were treated with different drug to determine the therapy for optimal relief of myotonia. Five patients responded to phenytoin, one to carbamazepine, three to acetazolamide and none to quinidine or procanamide.[7] Another double-blind study compared phenytoin and carbamazepine on myotonic patient; there was a good response to both drug but phenytoin should decreased efficiency at high-dose.[12] Dantrolene may be tried in patient who do not respond to usual drugs.[13] This patient responded well with phenytoin. Myotonic patient sensitive to many anesthetic drugs. Spinal and epidural anesthesia safer than general anesthesia. This patient also has a higher risk of pulmonary aspiration and post-operative pneumonia.[14] DNA mutational screening is not possible in our country, so we highlighted the clinical features and investigations to establish either dominant or recessive type myotonia congenita.

Progression of the disease continues to about thirty years of age and according to sun and Strib, the course of the illness thereafter remains unchanged.

Conclusion

When faced with a patient complaining of muscle weakness and stiffness, the neurologist must able to distinguish between dystrophic and non-dystrophic myotonia. If a careful medical history and neurological examination are undertaken in combination with electro-physiological studies, it is possible to determine which genetic test should be carried out to confirm clinical impression. This disease needed further investigations, advance treatment strategy and genetic counseling for better outcome.

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