ORIGINAL RESEARCH PAPER

Neurology

'FAINTING GOAT SYNDROME' – A RARE CASE OF MYOTONIA CONGENITA

KEY WORDS: channelopathy, EMG-NCV, congenital

Dr. Prajakta Mhatre III year PG Resident, MD Internal Medicine, MGM Medical College & Hospital, Kamothe, Navi Mumbai

Dr. Sandeep Rai* MBBS, DNB Medicine, Associate Professor, Department of Medicine, MGM Medical college & Hospital,Kamothe, Navi Mumbai *Corresponding Author

Dr. Kishor Jadhav MD Medicine, DM Neurology, Associate Professor, MGM Medical College & Hospital, Kamothe, Navi Mumbai.

ABSTRACT

Myotonia Congenita affects approximately 1 in 10,00,000 people worldwide. It is also called as Congenital myotonia. It is a congenital neuromuscular channelopathy that causes impairment of skeletal muscles (muscles that are used for movements).This condition is sometimes referred to as fainting goat syndrome. We, hereby, present a case of young male with features suggestive of myotonia congenita.[1]

CASE REPORT :

A 33 yrs old young male, right handed, married, driver by occupation, however part-time works as clerk in a pharmaceutical company, born of non-consanguinous marriage. He presented with complaint of stiffness of joints and body especially after taking rest which started since 10-12 years. It was insidious in onset and gradually progressive in nature. He complains that initially whenever he was doing any movement, its gets difficult, however subsequently as he keeps on moving, he feels better and can mobilize easily. He also falls easily when he is pushed.

No other complaints.

No significant details of family history noted, however his father had some difficulty in upsquat.

He denies addiction. No bowel & bladder incontinence.

On Examination, his pulse was 82 beats per minute which was regular in character. His blood pressure was recorded as 130/80 mm of Hg over the left brachial artery in sitting position. On detailed systemic examination of central nervous system, it was found that his higher mental functions were normal. All cranial nerves were intact. Sensory & motor system was intact.

However, it was observed that he had hypertrophy of calf muscles and tongue fasciculations were present.

On doing routine investigations of the patient, his complete blood count, liver function test, renal function test, serum electrolytes were within normal limits. 2-Decho was normal with LVEF 60%. No abnormality was detected in Chest Xray and ECG.

Nerve conduction and EMG findings were compatible with the diagnosis of Myotonia Congenita- dominant variety most likely.

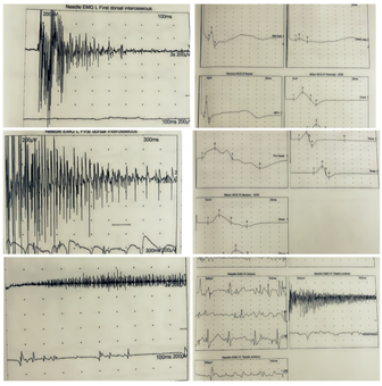

Fig 1: Nerve conduction & EMG study

Table 1 : Needle Electromyography

Muscle Name	Spontaneous Activity	Voluntary Activity	Interference Pattern	Remark
Left first dorsal Interosseous : (Ulnar Nr C8, T1)	Myoyonic discharges+	Normal	Full	Myotonic discharges with normal MUP configuration
Left Orbicularis Oris :	Myoyonic discharges+	Normal	Full	Myotonic discharges with normal MUP configuration
Right Deltoid (Axillary Nr C5, 6)	Myoyonic discharges+ (slow rate)	Normal	Full	Myotonic discharges with normal MUP configuration
Right Tibialis Anterior : (Deep Peroneal Nr, Common Peroneal Nr, Sciatic Nr L4,%)	Myoyonic discharges+	Normal	Full	Myotonic discharges with normal MUP configuration

DISCUSSION :

Myotonia Congenita is basically characterized by delayed relaxation after forceful contraction and rigidity.[2] In recessive mutations, these prolonged muscle contractions are more in leg muscles, however, in dominant mutations, it is more in hands, face and eyelids.[3] There are two main types i.e., Becker disease and Thomsen disease.[4] It was observed that in many patients being in cold environment aggravated the symptoms.[5]

The warm-up phenomenon was described in the year 1876, however its etiology is not clear. This means that if a person is sitting and then immediately he decides to walk, after few steps his muscles will stiffen because of which he usually slows down and then stops. But then after few steps as muscles loosen up he walks or climbs at normal pace. This effect usually lasts for about 5 minutes.[6] It was found that in the skeletal muscle there is Na + channel 1.4 inactivation that is supposedly responsible for warm up phenomenon.[7]

Myotonis Congenita is basically caused by a gene-CLCN1 encoding the CIC-1 chloride channel. In humans it is caused b y loss of function in the gene CLCN1.[8]

Myotonia Congenita affects approximately 1 in 10,00,000 people worldwide.[9]

CONCLUSION :

Myotonia Congenita is a rare disease and should always be suspected after a thorough clinical examination and should always be confirmed after genetic testing.

REFERENCES :

(1) Wikipedia- Myotonia congenital (https://en.wikipedia.org/ wiki/ Myotonia_ congenita)
(2) Gutmann, Laurie; Phillips Lh, Lawrence (2008). "Myotonia Congenita". Seminars in Neurology. 11 (3): 244–8. doi:10.1055/s-2008-1041228. PMID 1947487.
(3) Lossin, Christoph; George, Alfred L. (2008). "Myotonia Congenita". In Rouleau, Guy; Gaspar, Claudia. Advances in Genetics. 63. pp. 25–55. doi:10.1016/S0065-2660(08)01002-X. ISBN 978-0-12-374527-9.
(4) Rüdel, Reinhardt; Ricker, Kenneth; Lehmann-Horn, Frank (1988). "Transient weakness and altered membrane characteristic in recessive generalized myotonia (Becker)". Muscle & Nerve. 11(3): 202–11. doi:10.1002/mus.880110303.
(5) Nielsen, VK; Friis, ML; Johnsen, T (1982). "Electromyographic distinction between paramyotonia congenita and myotonia congenita: Effect of cold". Neurology. 32 (8): 827–32. doi:10.1212/WNL.32.8.827. PMID 7201578.
(6) Birnberger, K. L.; Rüdel, R.; Struppler, A. (1975). "Clinical and electrophysiological observations in patients with myotonic muscle disease and the therapeutic effect of N-propyl-ajmalin". Journal of Neurology. 210 (2): 99–110. doi:10.1007/ BF00316381. PMID 51920
(7) Lossin, Christoph (2013). "Nav1.4 slow-inactivation: Is it a player in the warm-up phenomenon of myotonic disorders?". Muscle & Nerve. 47 (4): 483–7. doi:10.1002/mus.23713. PMID 23381896
(8) Trip J, Faber CG, Ginjaar HB, van Engelen BG, Drost G (2007) Warm-up phenomenon in myotonia associated with the V445M sodium channel mutation. J Neurol 254(2):257-258
(9) Emery, Alan E.H. (1991). "Population frequencies of inherited neuromuscular diseases—A world survey". Neuromuscular Disorders. 1 (1): 19–29. doi:10.1016/0960-8966(91)90039-U. PMID 182277