ORIGINAL RESEARCH PAPER

Neurology

A RARE CASE OF DYSFERLINOPATHY – LIMB GIRDLE MUSCULAR DYSTROPHY TYPE 2B

KEY WORDS: LGMD2B, Dysferlin

Dr. Buddhadeb Majumder* Junior Resident, Dept Of General Medicine D Y Patil Medical College and Hospital, Nerul, Navi Mumbai*Corresponding Author

Dr. Pradeep Tiwari Consultant Neurologist D Y Patil Medical College and Hospital, Nerul, Navi Mumbai

Dr. Santwana Chandraka Professor and HOU, Dept Of General Medicin D Y Patil Medical College and Hospital, Nerul, Navi Mumbai

Dr. Nivedita Moulik Professor and HOD, Dept Of General Medicine. D Y Patil Medical College and Hospital, Nerul, Navi Mumbai

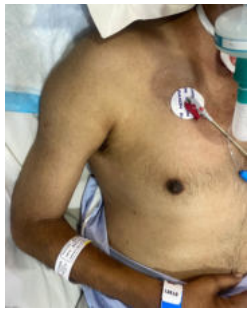
CASE REPORT

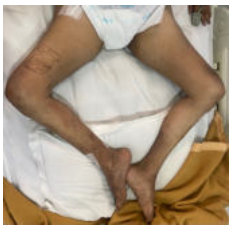
A 49 years old male presented with complaints of difficulty in breathing and swallowing since 1 month. Patient had a normal childhood till 7 years of age when he started developing bilateral lower limb weakness causing recurrent fall which progressed gradually over time and since 39 years of age he was unable to stand from squatting position without support and since 43 years of age he was not able to walk even with support.

On examination Hypotonia present in all the limbs, Power 0/5 in BL Lower Limb and 2/5 in BL upper limb. Deep-Tendon-Reflexes were reduced. On admission Patients oxygen saturation was 90% on room air and ABG was suggestive of Type 2 Respiratory failure. Patient was given NIV trial but Patient required to be intubated. On day 7 tracheostomy was done in-view-of anticipated prolonged ventilatory support and airway protection.

EMG was Abnormal in all 4 limbs. Serum CPK-884 mcg/L. Muscle Biopsy was done, which was suggestive of Muscular Dystrophy and IHC profile was Negative for Dysferlin. Patients Cardiac Status was within normal range.

Based on clinical and laboratory findings Diagnosis of DYSFERLINOPATHY – LGMD 2B was made. Symptomatic treatment was given to the patient. Patient was discharged on day 21 of admission on BIPAP support. Patient is being Followed up every 4 weeks since last 6 months. There is no further worsening of his Condition.





DISCUSSION

Mutation of the gene encoding for Dysferlin, located on Chromosome 2p13 lead to clinically Heterogenous Myopathy. Some patients show a Limb-Girdle pattern of weakness – LGMD2B. While others present with weakness and atrophy of the calf muscles – Miyoshi Myopathy.

Some patients may have earlier involvement of anterior tibial muscles. 80% patients of dysferlinopathy manifest as Distal Myopathy , whereas only 6% have LGMD pattern of weakness. Dysferlinopathies typically present in adolescence or early adult life. Progression is usually slow. Patients usually lose ambulation in their 2nd to 3rd decade of life. Rarely patients are able to walk on their own till late in life and can present late to the Healthcare facilities. Intra familial variabilities exist in the pattern of weakness and disease progression.

Several characteristics that can indicate towards Dysferlinopathy – (1) Autosomal Recessive inheritance or sporadic (2) Variable age of onset (3) Limb Girdle pattern of weakness with Gastrocnemius usually weaker than Tibialis anterior, although the weakness can be variable (4) Slow progression (5) variable clinical phenotype even within the same family.

There is no definitive treatment for Dysferlinopathy. Hence, treatment is mostly symptomatic relief. Goals of therapy include maintaining mobility and functional independence and managing associated complications and maximizing quality of life.

CONCLUSION

Dysferlinopathy has a wider clinical spectrum and the final diagnosis to be made by Dysferlin protein analysis of the biopsied muscle OR molecular analysis of Dysferlin gene. Cardiac or pulmonary involvement is uncommon but when it does occur, it is usually asymptomatic and late in the course of the disease. The management is supportive care.

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