Case Report**Gower Laing: Early adult onset distal myopathy type III (MPD I): A Case Report.****Thivacaren S¹, Suramiya AAA¹, Paherathy A¹, Arasalingam A²**¹Teaching Hospital Jaffna, ²Dept. Of Medicine, Faculty of Medicine, Jaffna.**Abstract:**

A rare case of sporadic Gower Laing: Early adult-onset distal myopathy type III (MDP I) presenting to a medical ward in northern Sri Lanka is presented here. A 39-year-old female present with difficulty in walking from age 10 and was investigated leading to the above diagnosis. This is a rare autosomal dominant distal myopathy and there have been no similar cases reported from the Northern Province of Sri Lanka, as well as in Sri Lanka.

Case History

A -39-year old female, presented with insidious onset difficulty in walking since 10 years of age. She denies worsening of weakness following exercise, exercise induced hematuria, cramps, muscle twitching or fasciculations. She was educated up to grade ten, had no learning difficulties or significant cognitive impairment and her education was interrupted due to difficulties in mobility. Birth and developmental history were unremarkable. At presentation she was able to carry out her activities of daily living with assistance. She had a cataract and conductive hearing loss. She did not have a history suggestive of endocrinopathies, seizures, headaches, or any other significant neurological conditions. Past medical history was significant for smear negative pulmonary tuberculosis at the age of 32 and completed the anti-tuberculosis (CAT 1 regimen) therapy without any medication induced

side effects. However, she had presented with post tuberculosis bronchiectasis at 37 years of age. She was not on any long-term medication. There was no evidence of a similar illness amongst first- and second-degree relatives for 2 generations. She was from economically unstable family.

She was cachectic and wasted with a BMI of 17kg/m². A tracheostomy scar was visible, and she had laboured breathing using accessory muscles of respiration. A facial asymmetry and a postural tremor was noted. Neurlogical examination revealed reduced visual acuity due to cataract; fundoscopy showed bilateral partial cataract with no evidence of optic atrophy, retinitis pigmentosa or any other background peripheral retinal abnormality. There was no ophthalmoplegia, but she had a unilateral lower motor neuron type seventh nerve palsy and wasting and fasciculations of the tongue. Upper limbs showed sparing of the intrinsic muscles of the hand, and mild weakness of distal muscles specially the finger extensors. There was significant wasting of the distal lower limbs, pes cavus and trophic nail changes. Distal power was significantly reduced in all muscle groups more in the anterior compartment specifically involving the extensor hallucis longus and the anterior tibialis with significant wasting of the anterior tibialis. She had mild proximal muscle weakness. Reflexes were symmetrical however ankle reflex was diminished. The remaining neurological examination was unremarkable.

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Submitted November 2021, Accepted December 2021

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There was no clinical signs of cardiomegaly or organomegaly. She was dyspnoeic and was being treated for a respiratory infection.

Her CPK levels were normal and all other biochemical and haematological investigations were normal except for microcytic hypochromic anaemia suggestive of iron deficiency anaemia. Her metabolic panel including thyroid function, serum calcium, magnesium and phosphate normal. Echocardiography was unremarkable with no evidence of cardiomyopathy or valvular lesions. Muscle biopsy from the calf revealed muscle tissue showing a mild variation of fibre size and shape, and significant amount of centralized nuclei (10%), with degenerating muscle fibres. Cytoplasmic inclusions, vacuoles suggestive of storage was not seen and there was no evidence of inflammation. Muscle biopsy was conclusive of a distal myopathy. Non contrast CT brain was unremarkable. Gene panel evaluation and further staining and histopathology studies were not possible due to logistic reasons. HRCT showed evidence of bronchiectasis.

With regards to neurological management, she was commenced on physiotherapy. She succumbed two weeks after presentation presenting with difficulty in breathing followed by a cardiac arrest. Postmortem was not performed.

Discussion

Based on the clinical and histopathological findings we considered the differentials for young onset distal myopathies which included Gowers Laing myopathy (early adult onset distal myopathy type III), Rod myopathy (distal myopathy 5), Williams myopathy and Nonaka myopathy (hereditary inclusion body myopathy 2). [1]. However the later

three were excluded on the basis of histological and certain distinctive clinical features. Based on the available clinical data we concluded that she had Gowers Laing myopathy (early adult onset distal myopathy type III – MPD 1) due to a sporadic genetic mutation. Sporadic and familial cases have been reported around the world.

Gower's Laing distal myopathy is an autosomal dominant early onset distal myopathy with predominant affliction of dorsiflexors of the ankle and extensors of the fingers. The hanging toe sign is characteristic due to weakness of the extensor hallucis longus. Weakness of neck flexors though common was not obvious in our patient. Another characteristic feature is a postural tremor which was seen in our patient. The progression is insidious, and the patient stays ambulant with a near normal life span in most cases. Cardiac involvement is rare with serum creatinine kinase being normal or mildly elevated. [1,2] MRI of the lower limbs have demonstrated that the tibialis anterior is the earliest and most commonly affected muscle. [3]

Histopathological changes are non-specific with the common features being fibre size variation, abnormally small type 1 fibers, type 1 or 2 fiber predominance, internal nuclei, minicores, and mitochondrial abnormalities. [1,4] A confirmatory diagnosis requires the presence of suggestive findings as well as a heterozygous pathogenic variant in the MYH7 gene via molecular genetic testing. [1]

Management includes physiotherapy and cardiac follow-up for the development of cardiomyopathy. Being an autosomal dominant disorder genetic counselling is important.

References

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