10.5005/jp-journals-10036-1204

CASE REPORT

A Case of Charcot–Marie–Tooth Disease with Marfanoid Features

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ABSTRACT

Charcot–Marie–Tooth disease (CMT)/Peroneal muscular atrophy is a diverse group of inherited chronic peripheral neuropathies. Afflicted patients develop progressive weakness and atrophy in distal-muscles of limbs and deformities of hands and feet. A case of Peroneal muscular atrophy (CMT) combined with Marfanoid Features, a coincidental congenital disorder is reported because of its rarity.

Keywords: Marfanoid features, Neuropathy, Peroneal muscular atrophy (Charcot–Marie–Tooth Disease).

How to cite this article: Biswas TK, Shenoi AS. A Case of Charcot–Marie–Tooth Disease with Marfanoid Features. MGM J Med Sci 2018;5(3):143-145.

Source of support: MGMIHS

Conflict of interest: None

INTRODUCTION

Charcot–Marie–Tooth disease (CMT)/Peroneal muscular atrophy is a diverse group of inherited chronic peripheral neuropathies. Afflicted patients develop progressive weakness and atrophy in distal-muscles of limbs with diminished deep tendon reflexes, and deformities of hands and feet.[1] CMT disease has three subtypes characterized by different electrophysiological and neuropathological features—CMT1 (Glial Myelinopathy) with slow motor-nerve conduction velocities and demyelinating neuropathy, CMT2 (axonal form) with normal or subnormal motor-nerve conduction velocities and axonal degeneration, and an "intermediate" CMT (myelinoaxonopathy), affecting patients from the same family who have either subnormal or diminished nerve conduction velocities. The most common CMT form is CMT.[1,2]

In this paper we report a rare case of CMT disease who also had characteristic musculoskeletal abnormalities of Marfan's syndrome.

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CASE REPORT

Patient Mr. A, a 42-year-old male, resident of Nanded district in Maharashtra, visited this hospital with complaints of gradually progressive difficulty in walking since 15 years. Patient would trip on himself while walking, associated with slipping of chappals. The patient can still get up from squatting position, though he requires a stick to walk. In addition he also complains of weakness of both upper limbs since 15 years with difficulty in buttoning/unbuttoning his shirt and in holding objects.

These symptoms forced him to discontinue farming, which was his sole means of income. The patient gave a history of trauma to the back 5 years earlier. The patient was taking drugs prescribed by his primary physician for 10 years without any improvement.

A detailed family history revealed that his sister, cousin brother, and nephew had similar problems. Physical examination revealed stable vitals, Marfanoid habitus of tall stature with long slender fingers (Arachnodactily) with height 181 cm > arm span of 173 cm, upper segment 84 cm < lower segment 89 cm (Fig. 1); Thumb's sign (Fig. 2) and Wrist sign present with a high arched palate. Pes cavus deformities of both feet (Fig. 3) were also noted. Neurological examination revealed bilateral wasting of the small muscles of hands (Fig. 4), along with distal muscles wasting of the lower limbs giving a characteristic "inverted champagne bottle" appearance (Fig. 1). He had no sensory deficit and no peripheral nerve thickening. He had no peripheral nerve thickening. All deep tendon reflexes were absent except Triceps jerk (2+) and Knee Jerk (3+). There were no spontaneous/inducible involuntary movements or cerebellar signs. Planters were bilaterally flexor. The patient had a high stepping gait with foot drop. The neuro physician consultation was obtained. Laboratory investigations revealed a normal hemogram with normal serum Vit B12 levels.

Magnetic resonance imaging (MRI) cervical spine revealed posterior bulge of C3-C4 disc indenting anterior subarachnoid space and vertebral osteophytes encroaching bilateral neural foramina. Besides, there was posterior herniation of C5-C6 disc indenting anterior subarachnoid space and vertebral osteophytes encroaching bilateral neural foramina indenting right exiting C6 nerve root. Screening through dorso lumbar spine revealed changes of lumbar spondylosis with posterior herniation of L3-L4,

Fig. 1: Inverted champagne bottle appearance of lower limbs and Marfanoid features

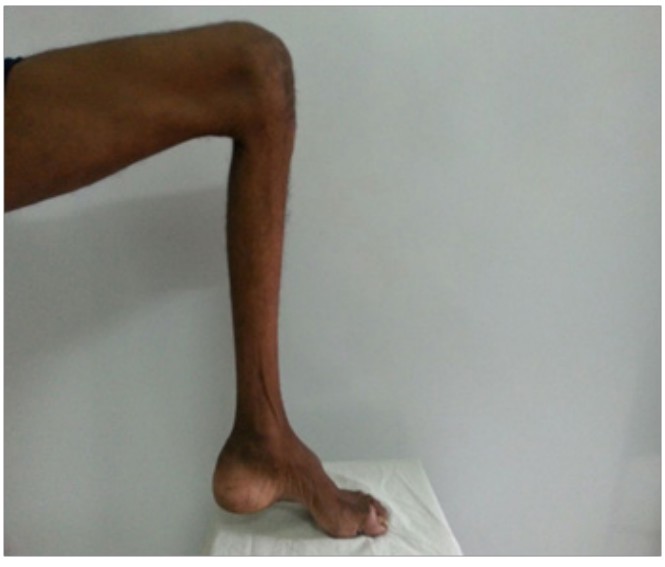

Fig. 3: Pes cavus deformity

L4-L5 and L5-S1 discs indenting thecal sac. He was being treated by his primary physician with pregabalin and muscle relaxant. The nerve conduction velocities (NCV) showed a generalized motor sensory neuropathy (axonal).

As further work-up for Marfan's syndrome, he had 2D Echo-cardiogram and Slit lamp examination of eyes, both of which were normal. Based on the clinical history, familial involvement, physical examination and investigation reports including electrophysiological studies conducted at this center, the patient was diagnosed as a case of peroneal muscular atrophy (Charcot–Marie–Tooth Disease) combined with Marfanoid Features.

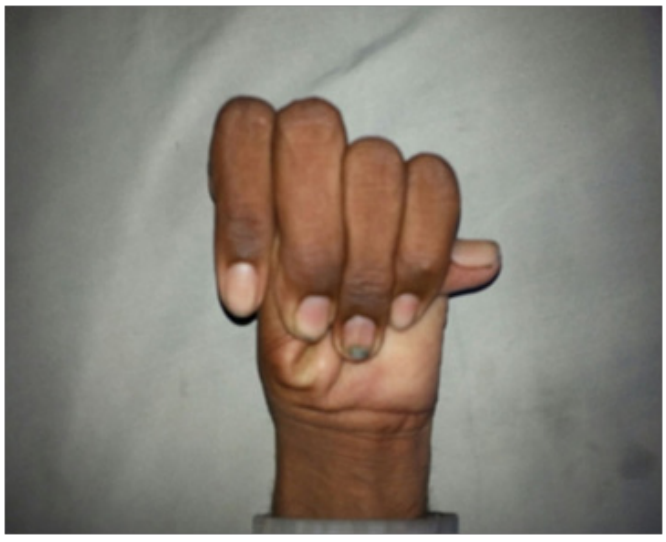

Fig. 2: Thumb sign

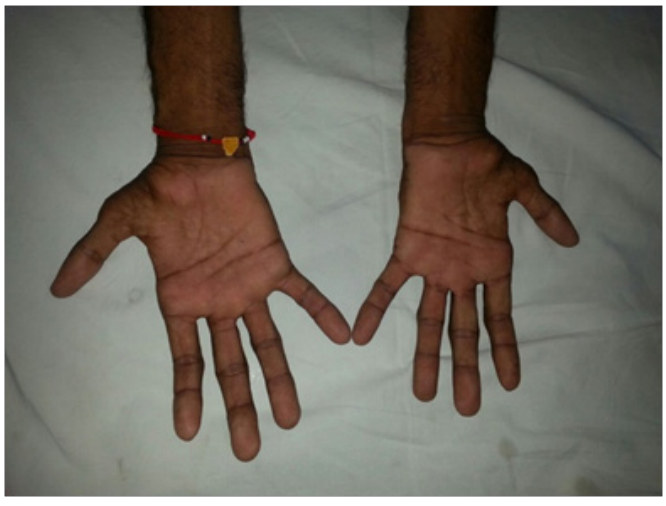

Fig. 4: Wasting of small muscles of hands

DISCUSSION

Peroneal muscular atrophy (CMT disease types 1/CMT1 and types 2/CMT2) was described in 1886 almost simultaneously by Tooth in England and by Charcot and Marie in France.

Charcot–Marie–Tooth type 1 (CMT1) is the most common form of hereditary neuropathy, with the ratio of CMT1:CMT2 being approximately 2:1.[3] Usual presentation is in the first to third decade of life with the weakness of distal leg muscles (e.g., foot drop). However individual patient may remain asymptomatic even late in life. Patients with CMT usually do not have sensory symptoms, which can be helpful in differentiating. CMT from acquired forms of neuropathy in which sensory symptoms generally predominate.

Often patient develops marked atrophy of distal peroneal muscles with tapering of the legs and hypertrophy of the proximal muscles giving 'inverted champagne bottle legs'/'stork leg' appearance (as in the present case).

Atrophy of the distal muscles begins in the feet and legs and later involves small muscles of the hands due to chronic degeneration of peripheral nerves and roots.. The extensor hallucis and digitorum longus, the peronei, and the intrinsic muscles of the feet are affected early in life, and this muscle imbalance produces the bony changes of pes cavus and pied en griffe (high arches and hammertoes). Pes Cavus deformity is likely a combined effect of CMT and Marfanoid features in this patient. Bilateral foot drop results in high "steppage" gait or slapping of feet.

The deep tendon reflexes are diminished or absent, and there may be sensory loss or impairment in the distal part of limbs to a variable extent. Thickening of peripheral nerves may occur. There was generalized are flexia except for triceps jerk (2+) and knee jerk (3+) which can be explained by an early feature of spinal cord compression as seen MRI spine. Inheritance in both CMT1 and CMT2 is usually autosomal dominant. Nerve conduction velocity is severely reduced in type 1 cases, moderately reduced in the X-linked form, and either normal or only slightly reduced in type 2. CMT1 is dominantly inherited myelinopathy and CMT2 is dominantly inherited axonopathy.[4]

There is usually no disturbance of autonomic function. Fixed pupils, optic atrophy, nystagmus, endocrinopathies, epilepsy and spina bifida, which have been reported occasionally in association with peroneal muscular atrophy, probably represent coincidental congenital disorders. The disease at present is incurable.

Orthopedic intervention (arthrodeses of the ankle in severe foot-drop) or devices (e.g., special shoes or fitments) may help a patient to walk with ease. The index patient is a case of CMT disease type 2/CMT2 with late-onset and slow progression. The presence of characteristic features of CMT disease with Marfanoid features, a coincidental congenital disorder makes this case a clinically interesting and rare entity.

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