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CASE REPORT

Charcot Marie Tooth Disease Type 1 - Rare but Commonest Hereditary Neuropathy

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Abstract: *Objective:* To present a case of Charcot Marie Tooth Disease. *Backgrounds:* A 22 years old boy presented with very slowly progressive symmetrical weakness of both lower limbs with distal muscular atrophy. He has a family member with similar problem. *Methods:* The patient was clinically examined and investigations done accordingly like Nerve Conduction Velocity studies (NCV). *Results:* The patient was diagnosed to be a case of Charcot Marie Tooth disease type 1 with classical features like" inverted Champagne bottle" like legs. *Conclusion:* Charcot Marie Tooth disease is rare and often undiagnosed yet it is the commonest hereditary neuropathy worldwide and India is not an exception.

Introduction

Charcot Marie Tooth (CMT) neuropathy comprises of a heterogeneous group of inherited peripheral nerve disorders. Overall, the worldwide incidence is 10 in 100000 [1]. An widely accepted classification pattern is Charcot Marie Tooth type 1(demyelinating form), type 2 (Axonal degeneration), Type 3 (Dejerine Sotta disease), Type 4, few X linked varieties and Hereditary Neuropathy with Pressure Palsies (HNPP). An alternate classification system is Hereditary Sensorimotor Neuropathies (HSMN), HSMN I (CMT -1), HSMN II (CMT -2), HSMN III (CMT iii) and HSMN IV (Refsum's Disease). CMT 1 can further be subdivided to 1A, 1B, 1C, 1D on the basis of chromosomal studies.

Case History

A 22 years old male presented with difficulty in running, jumping and recurrent tripping since childhood and difficulty in holding objects with his right hand.His walking was normal. There was no history of speech problem, difficulty in swallowing, hand tremor, hearing problems or any autonomic disturbances. His uncle had similar problems. He had no significant past history.

On examination his vitals were stable. Neurological examination showed bilateral symmetrical wasting and weakness of the Anterior Tibial, peroneii, calf muscles and the intrinsic muscles of the foot. The wasting of the lower limb muscles stopped abruptly at the level of lower third of the thigh both sides giving an "Inverted

Champagne bottle" like appearance (Figure 1 &2), pes cavus and hammer toes on both sides, varus deformity of right foot (Figure 2), loss of knee and ankle jerks bilaterally but plantar reflexes were flexor, his gait was high steppage variety. Also there was clawing of all digits on the right hand, impairment of sensation of crude and fine touch on stocking distributions bilaterally. Cranial nerves including fundoscopic examination and cerebellar functions were normal.

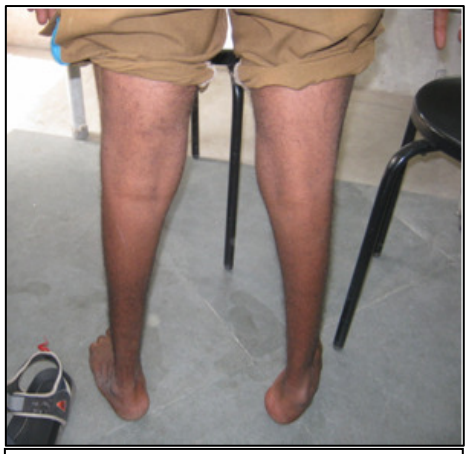

Figure-1: Charcot-Marie -Tooth disease from the back with the classical inverted "Champagne Bottle" appearance

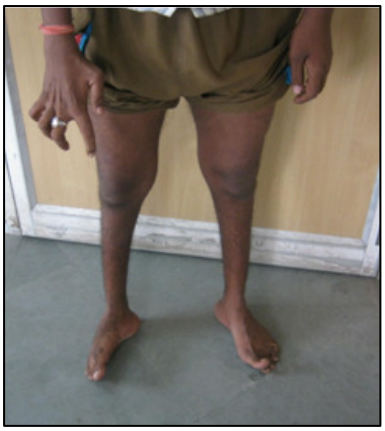

Figure 2: Charcot- Marie Tooth Disease from the with the classical 'inverted Champagne Bottle' appearance from the both mid thigh downwards , Complete right claw hand and varus deformity of the right foot

Investigations showed total White Blood cell count -9700/cuml with normal differential count , normal fasting , PP blood sugar, serum creatinine , Liver function test, sodium, potassium, Serum TSH ,Rheumatoid factor, freeT4 .Serum CPK/CPK-MB, LDH ,C reactive Protein, serum and urinary electrophoresis were normal, HIV 1 and II , HbsAg non reactive,. CSF analysis revealed, Total cell count -3 lymphocytes/ µl , glucose- 62 mg/dl, protein – 27 mg/dl , Chest Xray , ECG, USG whole abdomen, Echocardiography X ray lumbosacral and thoracic spine were normal. Nerve Conduction Velocity study of all limb showed demyelinating pattern with symmetrical, almost uniform slowing of all lower limb motor nerves to about 22 m/sec, no focal conduction block. Conduction velocity is approximately 25m/sec in forearm segment of median nerves. Sensory action potential was markedly diminished in both lower limbs with surface recordings. Our diagnosis was Charcot Marie Tooth disease type 1. Unfortunately patient did not afford Chromosomal analysis. The patient was sent to the Orthopaedics and Physical Medicine department for rehabilitative therapy.

Discussion

Charcot Marie Tooth disease (Hereditary Sensorimotor Neuropathy), usually an Autosomal dominat disease presents with very slowly progressive, distal symmetrical bilateral lower limb weakness with a strong family history and very little subjective sensory involvement. Basically any polyneuropathy advancing over 10 years is invariably genetic in origin [2]. Despite marked distal muscle involvement it is unlikely for them to lose ambulation. Our patient is ambulant till today since the onset of symptoms 6 years back. In CMT1A, the commonest subtype, there is duplication of the Peripheral Myelin Protein gene 22 (PMP-22) [3]. In our patient of CMT 1 progressive weakness of Tibialis Anterior led to Pes cavus, long toe extensors attempted to make up for the foot dorsiflexors like Tibialis Anterior causing hammer toes [4]. Tendon reflexes are usually lost early at ankle and later at the knee. Both the Vibration and big toe joint position senses are usually diminished. In our patient all of the above features were present. On NCV study motor and sensory conduction velocity are usually less than 60% of normal values in infant .Patients older than three years have conduction velocity 38m/sec in all peripheral nerves [4].Though family history is crucial for the diagnosis but the absence of family history may be due to Autosomal recessive inheritance and sometimes because of very few blood relatives or early death of a case before telltale signs could develop[5] .In our country absent family history is mostly due to improper information and poor education. Some patients are susceptible to complications during pregnancy, delivery [6] and neurotoxic damage by Vincristine if suffers from a malignancy in future.

Chronic Inflammatory Demyelinating Polyneuropathy, a close differential diagnosis can be ruled out by very slowly progressive history, normal CSF study, no conduction block in Nerve Conduction Velocity Studies (NCV) studies. Infact conduction block in NCV study is a certain sign of acquired demyelinating process [7]. Hereditary Neuropathy with Pressure Palsy (HNPP) can be ruled out by history of having no entrapment neuropathy in the past but chromosomal analysis is a must to know the subcategory.

Unfotunately treatment of this disease is limited. Treatment is mostly supportive. Early in the course of the disease strengthening exercise of feet with active feet stretching may be beneficial. Using ankle foot orthosis to stabilize the feet, correction of the foot drop may help reduce likelihood of falls.

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