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Charcot Marie Tooth Disease-A Case Report Name Of Institute :Sbks Medical Institute And Research Centre

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I. INTRODUCTION

Charcot -Marie-Tooth disease is most common genetic determined neuropathy and has overall prevalence of 3.8/100,000 population. It is transmitted as autosomal dominant trait with 83% expression,the 17p11.2 locus is the site of abnormal gene.Thegene product is peripheral protein 22(PMP22) .It is a hereditary motor and sensory neuropathy of the peripheral nervous system characterised by progressive loss of muscle tissue and touch sensation across various parts of the body. The typical clinical presentation includes distal muscle weakness and atrophy, but the severity and progression are largely variable. Improvements in supportive treatment have led to better preservation of patients' motor functions. More than 80 genes have been associated with CMT. These genetic discoveries, along with the developments of cellular and transgenic disease models, have allowed clinicians to better understand the disease mechanisms, which should lead to more specific treatments.

II. CASE REPORT

A 8year old male child was admitted in paediatric ward of Dhiraj hospital hadpresented withcomplain of difficulty in standing and walking with generalised weakness in all four limbs with lower limbs more and than upper limbs.On examination after admission it was noted that patient had following features: Upper limb - Bilateral Clawing withWrist drop present, Lower limb,complex foot deformity with

Left foot - Talipes equinus
Talipes calcaneovarous
Right foot - Talipes cavovarus

Along with bilateral foot drop.Gait deformity with high stepping gait more in left lower limb.
On Complete blood count-Hb-13.4, total count-9800, differential count neutrophil 56%,lymphocyte-39%, eosinophil 3%, monocyte-2%, basophil-0% .
Electrolytes -Na -140,K-3.5,Cl-101,Ca-8.1
Urea -35,creatinine-0.4,Bilirubin :Total -0.3,direct-0.2,indirect-0.1,SGPT-30,SGOT-16,CPK(Total)-116,
On DNA Test -Charcot-Marie-Tooth disease
On nerve conduction-study conclusion is sever Axonal ,sensori-motor neuropathy involving bilateral upper and lower limbs

III. DISCUSSION

Charcot-Marie-Tooth disease (CMT) is one of a group of disorders that cause damage to the peripheral nerves. Progressive muscle weakness typically becomes noticeable in adolescence or early adulthood, but the onset of disease can occur at any age. Because longer nerves are affected first, symptoms usually begin in the feet and lower legs and then can affect the fingers, hands, and arms.There is currently no cure for CMT but it can be managed with supportive therapy. CMT isn't usually life-threatening and rarely affects muscles involved in vital functions like breathing. People with most forms of CMT have a normal life expectancy.[1]

Typical early features include weakness or paralysis of the foot and lower leg muscles, which can cause difficulty lifting the foot (foot drop) and a high-stepped gait with frequent tripping or falling. Individuals also may notice balance problems. Foot deformities, such as high arches and curled toes (hammertoes), are also common in CMT. The lower legs may take on an "inverted champagne bottle" shape due to the loss of muscle bulk. As the disease progresses, weakness and atrophy may occur in the hands, causing difficulty with fine motor skills. Degeneration of sensory nerve axons may result in a reduced ability to feel heat, cold, and touch. The senses of vibration and position (proprioception) are often decreased in individuals with CMT. The disease also can cause curvature of the spine (scoliosis) and hip displacement. Many people with CMT develop

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contractures—chronic shortening of muscles or tendons around joints, which prevents the joints from moving freely. Muscle cramping is common. Nerve pain can range from mild to severe, and some individuals may need to rely on foot or leg braces or other orthopedic devices to maintain mobility. Some people with CMT experience tremor, and vision and hearing can also be affected.In rare cases, breathing difficulties may occur if the nerves that control the muscles of the diaphragm are affected.[1]

The gene mutations in CMT are inherited in three distinct patterns: autosomal dominant, autosomal recessive, and X-linked, all of which are tied to a person's chromosomes.

Different types of cmt.

- CMT1 is caused by abnormalities in the myelin sheath. The autosomal dominant disorder has six main subtypes.
- CMT1A results from a duplication of the gene on chromosome 17 that carries the instructions for producing the peripheral myelin protein-22 (PMP22). Individuals experience weakness and atrophy of the muscles of the lower legs beginning in childhood; later they experience hand weakness, sensory loss, and foot and leg problems.
- CMT1B is caused by mutations in the gene that carries the instructions for manufacturing the myelin protein zero (MPZ, also called P0), which is another critical component of the myelin sheath. Most of these mutations are point mutations.Other less common causes of CMT1 result from mutations within the SIMPLE (also called LITAF), EGR2, PMP22, and NEFL genes, respectively.

- CMT2 results from abnormalities in the axon of the peripheral nerve cell, rather than the myelin sheath, and is less common than CMT1,people with CMT2 often have less disability and sensory loss than individuals with CMT1. The onset of CMT2 is usually in childhood or adolescence. Some types of CMT2 may have vocal cord or phrenic nerve involvement, causing speech or breathing problems.

- CMT3, or Dejerine-Sottas disease, is a particularly severe demyelinating neuropathy that begins in infancy. Infants have severe muscle atrophy, weakness, delayed motor skills development, and sensory problems. Symptoms may progress to severe disability, loss of sensation, and curvature of the spine.
- CMT4 comprises several different subtypes of demyelinating and axonal and motor neuropathies that are inherited autosomal recessively.

- CMTX1 (also called CMT X, Type 1) is the second most common form of CMT. This X-linked disease is caused by mutations in a gene that provides instructions for making the protein connexin-32. The Males who inherit the mutated gene show moderate to severe symptoms of the disease beginning in late childhood or adolescence. Females who inherit a mutated gene often develop milder symptoms than males or do not show symptoms.[7][8][9]

Stabilisation of the ankles is a primary concern.In early stages stiff boots that extend to the mid calf are prescribed later when ankle weakens further,light weight plastic spiltsextending beneath the foot and around the back of the ankle can be custom made.External short leg braces may be required when foot drop becomes complete.In advance cases ofcompression neuropathy during sleep may be prevented by placing soft pillow beneath or between the lower legs,burningparasthesia not common but often are abolished byphenytoin,carbamazepine or gabapantin.Surgical fusion of ankle may be considered in some cases.Progressive resistance excersise for foot dorsiflexion may attenuate the progression of weakness[2][3][4].

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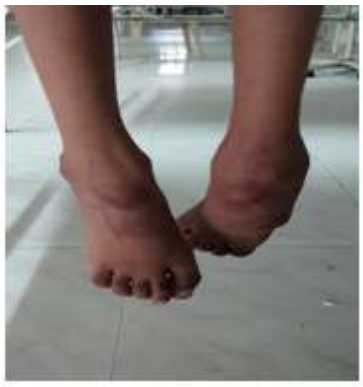

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