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In Syndrome

Charcot Marie Tooth Disease

Definition:

- It is the commonest inherited neuromuscular disorder.
- It affects the peripheral nerves and leads to progressive weakness of extremities.
- Occasionally it involves cranial nerves, other sites of the neuraxis, and other organ systems.

History:

This entity was first described in 1886 by Jean Marie Charcot and Pierre Marie from France and Howard Henry Tooth from England.

Etiology:

- Over 80% to 90% of the genetic abnormalities are due to copy number variation in PMP22 and mutations in GJB1, MPZ, and MFN2 genes.
- Copy number variation in PMP22 is the commonest cause of CMT.
- PMP22 is a large gene that is located in the middle of the 1.4 Mb regions in chromosome17p.12.
- This region is susceptible to frequent genomic rearrangements.

Epidemiology: 1 in 2500 people.

Classification:

- CMTs were initially named as hereditary motor and sensory neuropathies (HMSN) and classified as
- HMSN I: demyelinating subtype
- HMSN II: axonal subtype
- HMSN III: Dejerine Sottas disease
- HMSN IV: Refsum disease
- HMSN V: with pyramidal signs
- HMSN VI: with optic atrophy
- HMSN VII: with retinitis pigmentosa

Clinical Features:

- Distal symmetrical weakness, wasting, hypo/areflexia, skeletal deformities. These are more pronounced in lower limbs as compared to upper limbs.
- Commonly reported symptoms are difficulty in walking fast or running, tripping, falls, and twisting or spraining of ankles.
- Patients may have delayed motor milestones.
- During childhood, these patients are clumsy and 'slow' in sports, and not athletic.
- As the weakness progresses, patients develop foot drop and high-stepping gait. The weakness of hands manifests as difficulty in buttoning, zipping, and writing..
- Pes cavus, hammertoes and clawed hands in patients with long-standing disease due to the weakness of intrinsic muscles.
- Wasting of legs and distal thighs may seem like an inverted champagne bottle.
- Spinal deformities (scoliosis) may also occur.

Diagnosis :

- Electrophysiology
- Nerve Imaging
- Genetics
- Nerve Biopsy
- Evaluation of Systemic Involvement

Treatment :

- Treatment is mostly rehabilitative and symptomatic since there is no definite and effective disease-modifying treatment.

Prognosis :

- Life expectancy is usually not affected, but the disease may be severe when the onset is very early.

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