FOUR CASES WITH DIFFERENT TYPES OF CHARCOT MARIE TOOH AXONAL INVOLVEMENT

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Hereditary neuropathies are a disease group having heterogeneity that progresses slowly in the peripheral nervous system with demyelination and/or axonal loss. Charcot-Marie-Tooth (CMT) disease is also known as peroneal muscular atrophy or hereditary motor sensory neuropathy. Demyelinating type (CMT Type 1) and axonal type (CMT Type 2) are the two primary clinical phenotypes. Despite the significant developments in the genetic field in recent years, the diagnosis of a major part of axonal type CMT is still not made by a genetic mutation.

OBJECTIVE

The objective is to discuss the four CMT cases with rare axonal involvement presenting with gait disturbance with their similarities, differences, and genetic results.

CASES

This case report includes four patients presenting with gait disturbance, who were diagnosed within the past year. The patients' age, gender, history, family history, clinical findings, central imaging features, and electrophysiological and genetic findings are shown in detail in Figures 1 and 2 and in Tables 1, 2, and 3.

Table 1. Medical history

Case 1	Full term, C/S, 4,000 gr - Flexion posture in the right hand at birth
Case 2	Full term, NSVD, 1,500 gr - Toe walking and strabismus when she started to walk
Case 3	Full term, NSVD, 4,000 gr - Pressing the left foot on the side when she started to walk
Case 4	Full term, NSVD, 4,000 gr - Deformity in both feet at birth

Table 2. Electrophysiological* and MRI findings**

Case 1	*Sensorimotor polyneuropathy consistent with axonal degeneration **Normal brain + spinal MRI findings
Case 2	*Severe sensorimotor polyneuropathy with axonal degeneration and demyelination **Normal brain + spinal MRI findings
Case 3	*Normal **Brain MRI wide bilateral ventricles in the supratentorial area Lumbar MRI normal
Case 4	*Normal **Brain MRI temporal arachnoid cyst, ventricular asymmetry Spinal MRI normal

Table 3. Clinical and physical examination findings

Case 1	Normal mental findings, Diffuse hyperlaxity, high plate Thenar and hypothenar atrophy, hypoesthesia, and camptodactyly Chest wall deformity and scoliosis Muscle strength: 4/5 for upper extremity proximal and distal, 4/5 for lower extremity distal, and 3/5 for lower extremity proximal, atrophy in lower and upper extremity distal, bilateral absence of DTR, pes cavus deformity, rocker bottom feet, steppage gait
Case 2	Normal mental findings Diffuse hyperlaxity Small hands and feet, weak grip, diffuse hyperlaxity, thenar and hypothenar atrophy, and camptodactyly Muscle strength: 4/5 in all four extremities, atrophic distal lower extremity, bilateral absence of DTR, genu recurvatum, rocker bottom feet, prominent pes cavus deformity, and impaired gait pattern
Case 3	Normal mental findings Lower extremity: left leg 2 cm thinner than right Muscle strength: 4/5 for distal left lower extremity Left foot stepping in, left ankle spasticity, hypoesthesia of the left toes, and bilateral absence of DTR
Case 4	Normal mental findings Bifid uvula Muscle strength in distal upper and lower extremities: 4/5 Bilateral hypoactive DTR in lower extremities, rocker bottom feet, pes calcaneovalgus deformity, and impaired tandem gait

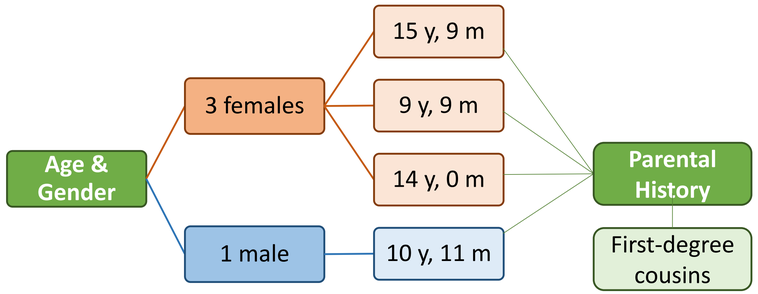

Figure 1. Age, Gender, and Parental History

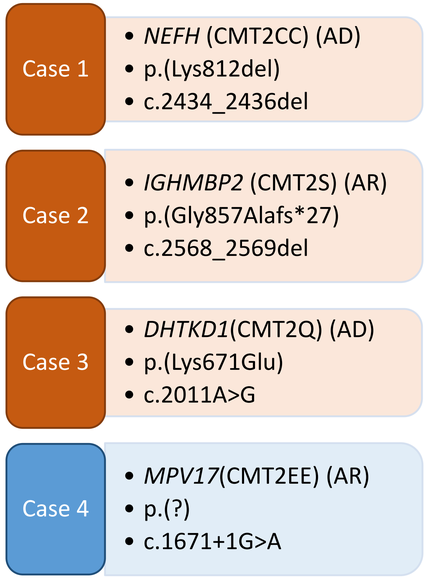

Figure 2. Genetic, Protein change, and Nucleotide change

CONCULSIONS

In conclusion, the subtypes of CMT2 are clinically similar, but the use of molecular genetic tests has made it easier to distinguish them in diagnosis. This study is presented to raise awareness that late diagnosed cases can actually be diagnosed even in the neonatal period with careful physical examination findings, to introduce 3 new mutations to the literature, to emphasize the differences as well as the similar features of the detected cases, and to facilitate the diagnosis of rare diseases.

REFERENCES

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