
A 12 Year Old Boy With Suspected Charcot-Marie-Tooth: A Case Report

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Abstract

Charcot-Marie-Tooth (CMT) is the most common hereditary neuropathy, with a prevalence of 1:2.500 in the Caucasian population. Clinical manifestations typically present during childhood or adolescence. A 12-year-old boy presented with difficulty walking. Alloanamnesis revealed that his growth and development had been appropriate for his age from birth until the age of 10 years. Over the past two years, he had developed gait abnormalities characterized by foot slapping while walking. Lower limb weakness had worsened seven days prior to hospitalization. The patient frequently fell, fatigued easily, and experienced pain in both legs. These symptoms were not preceded by fever or cough. Bowel and bladder functions were normal. He was born full-term, he had complete immunizations and no significant past medical history. His uncle had experienced similar symptoms. Physical examination revealed pes cavus and hammer toes. Neurological examination revealed lower limb muscle strength of 2/5, decreased patellar tendon reflexes, and negative pathological reflexes. Sensory function was difficult to evaluate. ENMG was not performed because the patient's parents refused referral. The patient showed clinical improvement following symptomatic pharmacological therapy and physical exercise therapy. The patient's clinical manifestations were consistent with typical features of CMT, including distal weakness, foot deformities, and loss of reflexes. Genetic testing, electrophysiological examination, and nerve biopsy could not be performed due to limited facilities. The clinician's role is crucial in establishing an initial diagnosis based on anamnesis and physical examination. Early detection may facilitate timely rehabilitation interventions to improve the patient's quality of life.

Keywords: Hereditary Neuropathy, Charcot-Marie-Tooth, Genetics

INTRODUCTION

Charcot-Marie-Tooth (CMT) or Hereditary Motor and Sensory Neuropathy (HMSN) is a progressive hereditary neurological disorder characterized by demyelination and/or axonal degeneration. The prevalence of CMT is estimated to be approximately 1:2500 in the Caucasian population (Carine et al., 2016).

CMT is characterized by progressive muscle weakness, muscle atrophy, and sensory disturbances. The clinical manifestations of this disease typically emerge during childhood or adolescence but may also present in adulthood. Early symptoms commonly include gait abnormalities, such as frequent tripping due to weakness of the distal muscles of the lower extremities. Patients with foot drop often exhibit a high-stepping gait, characterized by excessive knee elevation to allow the sole of the foot to clear the ground (steppage gait) (Stavrou et al., 2021).

The diagnosis of CMT is established based on history taking, physical examination, nerve conduction studies, and genetic analysis. However, limited facilities often pose a challenge. This case report aims to emphasize the importance of clinical awareness regarding CMT in children presenting with progressive weakness.

RESEARCH METHODS

Reported a 12-year-old boy presenting with a complaint of difficulty walking. Based on heteroanamnesis (alloanamnesis) with the patient's mother, it was found that from birth up to 10 years of age, the patient's growth and development were appropriate for his age

milestones. Over the last two years, changes in gait have been observed, such as stamping his feet when taking steps.

Lower extremity weakness worsened since 7 days prior to admission. The patient frequently tripped/fell, fatigued easily, and experienced pain in both lower extremities. The complaints were not preceded by fever, cough, or common cold. Bowel and bladder habits were within normal limits. The patient had a history of being born full-term with a birth weight of 4,300 grams. Immunization history was complete. Past medical history was denied. The patient's uncle experienced similar complaints.

Physical examination revealed a weak general condition, *compos mentis*, heart rate 118 bpm, respiratory rate 22 breaths/minute, temperature 36.4°C, and oxygen saturation 98%. On inspection of both lower extremities, deformities in the form of *pes cavus* and *hammer toes* were observed (**Figures 1 and 2**). Neurological examination showed that the muscle strength of both right and left lower extremities was decreased with a grade of 2/5. Patellar tendon reflexes were decreased, pathological reflexes were negative, and sensory function was difficult to evaluate.

Routine blood tests showed results within normal limits, including: hemoglobin 13.2 g/dL (14-18 g/dL), hematocrit 39.4% (42-52%), MCH 25.9 pg (27-31 pg), MCHC 33.5 g/dL (33-37 g/dL), MCV 77.2 fL (79-99 fL), and MPV 7.8 fL (9-13 fL). Plain lumbosacral radiography showed no abnormalities (**Figure 3**).



Figure 1. Pes Cavus

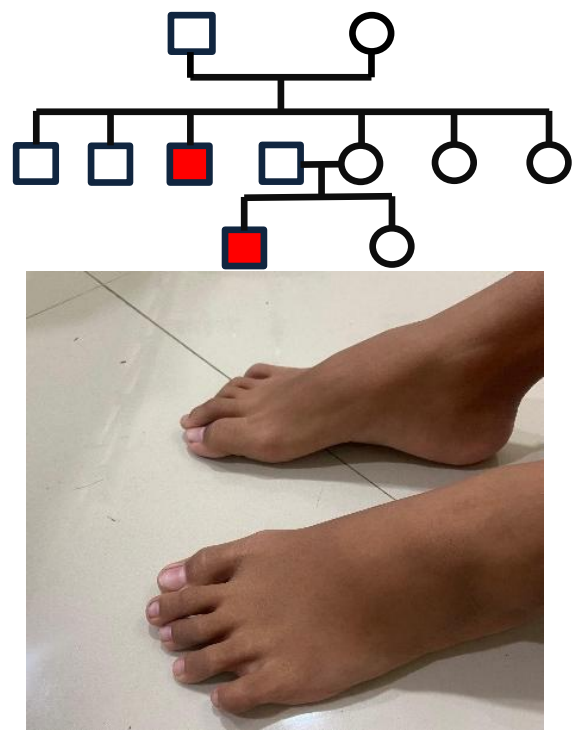


Figure 2. Hammer toes

Figure 3. Patient's family tree. There is one family member, specifically a maternal uncle, with similar symptoms



Figure 3. Lumbosakral X-Ray

An ENMG examination was not performed because the patient's parents declined the referral. However, clinical improvement was achieved through physical exercise therapy and symptomatic pharmacological treatment.

RESULTS AND DISCUSSION

The prevalence of Charcot-Marie-Tooth (CMT) disease is estimated at 1:2,500 among Caucasian population. As the most common hereditary neuropathy worldwide, it has an incidence of 1–3 cases per 100,000 individuals. To date, epidemiological data regarding CMT in Indonesia remain unavailable.

CMT encompasses a spectrum of neuropathic disorders caused by genetic mutations. Based on inheritance patterns and pathological characteristics, CMT is classified into several types: type 1 (CMT1, demyelinating), type 2 (CMT2, axonal), type 4 (CMT4, autosomal recessive), X-linked (CMTX), and dominant-intermediate (DI-CMT) (Ramchandren, 2017).

The patient's clinical manifestations are consistent with the classic presentation of Charcot Marie Tooth (CMT), namely distal weakness, foot deformities, and loss of reflexes. Ancillary investigations such as genetic analysis, ENMG examination, and nerve biopsy could not be performed due to limited facilities at a type D hospital. Therefore, the role of the clinician is highly crucial in establishing an initial diagnosis through history taking and physical examination. Early detection can accelerate rehabilitation interventions to improve the patient's quality of life.

Peripheral neuropathy in CMT can be inherited in an autosomal recessive, X-linked manner, or result from mitochondrial DNA mutations (Parmar et al., 2024). Relevant pedigree information includes premature death in the extended family and a history of undiagnosed walking difficulties. In this patient, a maternal uncle experienced similar symptoms (**Figure 3**), strengthening the suspicion of a hereditary neuropathy.

The diagnostic approach for CMT necessitates evaluating clinical evidence consistent with a genetic disorder, such as family history, age of onset, symptom progression, and developmental history. In this case, the presence of similar clinical manifestations in a family member provides substantial support for a genetic etiology. Additional diagnostic clues include a slowly progressive course over several years and an onset within the first decade of life, aligning with the classic presentation of CMT. The initial symptoms appeared at age 10 as gait disturbances, specifically foot stamping during ambulation. The patient also exhibited a steppage gait, characterized by high knee-lifting to facilitate foot placement. These clinical features are highly consistent with the known characteristics of CMT (Dai et al., 2025).

On physical examination, patients with hereditary motor and sensory neuropathy or CMT disease, typically present with progressive distal muscle weakness, including heavy legs, delayed ambulation, or loss of previously intact walking ability. Sensory disturbances, such as paresthesia and hypesthesia, foot deformities, and diminished or absent deep tendon reflexes are also common. In this patient, physical examination revealed pes cavus and hammer toes (**Figures 1 and 2**). Neurological evaluation demonstrated a bilateral reduction in lower extremity muscle strength, graded at 2/5 on the Medical Research Council (MRC) scale. Bilateral patellar tendon reflexes were diminished, pathological reflexes were absent, and sensory function was difficult to evaluate.

The patient achieved clinical improvement following pharmacological therapy tailored to symptomatic management. This regimen consisted of intravenous methylprednisolone (20 mg tid), paracetamol (300 mg tid), mecobalamin (500 mcg bid), and oral vitamin D (1000 IU qd), followed by physical therapy.

Management of hereditary neuropathy is primarily supportive and aimed at symptom relief. Available modalities include physiotherapy, occupational therapy, orthotics, and assistive devices to manage musculoskeletal dysfunction. Physical therapy regimens for CMT should emphasize muscle stretching to prevent contractures and mitigate muscle

strength decline. Comprehensive rehabilitation plays a pivotal role in preventing complications and enhancing the quality of life in CMT patients (Rudnik et al., 2020).

CONCLUSION

Charcot-Marie-Tooth disease should be included in the differential diagnosis of peripheral neuropathy in pediatric patients presenting with progressive lower extremity weakness. This case report underscores the importance of clinical awareness, particularly in resources-limited healthcare settings.

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