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Case Report

Sensory Impairment Related Upper Extremity Fine Motor Delay of an Eight Years Old Charcot-Marie-Tooth Type 1E Patient - A Case Report

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Abstract

Charcot-Marie-Tooth type 1E (CMT 1E) is a demyelinating neuropathy resulted from PMP22 gene point mutations, accompanied often with deafness. The aim of this case study is to offer a clear clinical picture for CMT 1E by reporting a case of a young patient, in order to assist clinicians with accurate and effective rehabilitation management so as to improving patient's quality of life. The patient of this study was an 8-year-old male who has been diagnosed with CMT 1E at 18 months with a history of both fine and gross motor development delay. The patient has been treated with both occupational and physical therapy simultaneously. Postural observation, neuromuscular examination, Manual Muscle Testing and Beery Visual Motor Integration were employed to estimate his functional impairment. Physical therapy resulted in fairly strong of lower extremity strength at the hip, knee and ankle joints. Abnormal gait pattern was found and identified as walking on heels with no toe: off. Balance deficits observed for posture and locomotion related with poor postural strength and endurance. Asymmetry occurred when hip rotated internally. Occupational therapy suggested weakness in motor coordination led to age inappropriateness in visual motor integration. Fatigue was experienced while the patient was commencing tasks of upper extremity coordination. The severe upper extremity fine motor control development delay is observed with peripheral sensory impairment.

Keywords: *Charcot-Marie-Tooth Type 1E; Postural Balance; Motor Function*

Abbreviations

DCD: Developmental Coordination Disorder; MC: Motor Coordination; VP: Visual Perception; VMI: Visual Motor Integration; OTE: Occupational Therapy Evaluation; CMT: Charcot - Marie - Tooth; CMT1 (1A, 1E, 1X) CMT2 (2A, 2P): Subtypes of Charcot - Marie - Tooth 1 and 2; GMCS: Gross Motor Control Skills; FMCS: Fine Motor Control Skills; PTE: Physical Therapy Evaluation; PO: Postural Observation; NE: Neuromuscular Examination; LE: Lower Extremity; MMT: Manual Muscle Testing; FIM: Functional Independence Measure Scoring

Introduction

Charcot: Marie: Tooth (CMT) disease is a genetically progressive degenerative disease, affecting peripheral nerves gravely. It also known as hereditary motor and sensory neuropathy [1]. which was found and identified by three physicians (Jean Martin Charcot, Pierre Marie and Howard Henry Tooth) in 1886 [2]. Gene mutation is the primary factor causing CMT with various clinical manifestations [1] that mostly are classified into two subtypes: CMT1 and CMT2.

Clinical presentation of CMT1 (demyelinating pattern of denervation, autosomal dominant or X: linked inheritance) are decreasing nerve conduction velocity (less than 60% of normal values), muscle weakness, sensory loss, and peripheral nerve trunks may be enlarged [3]. Based on genetic manifestation, CMT1 has been further divided into seven subtypes: CMT 1 to 1X. Symptoms of CMT2 (axonal pat-

tern of denervation, autosomal dominant or recessive inheritance), has been described as normal or slightly reduced nerve conduction velocities, preserved stretch reflexed except ankle jerks, and no nerve hypertrophy [3]. Seven subtypes of CMT2 (CMT2A to 2P) have been distinguished as well [28]. CMT1A is the dominant type among CMT cases [4] conducted numerously by researchers with specific presentation [5-7]. However, few information obtained about other types, especially CMT 1E, which is the clinic diagnosis subtype of the patient in this study. CMT 1E, with less than 5% of all CMT1, about 6000 in the US, is defined by demyelinating neuropathy resulted from PMP22 gene point mutations, accompanied with deafness [8]. Performances of patients with CMT 1E have been reported in only one study, that we can find, suggesting progressive gait instability, frequent fall and limitation in running in their childhood [9]. Therefore, no explicit characteristics for CMT 1E are determined due to its rarity, and more reports needed to document the symptoms of this subtype.

Onset of CMT appear in early childhood, but mild signs are able to be identified [10]. According to this, recently a few studies have unearthed the features of children with CMT, for gross motor control skills (GMCS) of lower extremities, which is defined as activating coordinated large muscle groups for locomotion involving balancing, walking, running, riding a bike or catching a ball, etc. [11]. Melissa., *et al*. reported poor balance due to ankle instability effected by muscles for inversion and eversion in most children with CMT [12]. Pagliano., *et al*. have detected involved dorsiflexior producing abnormality of walking and imbalance in majority of children with CMT1A [5]. For upper extremities, on the contrary, scarce findings have been reserved. Two reports [5,13] have noted children of CMT1A presented obstacle in hand works with attenuation of grip strength and sensory dysfunction, respectively. These symptoms are implicated to fine motor control skills (FMCS), which is considered as precise and refine movement integration by using hands and fingers [11] relying significantly upon simultaneous development of GMCS. Unfortunately, FMCS about CMT group has been barely explored, either with adults or children. We invested the effects of CMT 1E on FMCS in this report.

In addition, besides muscle contracture, anesthesia that considered as the most apparent characteristics, there are other clinical manifestation of CMT, such as frequent fatigue [14], hearing loss [15] and led patients of CMT with reductive quality of daily life.

The purpose of this study is to attempt to offer a clear clinical picture for CMT1E with FMCS in terms of reporting an 8: year patient with CMT 1E subtypes, in order to help clinicians, prescribe for rehabilitation treatment correctly so as to improving quality of life.

Subject

The patient of this study is an 8-year-old male with a diagnosis of Charcot Marie Tooth Disease type 1E, which was diagnosed at 18 months following a muscle biopsy. He also has a history of fine and gross motor delay. He was born at 32 weeks by C: section after his mother's water broke at 29 weeks. He weighted 3lbs 13oz at birth. He wears glasses because of a lazy eye history. Hearing loss occurred occasionally. Educational services, occupational and physical therapy are received at school. There are a series of assistance devices including bilateral hinged Ankle Foot Orthosis, a bath chair with back only, crocodile walker, amtryke, wheelchair used for him to attempt to obtain a normal functional behavior at school and home.

Physical Therapy Evaluation (PTE)

In accordance with his PTE, it showed his gross motor delay throughout three measurements: postural observation (PO), neuromuscular examination (NE), and musculoskeletal exam. Firstly, from the results of PO, it has been reported he was incapable to stand without a wide base of support, described specifically as bilateral knee recuvatum with forward lean of the trunk and thus increased postural sway. For sitting, he preferred a W: position for a long time due to reduction of muscle tone throughout trunk and bilateral upper extremity. Besides, asymmetry of lower extremity (LE) was noted as L > R when there was hip internal rotation and in: toeing position. Secondly, it was illustrated by NE there was chronic sensory impairment in bilateral LE from ankles down through the toes following light touch sensation test (patient closed eyes and tell therapist when he felt a pen point touch the skin). More over a new symptom of hands deprivation noted. Table 1. Presents the last and the most important musculoskeletal exam, Manual Muscle Testing (MMT), which is designed for evaluation of muscle strength as well as gross motor skills (Table 1 and 2).

	Function of the Muscle	Grade		
No Movement	No contractions felt in the muscle	0	0	Zero
	Tendon becomes prominent or feeble contraction felt in the muscle, but no visible movement of the part	T	1	Trace
Test Movement	**Movement in Horizontal Plane**			
	Moves through partial range of motion	1	2-	Poor-
	Moves through complete range of motion	2	2	Poor
	Antigravity Position			
	Moves through partial range of motion	3	2+	
Test Position	Gradual release from test position	4	3-	Fair-
	Holds test position (no added pressure)	5	3	Fair
	Holds test position against slight pressure	6	3+	Fair+
	Holds test position against slight to moderate pressure	7	4-	Good-
	Holds test position against moderate pressure	8	4	Good
	Holds test position against moderate to strong pressure	9	4+	Good+
	Holds test position against strong pressure	10	5	Normal

Table 1: *Manual Muscle Testing Procedures and Key to Muscle Grading.*

		Left	Right
Hip	Flexion	4	4 +
	Extension	3 +	3 +
Knee	Flexion	5	5
	Extension	5	4
Ankle	Dorsiflexion	4 +	4
	Plantarflexion	4 -	3 +

Table 2: *Lower Extremity Strength Scores (MMT) (0-5, 5 as the strongest).*

Following the MMT detailed in tables 1 and 2, it was observed that this child has sustained good muscle strength in LE except weakness of hip and knee extension corresponding to walking and standing with bilateral knee recurvatum, forward leaning of the trunk noted in PTE. Knee recurvatum was connected with excessive range of knee extension while other range of motion of LE was within normal limited. In addition, abnormal gait pattern was found and identified as walking on his heels with no toe: off motion at end of gait sequence. He was able to ambulate on uneven surfaces with assistant but had sudden movements of trunk and LE. It was investigated he required maximal assistant (In accordance to Functional Independence Measure Scoring (FIM) [29] maximal assistant means patient can perform 25%:49% of tasks) for going upstairs and had weak eccentric quadriceps control to descend downstairs. Endurance was poor as well when he was sitting with postural pelvic tilt. Loss of balance happened in movements of squatting, and from sitting to standing. Balance deficits also influenced his handling skills of age appropriateness such as throwing, kicking, catching a ball.

Occupational Therapy Evaluation (OTE)

From his OTE, he was able to answer age appropriate questions and tolerated well participating in tabletop and strength, endurance testing. In line with this, Beery Visual Motor Integration (VMI) [30] was utilized for FMCS referred to in daily routine, which is a developmental sequence of geometric forms to be imitated or copied with paper and pencil. It is designed to apply for individuals aged 2:100, evaluating the coordination capability of visual perception and finger: hand movements. There are two subtests executed: the visual perception and motor coordination. The former focus on identifying parts of children's bodies and similar but slightly different shapes by employing minimal motor requirements, whereas the latter need to a strong visual aid for controlling pencil through tracing mazes without going outside of the lines (Table 3).

	Visual motor integration	Visual perception	Motor coordination
Raw score	15	24	12
Standard score (85-115)	74	106	53
Scaled score	5	11	1
percentile	4th%	65th%	0.1st%
Age Equivalent	5 years, 6 months	9 years, 8 months	3 years, 9 months

Table 3*: Subtests of the Beery VMI (age: 8 years, 5 months).*

As Beery VMI suggested, his standard score was lower than the standard score with 96% behind the healthy peers. Age inappropriateness was noted prominently. However, interesting results were found from supplemental tests. The score of visual perception (VP) was between 85 :115, and age equivalent (9 years, 8 months) was beyond his actual age. In contrast, compared to VP, there's a significantly lower score presented in motor coordination (MC). Consequently, MC was reckoned as the primary factor affecting VMI, while VP was not the most critical one.

Restriction of hand: finger corporation was demonstrated via tabletop tasks as the following performances: 1. A weak tripod grasp of right hand with dropping pencil easily and obstacle in picking it up again; 2. Consistently using left hand to stabilize paper and as a gross help; 3. Tough hand: writing as a result of decreased dexterity in both hands, reduction of thumb and finger flexion, and shrinking thumb: finger opposition; 4. Cutting basic shapes with standard scissors with fair accuracy but frequent fatigues without age equivalent of grasp strength (Left: 17, Right: 15) compared with normative data of male children for 8:9 years (Left: 39.0 ± 9.3, Right: 41.9 ± 7.4).

Upper extremity strength had been measured likewise, showing for elbow flexion and extension (4/5) was better than for bilateral shoulders flexion, abduction and adduction (3+/5), as well as for wrist flexion and extension (3+/5) on the basis of MMT. Nevertheless, sensation loss for both hands combined with poor core stability, were recognized as negatively impacting FMCS, occupational performance, and self: care activities including bathing with moderate assistance (patient can perform 50% to 74% of task from FIM), wearing braces, shoes and socks with maximal assistance, cleaning himself with bowel movements with minimal (patient can perform 75% or more of task from FIM) to moderate assistance, opening hardly containers with lunch at school, buttoning and unbuttoning with assist, and disability of using bathroom at school.

Although CMT disease affected him with difficulties in daily life, it has been noticed he remained positive and optimistic psychology throughout both OPE and PTE, which also mentioned he was enjoying greatly with horseback riding, coloring, train table, golf, iPad, and play in general.

Discussion

The patient is an 8: year: old boy with diagnosis of CMT 1E, ambulating with assistance devices with abnormal gait pattern: heel: walking without toe: off, correlated to decrease of bilateral ankle plater flexion and dorsiflexion in agreement with Romildo Don., *et al*. [16] who reported patients with plantar flexor failure were lack of impulse at push: off. There was more consistent phenotype presented by Vince., *et al*. [17] suggesting patients with CMT1 had hardly walking with toe: off because of poor range of planter flexion. As reported, however, the biggest problem for him was core instability, which were similar in previous study, displaying patients with CMT 1A showed muscle deficiency of lower extremities affected posture control abilities significantly, mostly with severe planter: flexor muscle weakness [18]. The patient of this study showed knee recurvatum in connection with impairment of LE and trunk muscle strength. From this point, wasting of hamstring and quadriceps may have been related to knee hyperextension, assumed by Newman., *et al.* [19] without evidence in MMT. It substantiated deficits of knee extensor, yet, in this case with MMT. Moreover, by verified in kinetic analysis they found the same re-

sult of hip extensor that was moderately weak as in this case, which delineated reduction of power for hip extension muscle [19]. Though most signs of GMCS for this patient were semblable with other types cases, one difference noticed contrasted with anterior research. Dissymmetry during hip internal rotation and toe: in position was noted for this patient, which was against the study that showing symmetrical foot alignment among children of CMT1A [20].

FMCS depend on contemporaneous GMCS development, as mentioned above. In line with this, this patient showed delayed FMCS due to deferred GMCS but with more grave deficits. Fine motor control at the hand has been seen as an important parameter for measure FMCS. It has been supposed muscle strength of upper extremity were not the main factor impacting him with difficulty of movements regarding to MMT. Worsen sensation for both hands and deficits of trunk balance were the indeed principal factors, which was not approved with the study illustrating more serious motor loss than sensory for patients with CMT [21]. However, they also evaluated muscle was the better parameter corresponding to dexterity than sensory function [21] which was disparate in this case, reveling sensory defect was the major proportion for reduction of dexterity. Moreover, loss of position sense was represented by w:sitting position due to lower muscle tone MT. Walter., *et al*. [22] provided clinical situation about one patient of 42 years old with CMT1 referring poor locomotion with reduced MT in all limbs and loss of sensation, but little is known between muscle tone and mobility for patients of CMT, except it only has been notified lesion of FMCS and integration correlated to motor performance more strongly than to MT among patients with developmental coordination disorder [23].

Hearing barrier has been presented in this case in accordance with one research emphasizing children with CMT 1 and CMT 2 both suffered deferred auditory ability from disease [15]. In addition, there was vision deficits: amblyopia noted in his medical history. Regardless he finished the visual perception subtest as a higher score, it might not be excluding the potential that amblyopia was related to FMCS dysfunction, which indeed explored in one study highlighting reduction of FMCS existed in amblyopic children compared to age: matched control group [24]. Fatigue present in patients with CMT, as measured by evaluators [14] and they were prone to explain there were poor GMCS and anesthesia linked to fatigue. However, in this case, the patient was experiencing fatigue when he was commencing fine motor tasks rather than gross motor movements. That might be regarded as the consequence of poor core stability and bilateral non-cooperation derived from hypotonia and decrease of muscle strength for upper extremity.

Despite CMT IE is the primary disease infecting the patient we studied, similar symptoms arise in other disorders as well. As literature showed, developmental coordination disorder (DCD) has been in correlation with CMT disease as well. DCD is defending by delay of motor skills development, and in trouble with integration performance for daily activities (dressing, handwriting etc.) [25]. In addition, sensory processing disorder, is also called dyspraxia, and has similar symptoms emerging in literature involving hardness with handwriting and other fine-motor skills (buttoning, tying shoes, holding a pencil etc., and barely playing ball games [26]. The extraordinary finding in this case was noticed that there was a close relevance between premature and deficits of motor control skill. Afend., *et al*. had affirmed 40%:60% preterm infants suffered moderately from no developing cerebral palsy and presented both gross and fine motor ability frequently [27].

Conclusion

In conclusion, with this special case, the child with CMT 1E showing impaired posture control and mobile ability effected by reduced muscle tone and mild decline of muscle strength for lower extremity. Poorer FMCS dysfunction has also been observed accompanied with upper extremity sensory deficit. However, despite it has been offered valuable signs about CMT1E in this case, further information needed to investigate, particularly with common characteristics of more patients with CMT1E, and correlation between FMCS and CMT.

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