Case Report

Neurology - Research & Surgery

Severe Sensitive Axonal Neuropathy and Oropharingeal Dysphagia

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Received: 24 July 2018; **Accepted:** 14 August 2018

Citation: Marta Linares-Gago, Marina López Ruiz SLP, Belén Romero Romero, et al. Severe Sensitive Axonal Neuropathy and Oropharingeal Dysphagia. Neurol Res Surg. 2018; 1(1): 1-3.

ABSTRACT

Hereditary motor and sensory neuropathies (HMSN), also known as Charcot-Marie-Tooth (CMT) disease, are the most common degenerative disorders of the peripheral nervous system. More than 80% of CMT patients in Western countries have genetic abnormalities associated.

CMT 2 typical clinical features include: distal weakness, muscular atrophy, sensory deficits, osteotendinous reflexes decrease and feet deformities. Symptom onset happens before 5 years old. Not being usual the swallowing disorders in its typical clinical presentation. We report an unusual clinical case where dysphagia and feeding problems are the main consequence of this condition.

Keywords

Charcot-Marie-Tooth disease, Sensory Neurophaty, Dysphagia, Feeding disorder.

Introduction

Hereditary motor and sensory neuropathies (HMSN), also known as Charcot-Marie-Tooth (CMT) diseases, are the most common degenerative disorders of the peripheral nervous system. More than 80% of CMT patients, in Western countries, have genetic abnormalities associated. They are suspected to be caused by a specific mutation in one of several myelin genes that results in defects in myelin structure, maintenance, and formation. HMSN are subdivided into 7 types, being 1 and 2 types the most common [1,2].

CMT 2 typical clinical features include: distal weakness, muscular atrophy, sensory deficits, osteotendinous reflexes decrease and feet deformities. Clinic evolution is similar to CMT 1, but sensory symptoms are prevalent. A severe sensory and slightly motor slowed conduction velocity are described in electrophysiologic studies [3].

In CMT 1, the symptoms onset typically is present in second or third decade. While, in CMT 2, the symptoms appearance is earlier, happening before 5 years old. Highlighting lower distal limbs weakness over sensory symptoms [2-5]. Not being usual the swallowing symptoms in its typical clinical presentation.

Clinical Case

It´s described a 10-year-old girl followed in our dysphagia unit, since 6 years ago, due to drooling and chewing problems according to a neuropathy under study. She had showed generalized hypotonia in birth, with initial sucking problems that led her to an early hospitalization according to a dehydration with 4 days of life.

First Clinical Assessment

- No cognitive problems
- Normal language development with slight inaccuracy articulation, but appropriate speech intelligibility.
- Mouth opened in rest with hypotonic lower lip in association to persistent drooling.
- Hypotonic tongue resting in mouth floor and interposed between both dental arcades in rest.
- Left oral mucosa and left tongue border sensory loss
- Slight weakness limbs
- Sensory distal limbs deficits

- Light handling problems
- Mild inestability gait

First Feeding evaluation

Great chewing difficulties with impossibility for bolus preparation. Adequate mash food swallowing (Figure 1).

First Instrumental swallow assessment (flexible naso-fiber-endoscopy evaluation of swallowing: FEES):

- Post-swallow pharyngeal residuous
- Complete cleaning residuous with multiple swallows
- Slight weakness tongue propulsion
- Right oral sensory deficit

First electromyography-electroneurography (EMG-ENG): Sensory Peripheral Neuropathy without typifying
First Genetic analysis: inconclusive

She was attending to intensive myofunctional therapy for speech and feeding/swallowing improvement with slight changes. The persistent masticatory disorder justified the performance of a new EMG-ENG study, of the muscles involved in this process. In this study, signs of a Sensitive Axonal Neuropathy compatible with the CMT IIB type, was described. It was associated with Severe Axonal Motor Neuropathy in masticatory muscles, especially in the pterygoid muscles without affectation of the masseters (Figure 2).

Figure 1: Chewing difficulties with impossibility for bolus preparation.

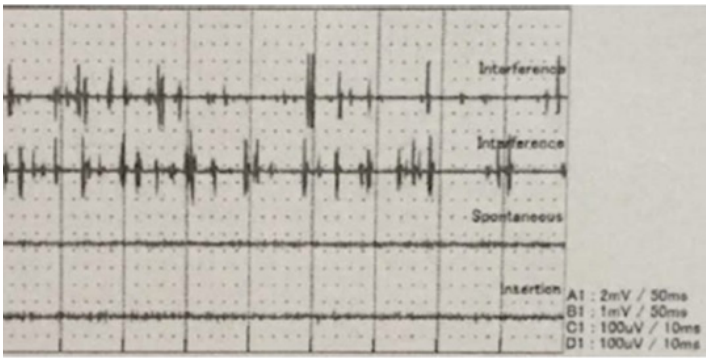

Figure 2: Electromyography-electroneurography.

After 6 years of therapy and following-up in our department, she doesn´t present any speech problem with a complete normal intelligibility. Rotatory masticatory function difficulties are still present, being cause of diet modifications. She carries out a pure vertical chewing, with secondary masseter hypertrophy and use of compensatory muscles. The weakness of the orbicularis of the lips and oral hypoesthesia persist and a light involuntary drooling due to this condition. Although, thin fluid intake is safe. So, currently, she has reached level 6 in the Functional Oral Intake Scale (FOIS), because she has only removed from her diet specific foods, regarding to their swallowing risks, as: hard meats, some fruits and sticky vegetables or cakes.

Nowadays, she walks independently, without balance disturbances. Though, she still shows slight fine motor skills difficulties due to distal limbs sensory loss. Because of this, she needs some help for specific complex task. Despite that, she is completely integrated in her environment without social limitations in the different life spheres.

Discussion

We describe an atypical CMT 2 clinical presentation, no published previously, where we observe its association to a severe chewing muscles sensory neuropathy that is the predominant symptom over classical CMT features [1-2].

Motor-sensory oral disorder, reported in this patient, can be the source of complications as development delay, malnutrition, dehydration, stomatognatic system pathology, and speech disorders. All these problems can be related to chewing muscles function deterioration, that control jaw movements in speech and feeding/swallowing function [6].

Our patient showed great masticatory difficulties what made bolus trituration imposible due to severe motor pterigoideus deterioration. These muscles are important in chewing and responsible of lateral movements during bolus preparation [7]. Because of that, she had difficulties in the introduction of solid foods in her diet as consequence of problems with bolus preparation. Chewing limitations has been associated to prolonged meals and drooling too. All these features led her slightly circumstances to social integration.

Feeding and swallowing diseases are common among neurological disorders, as cerebral palsy, brain injury…etc. Masticatory limitations bring on restrictive diets with a high risk of malnutrition. Besides, normal cognitive and nervous-inmunology-musculoskeletal system development is influenced by this situation, having been described the association among nutrition level, hospitalization/ medical consultation, and scholar days loss [8]. Actually, this condition could have important effects over adult health of the child [9-11] and social integration.

We describe an exceptional clinical case, where chewing muscles are severely affected in association of classical presentation in CMT 2. We didn´t find any previous reference in scientific literature, and no discussion about possible feeding/swallowing problems in these patients. Due to regular treatments are addressed to improve complaints related to limbs function, we want to highlight the possibility of clinical masticatory involvement with important effects in feeding and swallowing. It´s neccessary to confirm with future survey, the presentation degree of these features and in which CMT type is more affected.

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