

AUDITORY FUNCTION IN PATIENTS WITH CHARCOT-MARIE-TOOTH SYNDROME

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ABSTRACT

The Charcot-Marie-Tooth (CMT) disease is a neurological syndrome, which has seldom been associated with hearing dysfunction, particularly sudden sensorineural hearing loss (SNHL).

The disease of Charcot-Marie-Tooth is due to the alteration of one or more genes that give way to the formation of the axon and the genesis of the myelin, whose synthetic deficit involves the reduction of the speed of nervous management.

The aim of the study was to detect in individuals suffering from Charcot-Marie-Tooth syndrome with audiological signs and symptoms qualitative and quantitative type of hearing impairment and of these finds some assumptions on the location of the damage along the auditory nerve pathways.

Keywords: Charcot-Marie-Tooth syndrome, hearing loss, otofunctional evaluation, sound distortion.

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Introduction

The Charcot-Marie-Tooth (CMT) disease or hereditary motor and sensory neuropathy (HMSN) is a relatively common neurological syndrome, which has seldom been associated with hearing dysfunction, particularly sudden sensorineural hearing loss (SNHL). Families with autosomal dominant, autosomal recessive and X-linked forms of inheritance have been described.

It strikes one or more neuronal systems and can involve neurons of the central and peripheral motor system, so it can interest the faces of Goll and Burdach or the faces of spine cerebral⁽¹⁰⁾.

The onset may occur during late childhood or adolescence, may become progressive and severe or may spontaneously arrest at any time. Evolutionary trends overlapping was observed in other studies of childhood neurodegenerative diseases relating to immune character⁽¹⁷⁾.

Various conditions have been reported in association with CMT, including Friedreich's ataxia, myopathy, a combination of nephritis and sensorineural hearing loss and optic atrophy. Features of hereditary cerebellar ataxias, optic atrophy, and other cranial nerve involvement may occur in combination with the previously mentioned classic description.

In some cases, it is possible observe bilateral vocal fold paralysis, resulting in disorders of phonation, These changes are based on neurodegenerative disorders, resulting in very similar vocal demonstrations to those found in the larynx senile⁽¹⁴⁾.

Some aspects similar neurodegenerative can also be found in the epileptic disease, even if under the clinical profile diseases are widely different⁽⁶⁾.

From the anatomo-pathologic point of view microscopically signs of atrophy are observed with reduction of volume of long spinal lines and of

wide cerebral areas in correspondence to the stricken system.

Histologically you can observe atrophy of the nervous cells and primary degeneration of the fibers subject to the process of continuous demyelination distribution, the theca mieliniches reduced are inflated and fragmented into lumps. The protoplasmatic glia assumes the character of fibrous glia with consequent formation of glia scars responsible of the sclerotic hardening and wrinkling^(4,6).

Under the biochemical profile in which the pathology may be found the typical cellular alterations, oxidative stress, other neurodegenerative diseases and sometimes features similar to cellular aging^(2,3,11).

The disease of Charcot-Marie-Tooth is due to the alteration of one or more genes that give way to the formation of the axon and the genesis of the myelin, whose synthetic deficit involves the reduction of the speed of nervous management⁽¹⁾.

Two principal forms of the disease of Charcot-Marie-Tooth have been described:

- *Form demyelinated*: characterized by the proliferation of the cells of Schwann that are typically marked by the histological comparison of bulb formation of onion, the covering of the axon has been altered and the deficit myelinic initially induces a deceleration of the nervous management speed without clinical trouble but in the following phases the axon loses connection with the relative muscular fibers depriving it of some innervation making the apparent symptomatology;

- *Form axonale*: characterized from scarce demyelination without histological comparison of bulb formations of onion, the myelin is entire even if the axon introduces an alteration of its components with normal speed of nervous management⁽⁵⁾.

In the demyelination and in the axonale forms the nervous fibers struck are those longer which are destined to the muscles of the feet, of the legs, and of the hands but also the sensorial fibers run into degeneration and death. The disease of Charcot-Marie-Tooth is marked by a progressive distal neurogenic amyotrophy with ascending evolution, it initially strikes the muscles antero-external face of the leg with progressive steppage, without however to go up again above the third inferior of the thigh, while that of the hands is more late and it slowly goes up again to the forearms^(7,12).

The abolition of the achilles reflexes are precocious while those rotuleis are late, as generally its presence is found in foot cable and kyphoscoliosis.

In some forms there is also weakening of the respiratory muscles and of the phonation with compression of other organs among which the ear, phenomenon that can induce sensorial deafness.

In some cases the disease is also manifested with impaired auditory function, and specifically the component sensorineural retrocochlear^(9,15,16).

In some patients the audiological symptomatology is characterized by the presence of tinnitus in shades similar to that seen in expansive vascular disorders in cerebellar localization⁽¹³⁾.

The aim of the study was to detect in individuals suffering from Charcot-Marie-Tooth syndrome with audiological signs and symptoms qualitative and quantitative type of hearing impairment and of these finds some assumptions on the location of the damage along the auditory nerve pathways.

Materials and methods

The study has been effected on 3 subjects of female sex, affected by Charcot-Marie-Tooth disease, of age between 48 and 55 years, coming to our observation for the presence of bilateral hearing loss.

The procedural methodology was based on the execution of:

- anamnestic investigation for the collection of subjective data related to the presence of auditory deficit;

- otofunctional evaluation for the determination of what quantitative and qualitative characteristics of the auditory functionality through subjective and objective audiological tests:

- tonal audiometric examination for the survey of the auditory threshold;

- vocal audiometric examination, for the determination of the level of oral intelligibility. It consists of different phases characterized by methodic followings audiometric vocal examination, as: test of perception of quickly spoken altered or rasp, test of binaural fusion, test of spondaiche words divided out or SSW, test of the dichotic figures, test of the competitive sentences, test of models of frequency, test of the spoken at pass-low

- impedenzometric examination

- automatic audiometry

- auditory brainstem responses (ABR).

Results

The analysis of the results has underlined in all patients the presence of sensorineural hearing loss at the tonal audiometric examination.

In the tonal audiometric suprathreshold tests, the test of the threshold of decadence, has shown presence of bilateral adaptation, probably referable to phenomena of sonorous distortion in relationship to the time, sign of retrocochlear suffering⁽⁸⁾.

The study of impedance audiometry has noticed at the contralateral acoustic reflex threshold that the presence of normal threshold is reduced to low and averages frequencies and absent to the high frequencies.

The examination of the potential evoked auditory has brought in all cases the presence of layout not structured. As it regards the central acoustic evaluation and specifically in the test of fusion biauricular, all the subjects have had notable discriminative difficulties, with a score below the normal levels. Under conditions of competition in the test of words spaced out spondaic is noticed a serious bilateral deficit, as in the test of the dichotic figures.

In the test of the competitive sentences, the patients had shown a bilateral deficit that in the test to models of frequency, the seriously reached reduced scored, in the test of the spoken filtrate it passes-low, the oral understanding results particularly bilaterally compromised. It resulted in fact difficult for the patients to distinguish complexed acoustic stimuli and to listen under conditions of noise.

The battery of the peripheral test essentially showed a non remarkable degree of auditory deficit, while the test on ABR and center pointed out referable problems to the subjectivity symptomatology brought by the patients.

Conclusions

In cases observed in this study the audiologic and neurological research have clearly brought back the audiologic pathology to the disease of Charcot-Marie-Tooth, as so comparative research points out that this type of pathologies can implicate various sensorial dysfunctions and of the central nervous system.

In this disease there is probably a bilateral involvement of the auditory nerve even if the most serious deficit seems to be in the central auditory system.

In the patients observed a bilateral rollover of the function has been noticed PI-PB conforming to the lesion of the auditory nerve and or to the lesion of the encephalic trunk and sometimes to both.

The results of the tests of auditory decadence and of the automatic audiometry, were clearly indicative of central auditory lesions as shown by the absence of high frequencies of the thresholds of the acoustic reflex, that were present however in middle-inferior frequencies.

The tests of central acoustic evaluation have subsequently underlined the impairment of the decodificative abilities of the central nervous system and the involvement of the cortical function, element that at the light of the considerations brought it assumes meaning of centrality place of the lesions.

Then it can be concluded that in the patients neurological valued demonstration of degenerative type a retrocochlear level, of the eighth cranial nerve, of the encephalic trunk and of the cortical areas in all it seems that the auditory involvement is essentially imputable to central dysfunctions.

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