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AUDIOLOGY RELIEFS IN THE DISEASE OF CHARCOT-MARIE-TOOTH

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[Rilievi audiologici nella malattia di Charcot-Marie-Tooth]

SUMMARY

The disease of Charcot-Marie-Tooth is a degenerative disease of the nervous system, it strikes one or more neuronals systems and can involve neurons of the central and peripheral motor system, so it can interest Gall and Burdach faces or the spine-cerebral faces. From the histological point of view, we can observe an atrophy of the nervous cells and primary degeneration of the fibers subject to the process of continuous demyelinization distribution, the thesa mieliniches got reduced are inflated and are fragmented into lumps. Charcot-Marie-Tooth is due to the alteration of one or more geniuses that give way to the formation of the assone and the genesis of the myelin, whose synthetic deficit involves the reduction of the speed of nervous management. Two principal forms of the disease of Charcot-Marie-Tooth have been described: Form demyelinized and Form assonale. In the demyelinization and in the assonale forms the nervous fibers striked 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.

Key words: Degenerative disease, central and peripheral motor system, theca mieliniches, assone, demyelinization, nervous management

RIASSUNTO

La malattia di Charcot-Marie-Tooth è una malattia degenerativa del sistema nervoso, colpisce uno o più sistemi neuronali e può coinvolgere neuroni del sistema motore centrale e periferico, così come può interessare i fasci di Goll e Burdach o i fasci spino-cerebellari. Istologicamente si osserva atrofia delle cellule nervose e degenerazione primaria delle fibre soggette al processo di demielinizzazione a distribuzione continua, le guaine mieliniche si riducono, si rigonfiano e si frammentano in zolle. La malattia di Charcot-Marie-Tooth è dovuta ad alterazione di uno o più geni che provvedono alla formazione dell'assone e alla genesi della mielina, il cui deficit sintetico comporta la riduzione della velocità di conduzione nervosa. Sono state descritte due forme principali della malattia di Charcot-Marie-Tooth: assonale e demielinizzante. Sia nella forma demielinizzante che in quella assonale le fibre nervose colpite sono quelle più lunghe che sono destinate ai muscoli dei piedi, delle gambe e delle mani, ma anche le fibre sensitive vanno incontro a degenerazione e morte.

Parole chiave*: Malattia degenerativa, sistema motore centrale e periferico, guaine mieliniche, assone, demielinizzazione, conduzione nervosa*

Introduction

The disease of Charcot-Marie-Tooth is a degenerative disease of the nervous system, it strikes one or more neuronals systems and can involve neurons of the central and peripheral motor system, so it can interest the faces of Gall and Burdach or the faces of spine-cerebral. From the pathologic anatomy 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 demyelinization distribution, the theca mieliniches reduced are inflated and fragmented into lumps. The protoplasmatic glia assumes the character of fibrous glia with conseguent formation of glia scars responsible of the sclerotic hardening and wrinkling.

The disease of Charcot-Marie-Tooth is due to the alteration of one or more geniuses that give way to the formation of the assone and the genesis of the myelin, whose synthetic deficit involves the reduction of the speed of nervous management. Two principal forms of the disease of Charcot-Marie-Tooth have been described:

1) *Form demyelinized* characterized by the proliferation of the cells of schwann are typically marked by the histological comparison of bulb formation of onion, the covering of the assone 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 assone loses connection with the relative muscular fibers depriving it of some innervation making the apparent symptomatology;

2) *Form assonale* characterized from scarce demyelinization without histological comparison of bulb formations of onion, the myelin is entire even

if the assone introduces an alteration of its components with normal speed of nervous management.

In the demyelinization and in the assonale forms the nervous fibers striked 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.

The obolition of the achillesis 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 compromission of other organs among which the ear, phenomenon that can induce sensorial deafness.

Materials and methods

The study has been effected on two 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 bradyacusia. 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 characteristics of the auditory functionality through subjective test and objective audiologic:

1) *Tonal Audiometria* for the survey of the auditory threshold;

2) *Vocal Audiometria*, for the determination of the level of oral intelligibility, it foresaw different phases characterized by methodic followings vocal audiometriche: (Test of perception of quickly spoken altered or rasp, Test of biauricular 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);

3) *Impedenzometria*;

4) *ABR* (potential evoked auditory of the encephalic trunek);

5) *Effected central acoustic evaluation* using a tone from 500 Hz to 200 ms.

Results

The analysis of the results has underlined in both patients the presence of the examination tonal audiometric eliminar of sensorineural hearing loss. The vocal audiometriche evalution has given a sufficient degree of oral intelligibility appraised through special spondee.

To the tonal audiometric suprathresmold tests the test of the threshold of decadence, has shown presence of bilateral adaptation, probably referable to phenomenons of sonorous distorsion in relationship to the trime, sign of retrocochlea suffering, the study impedance audiometry the controlateral acoustic reflex threshold has noticed 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 both cases the presence of layout not structured, as it regards the central acoustic evaluation and specifically in the test of fusion bi auricular, 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 fidures.

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 under standing 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.

Conclusive considerations

In cases observed by us, 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. Within 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 audiometry automatic, 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 decotificative 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 retrococleare 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.

Bibliografia

1) Gozzano M.: *"Trattato delle malattie uditive nervose"*. 8° Ed. Padova Vallardi, NLE; 1981.
2) Tackmann W., Rad E.: *"Pattern shift visual evoked potentials in Charcot-Marie-Tooth disease"*. J. Neurol.; 1980.
3) Merritt H. H.: *"Neurologia"*. Vol. 2°, Napoli, Idelson; 1980.
4) Bess F., Josey A., Humes L.: *"Performance intensity functions in coclear and eighth nerve disorders"*. AMJ Otol.; 1979.
5) Fazio C., Loeb C.: *"Neurologia"*. Vol. 2°, Roma, SEU; 1995.
6) Jerger J., Jerger S.: *"Auditory findings in brain stem disorders"*. Arch. Otolaryngol; 1974.
7) Lynn G., Gilroy J.: *"Perception of rapidly alternating speech in patients with brain stem and cerebral hemi - sphere lesions"*, read bifore the ninth Colorado Otology. Audiology workshop; 1985.
8) Smith B., Resnik D.: *"An auditory test for assessing brain stem integrity: preliminary report"*. Laringoscope; 1972.
9) Swash M., Schwartz M. S.: *"Neuromuscolar disease"*. Springer, Verlag, 1988.
10) Katz J.: *"The staggered spondaic word test in keithr"*. Central auditory dysfunction; 1977.
11) Brunt M.: *"The staggered spondaic word test in Katz J."*. Handbook of Clinical Audiology; 1978.
12) Serra A., Maiolino L., Grasso D., Berretta M.: *"Studio ABR nella sclerosi multipla"*. Rivista italiana di Audiologia e foniatra 2; 1996.
13) Del Bo M., Giaccai F., Grisanti G.: *"Manuale di Audiologia"*. Ed. Masson; 1992.
14) Wrabetz L., Feltri M. L., Kleopla K. A., Scherer S. Inherited neuropathies: Clinical, Genetic aqnd Biological features. Chap. 39. In *"Myelin Biology and disorders"*. Editors: Lazzarini R. A., Griffin J. W., Lassman H., Nave K. A., Miller R. H., Trapp B. D. Elsevier Academic Press; 2004. Pagg: *905-951*.
15) Dematteis M. et al. Lancet 2001; 357*: 267-272*.
16) *"Surgery sometimes, Bracing Often, Caution Always"*. QUEST vol. 13, n. 5; september/october 2006.

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