The association of hereditary spastic paraplegia and hereditary motor and sensory neuropathy in the same family

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The association of hereditary spastic paraplegia (HSP) and sensory neuropathies have been reported in a number of cases. But it is rare to detect both entities seperately in different members of the same family. In the present study, we report clinical and electrophysiological findings of three members of a family. In two of the siblings the clinical picture was indistinguishable from "pure" hereditary spastic paraplegia, but electrophysiological studies revealed a predominantly sensory polyneuropathy. In the third case (the mother of the siblings), the clinical diagnosis was consistent with hereditary motor sensory neuropathy type 2, which was also confirmed by electrophysiological studies. We believe that, with the further genetic reevaluations, the hereditary spastic paraplegia with sensory abnormalities may take a new place in the classification of hereditary motor and sensory polyneuropathies, as a distinct entity.[Journal of Turgut Özal Medical Center 2(2):193-195,1995]

***Key Words**: Hereditary spastic paraplegia, hereditary motor and sensory neuropathy*

Aynı ailede herediter spastik parapleji ve herediter sensorimotor nöropatinin birlikteliği

Herediter spastik parapleji(HSP)'li hastalarda sensoriyel nöropati görülebileceği daha önce de bildirilmiştir. Ancak bu iki patolojinin aynı ailenin farklı fertlerinde ayrı ayrı gözlenmesi çok nadirdir. Bu yazımızda klinik ve elektrofizyolojik bulgularını bildirdiğimiz iki kardeşte klinik tablo "saf" herediter spastik parapleji'den ayrılamazken elektrofizyolojik incelemeler sensoriyel ağırlıklı bir polinöropatinin varlığını göstermiştir. Bu hastaların annesinde ise hem klinik tablo hem de elektrofizyolojik bulgular herediter motor ve sensoriyel nöropati Tip II ile uyumludur. Gerek klinik gerekse elektrofizyolojik olarak duyusal anormalliklerin eşlik ettiği herediter spastik paraplejilerin ileri genetik çalışmaların da yardımıyla, gelecekte diğer gruplardan ayrılarak herediter motor ve sensoriyel polinöropati sınıflaması içinde ayrı bir yer alacağına inanıyoruz. [Turgut Özal Tıp Merkezi Dergisi 2(2):193-195,1995]

***Anahtar Kelimeler** : Herediter spastik parapleji, herediter motor ve sensoriyel nöropatiler*

Association of hereditary sensorymotor neuropathies with pyramidal features has been reported in a number of cases but there is still a lack of consensus on the terminology of diagnosis in these cases. "Hereditary motor and sensory neuropathy (HMSN) Type V", "peroneal muscular atrophy with pyramidal features" and "hereditary spastic paraplegia associated with sensory neuropathy" were, all, used for the similar clinical pictures in previous reports[1-5]. However, the cases in whom the clinical picture is indistinguishable from that of "pure" hereditary spastic paraplegia (HSP) with a sensory neuropathy which is revealed electrophysiologically, form an intresting group and it is much more rare to detect both entities separately in different members of the same family. In the present study we report clinical and electrophysiological findings of three members of such a family.

CASE REPORTS

Case 1: This 16-year-old female, who was the fourth of six siblings, could never be able to walk without help but can stand up with the aid of two sticks. On

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the admission, there was spastic paraplegia with hyperreflexia, ankle clonus and extensor plantar responses. There was not any evidences of abnormalities of any type of sensory modalities, either in the the history or in the neurologic examination. Motor and sensory examinations, tonus and deep tendon reflexes were normal in the upper limbs. Kyphoscoliosis and bilateral pes cavus were observed in the physical examination. Electrophysiological studies revealed a sensorimotor neuropathy, which was predominantly sensorial (Table I).

There was not parity between the parents. After having the family history of two elder brothers who also had similar symptoms and mother who had a difficulty in walking, the family members were invited for examination. One of the elder brothers and the parents of the present patient were also examined. The neurologic and the electrophsiologic examinations of the father were normal.

Case 2: The elder brother of the first case was an 18-year-old male. He also could not be able to walk until he had had tenotomies of the Achilles tendons at the age of 12. Since then, he could be able to walk for a few hundred meters with the aid of two sticks. There were spasticity and hyperreflexia in the lower limbs. Bilateral ankle clonus and bilateral extensor plantar responses and an additional impairment of the position sense in the ankles, which was thought to be due to the previous surgery that he had had, were also observed. Tonus, deep tendon reflexes, motor and sensory examinations were normal in the upper limbs. Clinical diagnosis was hereditary spastic paraplegia. However, electrophysiological studies revealed a predominantly sensory polyneuropathy in this case, too (Table I).

Case 3: Slight weakness in the limbs, which is predominant in the distal extensor muscles, absent DTRs and inability to stand still were observed in the neurologic examination of the mother of the siblings, who was a 54-year-old female. She was walking in a wide based manner. She refused any sensory abnormalities but a slight impairment in the pinprick and vibration sensory modalities were observed. The clinical diagnosis was hereditary sensorymotor neuropathy type 2, which was also confirmed by electrophysiological studies (Table I).

In none of the patients any trophic ulcers and mutilation were not observed. Complete blood count and biochemical profile were within normal levels. None of the patients consented for sural nerve biopsy.

Table I. ENG findings of the cases

Case No	Nerve	Motor DL	Motor Amp.	Motor Veloc.	Sensory DL	Sensory Amp.
Case 1	R sural	couldn't be obtained				
	Peroneal	18.4	0.20	20.5 m/sn		
	Tibial	19.2	0.25	21.6 m/sn		
	Ulnar	14.4	3.84	12.5 m/sn	13.4	7.2
	L peroneal	19.6	0.45	11.9		
Case 2	R sural	couldn't be obtained				
	Peroneal	20.6	0.31	22.5 m/sn		
	Tibial	20.1	0.26	25.5 m/sn		
	Ulnar	13.0	4.1	18.0	15.5	6.8
	L peroneal	19.6	0.45	11.9		
Case 3	R sural	couldn't be obtained				
	Peroneal	couldn't be obtained				
	Tibial	14.4	0.32	30 m/sn		
	Ulnar	7.6	5.28	23.2 m/sn	6.8	5.2
	L peroneal	22.0	0.28	18.3		

DL : Distal latans

DISCUSSION

Hereditary spastic paraplegia (HSP) appears either in "pure" or "complicated" forms[1-6]. In the complicated forms, it is associated with amyotrophy, mental retardation, extrapyramidal features, cerebellar ataxia and deafness[1-4]. Its association with sensory neuropathies is also of special intrest and has been reported in a number of cases previously[1-5].

The most striking feature of the present family is the comorbidity of hereditary motor and sensory neuropathy (HMSN) and hereditary spastic paraplegia (HSP) with sensorimotor neuropathies in different members of the same family. The clinical and electrophysiological features was consistent with HMSN Type II in the third case, where the clinical picture, with ankle clonus, increased deep tendon reflexes, extensor plantar responses and spasticity in the lower limbs, was indistinguishable from "pure" hereditary spastic paraplegia in the first two cases. There was not any sensorial abnormalities but electrophysiological studies revealed sensorimotor neuropathy, which was predominantly sensorial, in first two of the cases.

Histologic findings in the sural nerve biopsies of three members of a large kinship of hereditary spastic paraplegia with electrophysiologically evident polyneuropathy, which was clinically indistiguishable from "pure" form of the disease, were reported recently[4]. Very similar pathologic features in these three cases point out that neuropathy is an integral part of the syndrom in this particular kinship[4].

Unfortunately we could not perform sural nerve

biopsies in the present cases. However, co-morbidity of HMSN type II and HSP with sensory neuropathy in different members of the same family indicates that association of hereditery sensory neuropathy with spastic paraplegia forms a distinct syndrom with an otosomal inheritance.

Further genetic researchs is needed to answer the question, whether the pyramidal tract features with sensory neuropathy (that can be clinically diagnosed as "hereditary spastic paraplegia") is a form of hereditary sensorimotor neuropathies or a distinct entity. We believe that, the hereditary spastic paraplegia with sensory abnormalities will take a new place in the classification of hereditary sensorymotor neuropathies, as a distinct entity.

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