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Comparative Anthropological Investigation of Patients with Definite Forms of Neuromuscular Diseases

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The material of this study includes 20 patients with definite forms of neuromuscular diseases — Hereditary motor and sensory neuropathy — Lom (HMSNL) and congenital cataracts facial dismophism neuropathy syndrome (CCFDN). Both diseases differ at present mainly on the basis of detail investigations of their ethiology, pathogenesis and clinical symptoms. The AIM of the present investigation is to enrich the phenotype characteristic of the diseases with the methods of anthropology; and to investigate the possibilities of their delimination on the basis of a comparative analysis of cephalometric and cephaloscopic characterization. The features used in the anthropological programme are by M a r t i n, S a l l e r [2]. The results show that more informative are the cephalometric features. The significant bilateral facial asymmetry established in both groups of patients prove the manifestation of an adverse effect of different genetical and exogeneous factors on the homeostasis of the patients' development.

Key words: cephalometry, cephaloscopy, asymmetry, neuromuscular diseases.

Introduction

The hereditary neuromuscular diseases are a heterogeneous group of diseases attacking the skeletal musculature and the peripheral nerves (including the diseases of neurons' cell bodies). The essential differences regarding their genetical basis, hereditary type, clinical symptomes, pathogenesis, laboratory analyses and dissemination between the population necessitates the use of many different methods in their investigation.

The AIM of the present study is to generalize in a comparative aspect the data of definte anthropological methods applied in the investigation of two kinds of neuromuscular diseases — Hereditary Motor and Sensory Neuropathy-Lom (HMSNL) and Congenital Cataracts Facial Dysmorphism Neuropathy Syndrome (CCFDN) with a view to investigate the possibilities of their additional delimination on an anthropological basis.

Both neuromuscular diseases are spread only between Gypsy populations [1, 3, 5].

Material and Methods

Subject of the study are 20 patients (gypsies): — 10 patients (2 males and 8 females) with HMSNL disease and 10 patients (4 males and 6 females) with CCFDN syndro-

T a b l e 1. Investigated anthropological features

No	Cephalometrical features	No	Cephalometrical investigated bilaterally features	No	Cephalocsopical features
			horisontal		
1	Head length (gl-op)	1	Frontotemporale - Nasion	1	Skin colour
2	Head breadth (eu-eu)	2	Entokanthion - Ektokanthion	2	Hair colour
3	Minimal frontal diameter (ft-ft)	3	Tragion - Pronasale	3	Hair form
4	Bizygomatical diameter (zy-zy)	4	Tragion - Subnasale	4	Eyebrow hairiness
5	Bigonial diameter (go-go)	5	Tragion - Alare	5	Beard hairiness
6	Trichion-nasion distance (tr-n)	6	Tragion - Chelion	6	Chest hairiness
7	Physiognomical face height (tr-gn)	7	Gonion - Gnathion	7	Body hairiness
8	Morphological face height (n-gn)	8	Preaurale - Postaurale	8	Eye colour
9	Morphological upper face height (n-pr)		vertical	9	Epicanthus
10	Physiognomical upper face height (n-sto)	1	Ektokanthion - Chelion	10	Eyelid fold
11	Nasal-subnasal length (n-sn)	2	Ektokanthion - Gonion	11	Brightness of eyeslit
12	Nasal-pronasal length (n-prn)	3	Entokanthion - Chelion	12	Forehead slope
13	Nose protrusion (sn-prn)	4	Tragion - Gonion	13	Overbrow arcs
14	Philtrum length (sn-sto)	5	Tragion - Frontotemporale	14	Horizontal face profile
15	Labrale sup.-labrale inf. distance (ls-li)	6	Superaurale - Subaurale	15	Cheek-bone swelling
16	Interocular diameter (en-en)		oblique	16	Chin form
17	Biocular diameter (ex-ex)	1	Tragion - Ektokanthion	17	Ear form
18	Nose breadth (al-al)	2	Tragion - Entokanthion	18	Lobulus auriculae
19	Lip length (ch-ch)	3	Tragion - Nasion	19	Nose form
20	Head circumference (g-op)	4	Tragion - Gnathion	20	Profile of nose-bone base
		5	Gonion - Subnasale	21	Profile of nose-cartilage base
		6	Gonion - Chelion	22	Nose apex
		7	Gnathion - Chelion	23	Nose wings swelling
				24	Nose orifice form
				25	Nostrils axe
				26	Upper lip profile
				27	Lip thickness

me.The patients are at the age 20-55 years.The programme of their anthropological study includes: 20 cephalometric measurements and 21 bilaterally measured cephalometric ones, as well as 20 cephaloscopic features (Table 1) all of them taken by the method of M a r t i n, S a l l e r [2].

The genetical analysis of HMSNL disease and CCFDN syndrome shows an autosome-recessive type of inheritance for both diseases. The genes recently have been mapped to the telometric regions of chromosome 8q24 for the HMSNL, and respectively of chromosome 18q for the CCFDN.

The summarized phenotypic characteristic includes a combination of neurological and otoneurological manifestations for the HMNSNL disease, and neurological, ophtalmological and hormonal ones for the CCFDN syndrome. For both neuro-muscular diseases the neuropathy is demyelinating in type, distal weakness and wasting are evident first in the lower limbs and later in the upper ones, as well as areflexia, ataxia, and palm and foot deformities are observed. The stage of their manifestation, as well as the age period in which they become evident, however are specific for every disease. The last mentioned is connected probably with the specific gene expression responsible for the manifestation of the diseases.

Results and Discussion

The cephaloscopic characterization of the patients with HMSNL and CCFDN shows similarity at a certain degree regarding the bigger part of the taken from us cephaloscopic features. All the patients have black, lank and soft hair, and swarthy skin. The eyes are spindle-shaped, with wide opened eyelid slit, horizontally situated, with external end slightly picked up. The colour of iris range from dark to light brown. The straight and slightly protruded noses prevail. The lips are procheilia and orthocheilia. The ear is with well-formed curve shape, which curve extends in many cases till 2/3 parts from the length of the external edge of the ear. Presence of the Darwin's tuberculum is found comparatively often from middle to big protrusion in the separate patients.

The general cephaloscopic characterization of the patients with CCFDN gives grounds to define two face types. Bigger height measurements of the lower jaw, rhomboid face configurations, flat profile, wide and straight or slightly sunken chins are typical for the first type. The strongly expressed prognathism (as a simple or alveolar one), narrowed horizontal profile of the face, slightly or strongly sunken chin are the cephaloscopic features characteristic for the second face type. Many tremas are also observed in the patients with the second face type, as a result from their strongly expressed prognatism.

A presence of bilateral asymmetry is obvious by a visual inspection at the patients with both neuromuscular diseases (HMNSL and CCFDN). In the patients with HMSNL the asymmetry affects different face's parts, while in the patients with CCFDN the asymmetry affects mainly the lower face's part.

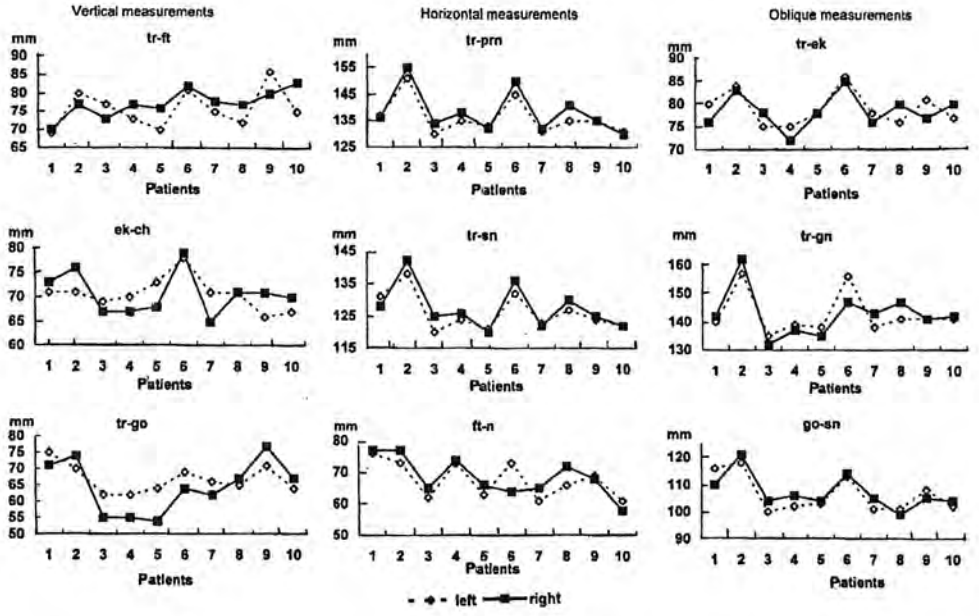

Fig. 1. Most significant manifestations of true facial asymmetry in three complexes of cephalometric measurements of patients with HMSNL

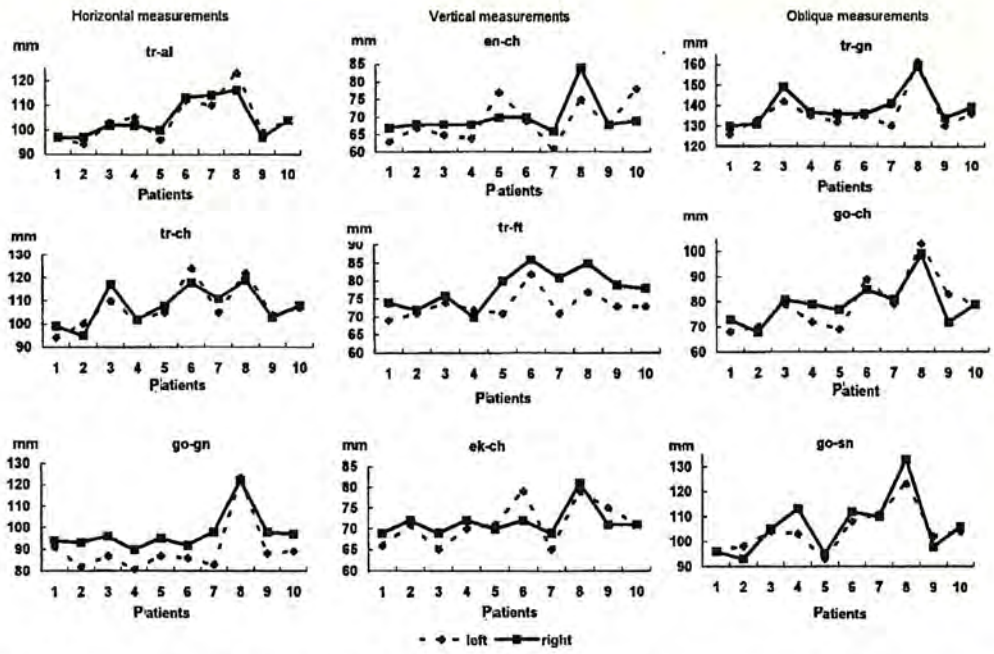

Fig. 2. Most significant manifestations of true facial asymmetry in three complexes of cephalometric measurements of patients with CCFDN

The comparative analysis of bilateral cephalometric measurements proves a presence of facial asymmetry in the patients with both diseases. Real facial asymmetry (differences over 2 mm) is found for all cephalometric measurements regardless their direction in the patients with HMSNL. As distinguished from them, real asymmetry of the CCFDN patients is found mainly for the lower jaw's measurements. The summarised analyses shows that for the patients with HMSNL the facial asymmetry predominates in right for the horizontal and oblique cephalometric measurements, and for the vertical ones — in left. For the patients with CCFDN respectively, the facial asymmetry predominates in right for the horizontal parameters, and in left for the oblique ones. Regarding the vertical cephalometric measurements, an equivalent manifestation of a facial asymmetry in right and left is established.

Biggest symmetry (differences till ± 2 mm) is observed about the cephalometric measurements (en-ch), (go-ch) and (tr-al) for the patients with HMSNL, and respectively about (en-ek) and (tr-ek) for the patients with CCFDN. The highest degree of real facial asymmetry (differences over 2 mm) is established about the horizontal measurements (tr-prn), (tr-sn) and (ft-n); the oblique measurements (tr-ek), (tr-gn) (go-sn), and the vertical ones (tr-ft), (ek-ch) (tr-go) for the HMSNL patients (Fig.1). Highest degree of real facial asymmetry is found about the horizontal measurements (tr al), (tr-ch), (go-gn); the oblique measurements (tr-gn), (go-ch), (go-sn); and the vertical ones (en-ch), (tr-ft), (ek-ch) in the CCFDN patients (Fig.2). Analysing the facial asymmertry in the cephalometric measurements with different direction can be concluded that the patients with HMSNL have relatively most symmetrical horizontal measurements, and the patients with CCFDN — most symmetrical oblique ones. As a whole, a tendency for higher facial asymmetry expressed is established with the ages' advance for the patients with both diseases.

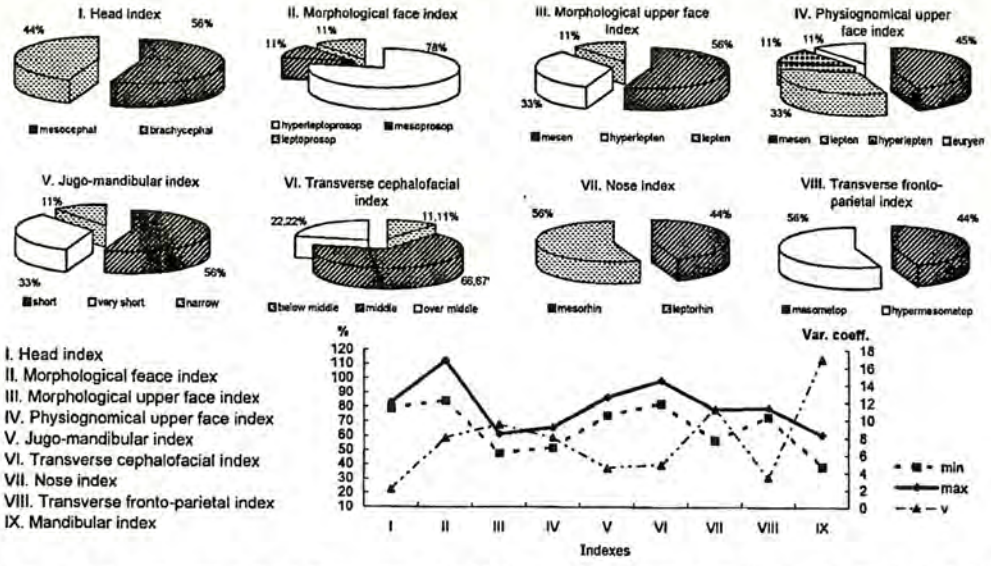

Fig. 3. Cephalometric indices in patients with HMSNL (*A*) and variation limits and coefficient of veriability (*B*)

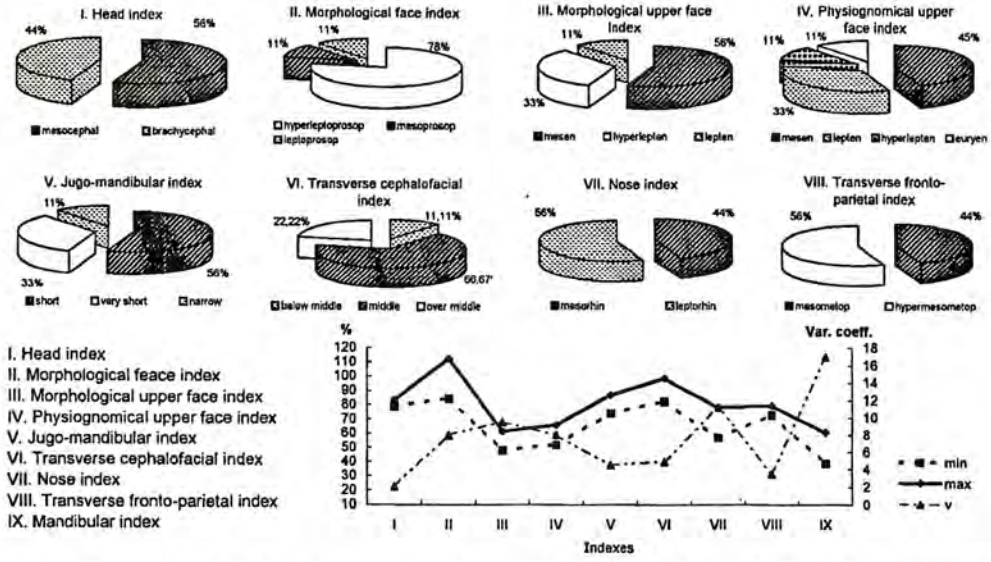

Fig. 4. Cephalometric indices in patients with CCFDN (*A*) and variation limits and coefficient of veriability (*B*)

On the basis of data about the cephalometric features' complex is observed a tendency to decrease of biparietal width, physiognomical and morphological face height, and nasal height in patients with HMSNL compared to the same measurements in healthy Bulgarians [4,6]. Decrease of head length, biparietal and bizygomatic diameters, morphological face height, nasal-pronasal length and head circumference is observed in the patients with CCFDN syndrome. About minimal frontal diameter,

bigonial diameter, interocular breadth and external bi-ocular breadth for the patients with HMSNL, and respectively abour head circumference, nose breadth and interocular breadth for the patients with CCFDN — a tendency to increase of these measurements is observed compared to the same ones in the healty Bulgarian population [4, 6].

The index cephalometric characterization of both diseases analysed in a comparative aspect shows a predominance of the mesocephalic, mesoprosopic, leptorrhinic and eurymetopic types in the patients with HMSNL, and respectively the brachycephalic, hyperleptoprosopic, and mesometopic types (to the limit of eurymetopic ones) for the patients with CCFDN (Fig.3 and 4). Most variables are the nose index, the morphological upper face index and the jugo-mandibular one in the HMSNL patients (Fig.3). And for the CCFDN patients, most variables are the mandibular index, the nose index, the morphological upper face index and the physiognomical upper face one (Fig.4).

The comparison between the patients with both diseases according to the physiognomical face height and both its parts (physiognomical upper face height and mandibular height) shows that the faces of patients with CCFDN syndrome are more long, as about absolute measurements, so for the indexes (towards the maximum interzygomatic breadth). These findings are basically due to the considerably higher lower jaw of the patients with CCFDN syndrome, and the higher mophological face height and physiognomical upper face height. The upper part of the face in the CCFDN patients is longer than that one of the HMSNL patients according to the comparative analysis of the physiognomical upper face height analysed separately. In maintenance of this conclusion are also the data from the comparative analysis of the mandibular and jugo-mandibular indexes. According to them (these indexes), the lower face part of the HMSNL patients is more round, and for the CCFDN patients it is more volumetric and prolonged. The comparative analysis of the values from thransversal cephalo-facial index determinate the patients with CCFDN syndrome as persons with poorly developed face part toward the cerebral one of the head. The opposite tendency is observed in the patients with HMSNL disease, which possibly is due to the high values of their maximal interzygomatic breadth and the relative deminution of the biperietal breadth.

Conclusion

In conclusion we can generalise, that the comparative analysis of the cephalometric and cephaloscopic characterization of the patients with HMSNL and CCFDN gives a real possibility for an additional delimination of both diseases on an anthropological base. In this meaning more informative are the cephalometrical features. As a whole the data from the anthropological investigation enrich considerably the phenotype characterization of both diseases, which till now are investigated mainly on data from their clinical, neuromorphological and electromyographical analyses. The different trends traced out in the manifestation of the studied anthropological features are determined first of all from the differences of the expression of the genes responsible for both diseases. The bilateral face asymmetry in HMSNL disease and CCFDN syndrome proves the negative influence of different genetical and exogeneous factors on the homeostasis of the individual development of the patients. The appropriate anthropological methods applied can give the possibility in the future study of both diseases for a precise assessment of these factors.

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