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EPIDEMIOLOGICAL CHARACTERISTICS OF HEREDITARY MOTOR AND SENSORY NEUROPATHY IN THE SUMY REGION

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The aim of the study *was to investigate the characteristics of the spread of hereditary motor and sensory neuropathy type 1A in the Sumy region, in terms of administrative units and specific ethnic groups of the population.*
Materials and methods. *An epidemiological study of hereditary motor and sensory neuropathy (HMSN) type 1A in the Sumy region was conducted based on patient referrals to various healthcare facilities from 2014 to 2017. The prevalence of HMSN in the Sumy region was studied using an epidemiological approach. The prevalence rate of HMSN was calculated for different administrative units of the Sumy region, as well as for specific ethnic groups of the population.*
Results. *The study registered 67 patients with hereditary motor and sensory neuropathy type 1A in the Sumy region. The prevalence rate of hereditary motor and sensory neuropathy type 1A in the Sumy region was 5.96 per 100,000 population. Of the HMSN patients, 47.8 % (32 individuals) resided in urban areas, while 52.2 % (35 individuals) lived in rural areas. In terms of gender distribution, 59.7 % (40 individuals) were female, and 40.3 % (27 individuals) were male. Most patients were of Ukrainian ethnicity (77.6 %), followed by Russians (20.9 %). Belarusians accounted for 1.5 % of HMSN patients in the Sumy region. Among other ethnic groups present in the Sumy region, HMSN type 1A was not detected.*
Conclusions. *Hereditary motor and sensory neuropathy type 1A is unevenly distributed in the Sumy region. The heterogeneous distribution of the disease is primarily attributed to the characteristics of settlement in specific geographic regions, which have led to the formation of territorial, national, and religious isolates with the accumulation of genetically heterogeneous types in these populations, resulting in an increased genetic burden*
Keywords: *epidemiology, prevalence rate, population size, ethnic groups, hereditary motor sensory neuropathy, Sumy region*

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1. Introduction

Hereditary motor-sensory neuropathies are the most common hereditary neuromuscular disorders [1]. It is difficult to estimate the prevalence of hereditary motor-sensory neuropathy due to the wide variability of clinical symptoms and different forms of the disease [2]. These difficulties explain the high variability of the prevalence rates reported in epidemiological studies: for example, in Libya per 100 thousand population – 8 cases, Nigeria – 10, South Wales – 17, Northern Sweden – 20, Northern Spain – 28 [3].

Most epidemiological studies have analyzed either all forms of hereditary motor-sensory neuropathy, or hereditary motor-sensory neuropathy type 1, as the most common subtype of the disease.

The presence of other hereditary neuropathies is more common with age, and thus hereditary motor-sensory neuropathy may be more difficult to distinguish from other neuropathies.

According to Mladenovic et al., the prevalence of hereditary motor-sensory neuropathy varies in different population groups and different regions, even within a country [4].

For example, isolation leads to an increase in the degree of relatedness of group members, which inevitably contributes to the growth of inbred offspring in generations. That is why concentrations of genes can be observed in isolated groups connected by a common origin [5]. Hereditary motor-sensory neuropathies have little effect on life expectancy, which causes their accumulation in individual families and in populations as a whole [6].

If the size of the population is limited, then gene frequencies can undergo changes in generations that are completely independent of the influence of mutations, migrations, and selection. There are differences between the gene frequencies of offspring and the cohort of parents [5].

The prevalence of HMSN in different populations varies in a wide range, the most relevant at the initial stage of the genetic-epidemiological study of HMSN is the determination of the population frequency of this mutation in a specific region [7].

Identifying the territorial features of the distribution, spectrum, and variability of HMSN manifestations is the basis for creating an effective monitoring system, as well as developing diagnostic and preventive methods optimal for a specific region. The study of the regional epidemiological features of HMSN distribution can also contribute to the rational allocation of existing and potential resources by health care institutions. In this regard, it is relevant to study the peculiarities of the spread of hereditary motor-sensory neuropathy type 1A in the Sumy region.

The aim of the study – study of the peculiarities of the distribution of hereditary motor-sensory neuropathy type 1A in the Sumy region in terms of administrative-territorial units and individual ethnic groups of the population.

2. Materials and methods

Epidemiological research of hereditary motor-sensory neuropathy type 1A in the Sumy region was carried out based on patient referrals to various medical and preventive institutions (polyclinics, medical genetic centers, archives of the Ministry of Health and Welfare) in the period from 2014 to 2017. Family members of patients with hereditary motor-sensory neuropathy type 1A were also examined. According to the results of the molecular genetic examination, hereditary motor-sensory neuropathy of type 1A was detected in mildly symptomatic family members of several patients.

The study was carried out based on the Educational and Scientific Institute of Postgraduate Education of the Kharkiv National Medical University. All patients who were involved in the study gave their consent to the processing of personal data.

The commission on ethics and bioethics of the Kharkiv National Medical University at its meeting (protocol No. 5 dated February 1, 2023) decided that the materials of this study submitted for examination meet international ethical requirements and do not violate ethical norms in science and standards for conducting biomedical research.

The work was carried out in accordance with the legislation in force in Ukraine, the Law of Ukraine "On Medicinal Products", 1996, Art. 7, 8, 12, principles of ISN GCP (2008), order of the Ministry of Health of Ukraine No. 690 of 23.09.2009 "On the approval of the Rules for conducting clinical trials and examination of clinical trial materials and the Standard Regulation on the Commission on Ethics", with changes and additions. The research was carried out with minimal psychological losses on the part of the patients and was conducted in accordance with ethical requirements in accordance with maintaining the confidentiality of the information obtained in the research process.

To study the prevalence of hereditary motor-sensory neuropathy (HMSN) in the Sumy region, an epidemiological method of studying morbidity was used [8].

The diagnosis of HMSN was made in accordance with the recommendations of the WHO Research Group on Neuromuscular Diseases based on diagnostic criteria [9].

The prevalence rate of hereditary motor-sensory neuropathy was calculated both for various territorial-administrative units of the Sumy region and for individual ethnic groups of the population according to the following formula:

$$Prevalence\ rate = \frac{number\ of\ registered\ patients}{population\ size} \cdot 100 \text{ thousands.}$$

Data on the population in various territorial-administrative units of the Sumy region were taken from the statistical collection of the State Statistics Service of Ukraine for 2015 [10], and information on the population in individual ethnic groups was obtained from the data of the All-Ukrainian population census of 2001 [11].

Descriptive statistics for qualitative accounting features are presented in the form of absolute values and percentages.

3. Research results

According to the data of the State Statistics Service of Ukraine, as of January 1, 2015, the total population of Sumy region was 1,123,448 people, in particular, the urban population was 768.4 thousand people (68.4 %), the rural population was 355.0 (31.6 %) thousand people. According to the gender structure, women predominate, so as of January 1, 2015, there were 610,000 women (54.3 %) and 513,400 men (45.7 %) in the region. The advantage of the number of women over men is due to a number of factors, among which the most important are a lower mortality rate among the female population under the age of 60 and labour migration of men. The average age of the population of the Sumy region is 41.7 years, in particular in cities – 40.3, in rural areas – 44.4 years [10].

Table 1 presents the results of a study of the prevalence of hereditary motor-sensory neuropathy in the Sumy region and the city of Sumy in the period from 2015 to 2010.

According to the results of the study, 67 patients with HMSN type 1A were registered in the Sumy region. The prevalence of hereditary sensory-motor neuropathy type 1A in the Sumy region is 5.96 per 100,000 population.

47.8 % (32 persons) of patients of the Sumy region live in cities and 52.2 % of patients (35 persons) live in rural areas. The number of urban and rural population in the Sumy region in 2015 was 61.82 % and 38.18 %, respectively, that is, the distribution of patients with hereditary sensory-motor neuropathy type 1A in the Sumy region is not determined by demographic ndicators.

In the structure of patients, 59.7 % (40 people) were women and 40.3 % (27 people) were men.

As the results of the research showed, hereditary motor-sensory neuropathy type 1A was detected in 7 cities of the Sumy region, of which 4 cities of regional importance (Sumy, Konotop, Okhtyrka, Shostka) and 3 cities of districts importance (Bilopillia, Krolevets, Putivl). Among the urban population of the Sumy region, the prevalence rate of hereditary sensory-motor neuropathy type 1A is 7.6 per 100,000 population.

Table 1

Prevalence of hereditary motor-sensory neuropathy in the Sumy region

The name of the territorial-administrative unit	Total population	Population size		HMSN prevalence rate	Number of HMSN patients
		urban	rural		
Sumy region	1 123 448	768 390	355 058	5.96	67
Cities of regional importance					
Sumy	268 642		–	5.2	14
Glukhiv	34 020	34 020	–	–	–
Konotop	88 252	88 252	–	5.6	5
Lebedyn	25 721	25 721	–	–	–
Okhtyrka	48 817	48 817	–	6.1	3
Romny	41 364	41 364		–	–
Shostka	77 847	77 847		5.1	4
Cities of districts importance					
Bilopillia	16 598	16 598	–	12.0	2
Burin	8 920	8 920	–		
Vorozhba	7 262	7 262	–		
Druzhba	4 919	4 919	–		
Krolevets	23 404	23 404	–	12.8	3
Putivl	16 009	16 009	–	6.2	1
Seredyna-Buda	7 144	7 144	–		
Trostyanets	21 003	21 003	–		
Districts of the region					
Bilopilsky	51 212	30 531	20 681	15.6	8
Burynskyi	25 580	8 920	16 660		
Velikopysarivskyi	19 466	7 330	12 136	5.1	1
Hlukhivskyi	23 672	3 891	19 781		
Konotopskyi	29 695	2 576	27 119	10.1	3
Krasnopilskyi	28 983	10 431	18 552	–	–
Krolevetskyi	38 864	23 404	15 459	5.1	2
Lebedynskyi	45 721	7561	25 543	8.7	4
Lipovodolinskyi	19 111	5 252	13 879	–	–
Nedrigailivskyi	24 811	8 724	16 087	–	–
Okhtyrskyi	26 794	2 413	24 381	–	–
Putivlskyi	28 156	16 009	12 147	–	–
Romenskyi	33 778	33 778	-	11.8	4
Sredino-Budskyi	16 689	9 053	7 636	–	–
Sumskyi	62 381	10 868	51 513	11.2	7
Trostyanetskyi	35 766	21 003	14 763	8.4	3
Shostkinskyi	20 876	7 070	13 806	14.4	3
Yampilskyi	24 199	16 272	7 927	–	–

Hereditary motor-sensory neuropathy type 1A was detected in 9 administrative districts of Sumy region: Bilopolskyi, Velikopysarivskyi, Konotopskyi, Krolevetskyi, Lebedynskyi, Romenskyi, Sumskyi, Trostyanetskyi, Shostkinskyi. The average value of the prevalence of hereditary motor-sensory neuropathy type 1A among the rural population of the Sumy region is 10.0 per 100,000 population. In the identified administrative districts, the highest prevalence rate was observed in the Bilopolskyi district – 15.6 per 100,000 population; the lowest – in the Velikopysarivskyi and Krolevetskyi administrative districts – 5.1 per 100,000 population.

The analysis of the results of the clinical and genealogical study of patients in the Sumy region allowed us to establish that in 20.9 % of patients (14 people) hereditary motor-sensory neuropathy type 1A was represented by the only patient in the family. 79.1 % (53 persons) had a family history of the disease.

The distribution of patients with hereditary motor-sensory neuropathy type 1A in the Sumy region by age is presented in Table 2.

The study also analyzed the prevalence of hereditary motor-sensory neuropathy in the Sumy region among ethnic groups.

Table 3 presents the distribution by ethnic groups of patients with hereditary motor-sensory neuropathy in the Sumy region.

Table 2

Distribution of patients with hereditary motor-sensory neuropathy type 1A by age in the Sumy region

Age group	The number of patients with HMSN type 1A		
	women	men	total
II decade (18–19 years)	4	2	6
III decade (20–29 years)	6	5	11
IV decade (30– 39 years)	11	7	18
V decade (40– 49 years)	9	7	16
VI decade (50– 59 years)	6	3	9
more than 60 years	4	3	7

Table 3

Distribution by ethnic groups of patients with hereditary motor-sensory neuropathy in the Sumy region

Ethnic group	The number of ethnic groups	The share of the ethnic group in the total population structure, %	The number of patients with HMSN 1A type in the ethnic group (n)	HMSN prevalence rate by ethnic group
Ukrainians	997 621	88.8	52	5.2
Russians	105 604	9.4	14	13.2
Belarusians	3 370	0.3	1	17.8
Romani	1 396	0.1	–	–
Armenians	1 217	0.1	–	–
Moldovans	1 123	0.1	–	–
other nationalities	13 484	1.2	–	–

As the results of the conducted research showed, the vast majority of patients were represented by Ukrainians (77.6 % of patients). The second largest group of patients are Russians (20.9 %). 1.5 % of patients with hereditary motor-sensory neuropathy type 1A in the Sumy region were Belarusians. Among other ethnic groups represented in the Sumy region, hereditary motor-sensory neuropathy type 1A was not detected. Due to the predominance of Ukrainians in the population structure of the Sumy region (88.8 %) and the significantly smaller number of other ethnic groups, the highest rate of prevalence of hereditary motor-sensory neuropathy type 1A was determined among Ukrainians. However, because of "large" and "small" samples, data on the prevalence of hereditary motor-sensory neuropathy type 1A among other ethnic groups represented in the Sumy region are not representative and cannot be transferred to the entire population.

4. Discussion of research results

Based on the results of the research, it was established that the prevalence of hereditary motor-sensory neuropathy type 1A in the Sumy region is 5.96 per 100,000 population. Hereditary motor-sensory neuropathy type 1A is unevenly distributed in the Sumy region. Features of the prevalence of hereditary motor-sensory neuropathy type 1A in the Sumy region are like the features of the prevalence of the disease in other populations.

Comparison of the obtained prevalence rate of hereditary motor-sensory neuropathy type 1A in the studied region of Ukraine with the prevalence rates of the disease in different populations of the world determined in previous studies is complicated due to the difference in the methodologies for determining the prevalence in different studies.

Thus, according to literature data, most prevalence rates of hereditary motor-sensory neuropathy type 1A vary in the range from 4.0 to 15.0 per 100,000 population, and the prevalence rate of all genetic forms of hereditary neuropathies ranges from 0.1 to 41.0 per 100,000 population and is characterized by high geographic variability [9].

The relatively low rate of prevalence of hereditary motor-sensory neuropathy type 1A in the studied region of Ukraine may be due to difficulties in diagnosing the disease due to pronounced clinical polymorphism, mild clinical manifestations at the beginning of clinical manifestation, which cause a low rate of referral of patients to medical institutions at the onset of the disease, and the presence of sporadic cases with late manifestation.

The heterogeneity of the prevalence of hereditary motor-sensory neuropathy type 1A in certain territorial-administrative regions of the studied region of Ukraine is associated with a low number of factors, the most important of which are the demographic indicator, the sex-age composition of the population, the level of genetic load with the accumulation of genetic "cargo" in individual families and geographical areas, which is due to the autosomal dominant type of inheritance.

The uneven prevalence of hereditary motor-sensory neuropathy type 1A among the ethnic groups represented in the Sumy region is associated with the effect of "small samples" and the unrepresentativeness of the data, which cannot be transferred to the population as a whole.

Study limitations:

1. A small sample of HMSN patients.

2. Complicated diagnosis of the disease due to pronounced clinical polymorphism.

Prospects for further research. The obtained results can be used to identify the territorial features of

distribution, spectrum, and variability of HMSN manifestations in other regions of Ukraine.

5. Conclusions

1. The prevalence of hereditary motor-sensory neuropathy type 1A in the Sumy region is 5.96 per 100,000 population.

2. Hereditary motor-sensory neuropathy type 1A is spread unevenly in the Sumy region.

3. The main reason for the heterogeneity of the spread of the disease is the features of the settlement of certain geographical regions, which contributed to the formation of territorial, national, and religious isolates with the accumulation of heterogeneous types in the population of these regions, causing the growth of genetic "cargo".

Conflict of interests

The authors declare that they have no conflicts of interest related to this study, particularly financial, personal, authorship, or otherwise, that could affect the study and its results presented in this article.

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Data availability

Data will be provided upon reasonable request.

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