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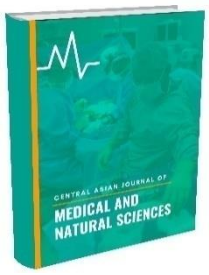

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# Hereditary Diseases of the Nervous System, Their Prevalence and Epidemiological Status

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**Abstract:** Hereditary diseases of the nervous system have a high proportion in the overall structure of neurological pathology. The particular relevance of this problem is due to the lack of effective treatment methods and the existing possibility of repeated cases in subsequent generations. The study of neurohereditic diseases is a complex task not only of modern neurogenetics, but also of clinical neurology. This is due to the presence of characteristic features that distinguish neurohereditic diseases from other pathologies of the nervous system.

**Key words:** Hereditary diseases, clinical polymorphism, epidemiology, Human genome.

**Relevance.** The health of the population occupies one of the first places in the system of vital values of any state. Reducing morbidity and preserving the health of the population are the most important socio-economic tasks of the country and they are particularly acute today for Uzbekistan, since such factors as the current population decline, along with high morbidity and mortality, are a real threat not only to the development of the country, but also to its national security.

As you know, hereditary diseases of the nervous system are one of the important medical and social problems. They make up a significant share in the burden of monogenic diseases and in the structure of neurological pathology, especially in children and adolescents [1]. Hereditary diseases of the nervous system manifest more often in adolescence and are steadily progressive without treatment, leading patients to severe disability.

Excessive diversity of nosological forms, genetic heterogeneity and pronounced clinical polymorphism of hereditary diseases of the nervous system not only complicates their diagnosis, but also complicates the conduct of medical and genetic counseling in families burdened with these ailments [3].

The first place among all hereditary monogenic neurological diseases is occupied by hereditary neuromuscular diseases (NMD), which include progressive muscular dystrophy, which is a clinically and genetically heterogeneous group with primary damage to skeletal muscles of a non-inflammatory nature, which is also characterized by a wide nosological spectrum and pronounced clinical polymorphism [2].

In recent years, significant progress has been made in the study of PMD, associated with the disclosure of the structural and molecular foundations that regulate the functioning of muscle fiber. However, despite the fact that active attempts of gene therapy and the widespread introduction of prenatal diagnosis of PMD into practice, the need for timely allocation of the development of preventive measures, social rehabilitation and adaptation of patients continues to be one of the most important urgent problems of medical science [7].

Thanks to the progress in the development of society and medicine, at the end of the 90s of the XX century, the concept of genomic or personalized medicine was formed, which is based on the principles of the classics of patronymic medicine of the XIX century. The principle of a personal approach to the patient received a new reading in the XXI century in the form of the main postulate of personalized medicine: "routine use of genotypic analysis, usually in the form of DNA testing, in order to improve the quality of medical care." New approaches make it possible to move from the "response" medicine that exists today, which deals with an existing disease and fights symptoms, to personalized or genomic medicine. Leroy Hood, a Nobel Prize lauret and one of the founders of personalized medicine, defines it as medicine of four "P": predictive (aimed at predicting the disease before its symptomatic appearance); preventive (preventing disease); personalized (taking into account individual, primarily genetic characteristics of the patient) and participatory (participatory medicine, i.e. implying the active participation of the patient in the identification of his genetic characteristics and preventive measures) [1, 2].

## 1. Analysis of literature data and problem statement

The implementation of such projects as "Human Genome", "Genomes of races", "1000 and 1 genome", "GWAS" (Genome-Wide Association Study), allowed us to accumulate a huge amount of information about monogenic hereditary diseases (NB), multifactorial (polygenic) diseases and various genetically determined predispositions. Based on studies of the human genome, it was possible to identify not only the genes that cause monogenic NB, but also mutations that lead to the most frequent, so-called multifactorial diseases resulting from the interaction of genetic factors and unfavorable environmental conditions. It is proved that the etiopathogenetic basis of such multifactorial diseases as essential hypertension, diabetes mellitus, bronchial asthma, atherosclerosis, some forms of cancer is a genetic factor, namely the presence of so-called "predisposition genes", the damaging effect of which is realized against the background of adverse environmental factors [3-5].

And if for multifactorial diseases, genetic changes are not the only ones determining the development of the disease, then for monogenic NBS they are the etiological cause. Taking into account the absence of pathogenetic methods of treatment of most forms of NB to date, the most effective method of combating this pathology is not so much prevention, but first of all, the prediction of the occurrence of the disease. The words of the French scientist Jean Dosse, founder of predictive medicine, Nobel Prize winner: "to prevent a disease, it must be foreseen" are now more relevant than ever [6].

Hereditary diseases of the nervous system (NBNS) make up a significant part of the structure of monogenic NBS. They account for a significant part, more than 5%, of all neurological pathology in modern society. NBNS occupy a special place both among all forms of hereditary human diseases and pathology of the nervous system, which is associated with their overall high prevalence, a wide variety of nosological units, significant phenotypic polymorphism and pronounced genetic heterogeneity. Most NBNS are of a severe progressive nature, often lead to early disability, and sometimes death of the patient, while effective pathogenetic treatment has not been developed to date [4, 7, 8].

The individual approach, as the basis of the principle of personalized medicine, is partly due to the presence of pronounced clinical polymorphism of many diseases, manifested in the development of individual individuals of various degrees of pathological manifestations from erased to severe forms.

The development of NB occurs as a result of the influence of an etiological factor in the form of strictly deterministic gene mutations that have passed through the evolution of tens and hundreds of years of natural selection through the acquisition of a certain type of inheritance and molecular genetic features, as well as a clear mechanism of pathogenesis, fully controlled by gene activity. In this regard, it would be possible to assume the presence of strictly unified clinical manifestations within a certain nosological form. However, many NBNS are clinically and genetically heterogeneous with a wide variability in the age of manifestation of the disease, its duration and duration of life, the completeness and severity of clinical manifestations, as well as the depth of the pathological process. The clinical polymorphism of NBNM significantly complicates their differential diagnosis and, accordingly, the prevention of their possible development in the population [3, 4, 9].

For the first time, clinical polymorphism of the NBNS began to be deeply analyzed back in the 20-30s of the XIX century by an outstanding neurologist and geneticist S. N. Davidenkov, who proved that its main causes are the environment, the nature of mutations and specific genotypic features of the population; genetic heterogeneity was also revealed, manifested in the occurrence of various genotypes of the disease and masquerading as clinical polymorphism.

However, despite a fairly wide clinical polymorphism, the absence of a smooth transition from norm to pathology is characteristic of all NBS, and even mild forms of the disease have, although very minimal, but significant diagnostic signs for this disease, which corresponds to the basic genetic rule: the normal genotype determines the normal phenotype, and the mutant genotype determines the mutant phenotype. [8, 12].

Taking into account the above, early diagnosis and prevention of NBNS are of great medical, social and economic importance, the full implementation of which is impossible without an idea of the etiology, pathogenesis and features of their spread.

**2. Population-epidemiological aspects of hereditary diseases of the nervous system**

Significant priority is currently being given to research that allows us to assess the population frequency of hereditary pathology and to study their clinical polymorphism and genetic heterogeneity. Many publications of both domestic and foreign authors are devoted to the epidemiology of individual NBS in different populations, indicating that there are no NBS with the same population frequency of occurrence and the spectrum of gene mutations that determine their development.

The study of the prevalence of NBNS is important both from a theoretical and practical point of view. The theoretical significance lies in determining the model of the genetic structure of the population, taking into account the territorial distribution of mutant genes and the features of genetic differentiation of the population, as well as the genetic burden of the population, which is defined as a decrease in the average fitness of the population compared to the population, all individuals who have a genotype that ensures maximum fitness. From a practical point of view, the identification of population-territorial features of the distribution, spectrum, variability of manifestations and the genetic nature of hereditary pathology is the basis for creating an effective system for monitoring them, as well as developing diagnostic and preventive methods that are optimal for a particular region [17-20].

In the literature, the possible causes of inter-population differences in the prevalence of certain forms of hereditary diseases of the nervous system are very intensively discussed, the primary of which are molecular genetic features peculiar to specific populations. According to the modern theory of evolution, the main factors determining the formation of the gene pool of various populations are natural selection, migrations, mutations and random events. As a result of their combined impact in specific environmental conditions, the integration of unique features of the structure of certain groups occurs, manifested in the form of differentiation of different populations in the feature space.

The dynamics of modern microevolutionary processes is quite complex and ambiguous, and, in general, differs significantly from that in small isolated populations of the past. Recent decades have been characterized by an increase in urbanization; a significant increase in the scale of migrations and, as a consequence, the expansion of the boundaries of breeding areas, an increase in the proportion of interethnic marriages and the associated recombination of genes, which together creates prerequisites for the formation of new genotypes and a significant increase in the level of genetic variability. Considering also that along with the listed factors, there is a significant decrease in the role of natural selection and the influence of a large number of mutagenic factors, it can be assumed that the genetic load in human populations is increasing. The founder of population genetics G. Meller spoke about this back in the 1950s: "Many genotypes that had reduced fitness and were rejected by natural selection in modern society live to reproductive age and leave offspring with reduced fitness as well; as a result, the genetic burden is growing, and the fitness of modern populations is falling, which can have dramatic consequences for all mankind ..." [4, 21-24].

It should also be noted that the differentiation in the burden and diversity of NB is revealed not only between different populations, but also among individual groups of the same population species living in different climatic, geographical and ecological zones. Consequently, the results obtained in the study of one local ethno-territorial region cannot be transferred to the entire population as a whole, and therefore the study of the patterns of formation of the structure and dynamics of the genetic characteristics of the population of individual regions is relevant, scientifically sound and logical [15, 23].

An important feature of the Ukrainian population is the large anthropological and cultural-ethnographic diversity of local territorial-ethnic areas associated with the complex historical process of their formation in the process of settling the area of the Ukrainian population, which since ancient times has been located at the intersection of the forest and forest-steppe of Eastern Europe, the Eurasian steppes and the Balkan-Carpathian historical region. The main territorial historical and ethnographic regions of Ukrainians are distinguished: Middle Dnieper, Podillia, Galicia, Zaporozhye, Slobozhanshchina, Polesie, Transcarpathia, Bukovina, Pokutye, Tavria, Donbass, Southern Bessarabia [25, 27, 29].

A questionnaire survey of a wide range of medical information sources is used, suggesting the identification of approximately 500 forms of the spectrum of monogenic pathology and the subsequent examination of identified patients by highly qualified specialists consisting of a neurologist, pediatrician, optometrist and dermatologist. The final verification of the diagnosis, if necessary, is carried out on the basis of highly informed diagnostic methods in medical and genetic centers.

In the standard of epidemiological research of hereditary pathology, a limited geographical region with a population of 0.5 to 5 million people is used, which makes it possible to avoid a negative impact on the indicators of any family with several affected. This region should not be excessively large, which is necessary to ensure reliable detection of cases.

The population-genetic method will allow us to get a unified idea of the components of the load of hereditary pathology (burden, spectrum), as well as about the factors correcting the quantitative and qualitative distribution of various diseases in the structure of the load (autosomal dominant BP, autosomal recessive AR and X-linked forms).The territory of Slobozhanshchina belongs to the second geographical zone of the first cluster of the Ukrainian population and corresponds to the Central Ukrainian anthropological type, which, in turn, is divided into western and eastern variants [27,30]

**Conclusions**

In conclusion, it should be noted that the epidemiological and molecular genetic foundations of most NBNS remain largely unexplored; their etiology and pathogenesis are unclear, but it is obvious that

the mechanisms of their formation are complex and diverse. And it is these diseases that currently have no adequate methods of treatment other than preventive, i.e. aimed at preventing the disease. Therefore, in order to ensure the most effective diagnosis and prevention of NBNS, it is necessary to study their prevalence, clinical and molecular genetic characteristics in individual regions and ethnic groups.

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