

Khudoikulova Nasiba Ismailova  
Lecturer at the Department of Medical Biology  
Bukhara State Medical Institute  
Khudoykulova N.I.

## INDICATORS OF HEREDITARY DISEASES IN THE BUKHARA REGION

### ИРСИЙ КАСАЛЛИКЛАРНИ БУХОРО ВИЛОЯТИДА УЧРАШ КУРСАТКИЧЛАРИ

Худойкулова Насиба Исмаиловна  
Тиббий биология кафедраси уқитувчиси  
Бухоро Давлат тиббиет институти

«Бухоро минтақасида 2021-2024 йиллари мобайнида туғма аномалиялар ривож (ТАР) ни ўрганиш бўйича эпидемиологик изланишлар олиб борилди. Бу изланишлар натижасида шу аниқландики, шу ТАР лар, яъни туғма юрак нуқсони (пороки) ва Даун синдроми аномалиялари бошқа патологиялар ўртасида энг кўп учраши кузатилди.

**Калит сўзи:** Даун синдроми, аномалия, туғма аномалия, Полидактилия, Синдактилия.

## ИНДИКАТОРЫ НАСЛЕДСТВЕННЫХ БОЛЕЗНЕЙ В РЕГИОНЕ БУХАРЫ

Проведено эпидемиологическое исследование по изучению распространения врождённых аномалий развития в Бухарской области за период с 2021 по 2024 годы. Было показано, что такие как врожденный порок сердца и синдром Дауна, были наиболее распространенными патологиями среди других врожденных аномалий развития.

**Ключевые слова:** синдром Дауна, аномалия, врожденные аномалии, полидактилия, синдактилия.

## INDICATORS OF HEREDITARY DISEASES IN THE BUKHARA REGION

Carried out an epidemiological study on the prevalence of congenital anomalies of malformations, (CAM) in Bukhara region for the period from 2021 to 2024. It was shown that these CAMs as congenital heart defect and Down's syndrome were the most common pathology among other CAMs.

**Key words:** *Down syndrome, anomaly, congenital anomalies, polydactyly, syndactyly.*

It is well known that. The Bukhara region is an important economically developed industrial region located in the arid zone of Uzbekistan, which is characterized by the presence of a sharply continental climate, a high level of insolation, a high degree of technogenic impact caused by the operation of industrial enterprises.

The complex of technogenic factors affecting the health of the local population can be attributed to the so-called "controlled risk factors", on the basis of which it seems promising to develop and implement a target program for the prevention and treatment of congenital malformations congenital anomalies in the region.

In recent years, a continuous growth of VAR has been detected in the world. An increase in the relative indicators of hereditary and congenital pathology in the structure of morbidity and mortality of newborns and young children was noted.

It is shown that more than 10% of all conceptions in the human population are accompanied by congenital anomalies of which 0.5% are chromosomal diseases, 0.7% are molecular, 1.8% are polygenic pathologies, the remaining 7% are hereditary predispositions. 4-6% of children with congenital anomalies are born annually in the world, and in many countries, fetal and newborn congenital

anomales are among the first in the structure of mortality .

In connection with the above, the study of the frequency of spread, early diagnosis, prevention and treatment of congenital anomalies is one of the urgent tasks of modern medicine.

The purpose of the work is to analyze the frequency of distribution of a number of VAR in the Bukhara region.

Material and methods of research. We conducted prospective epidemiological studies that included detailed registration of congenital anomalies in the Bukhara region. It should be noted that only based on accurate data on the frequency of occurrence of congenital anomalies in the regions, and then in Uzbekistan, it is possible in the future to judge the epidemiological picture and the frequency of occurrence of congenital anomalies , to conduct a medical examination of children with congenital anomalies and determine the required amount of pediatric and other specialized types of treatment .

Individual examination cards were developed for each child with congenital anomalies with the introduction of objective and reliable information there, starting from the beginning of the mother's pregnancy, the development of the child - to the profiling clinics, taking into account the diagnoses.

In this study, archival materials of maternity hospitals in the city of Bukhara were used .- The Bukhara region, as well as the data of the Bukhara Regional Screening Center. In maternity hospitals, all the birth histories and the history of the development of newborns for the period from 2020 to 2024 were studied and analyzed.

Results and discussion. The results of the research showed that 84,159 live children were registered in the settlements/ Bukhara region for 4 years.

The prevalence rates of 6 types of congenital anomalies were analyzed (Table). Of these, the following types of congenital anomalies were most common: congenital heart defects, Down syndrome, spinal hernia and polydactyly.

For example, the frequency of congenital heart defects was 0.48 or

The second group of VPD, namely congenital torticollis and syndactyly, occurred with a lower frequency compared to the above-mentioned congenital anomalies and according to the frequency of epidemiological prevalence in the Bukhara region, they can be conditionally attributed to the group of pathologies with a moderate nature of the spread of congenital anomalies (Table).

Congenital torticollis was registered with a frequency of 0.27 or 1 case per 3659 newborns. Syndactyly in terms of the frequency of occurrence of congenital anomalies was in last place: 0.14 or 1 : 7013.

The results obtained indicate that the epidemiological picture of the situation in the Bukhara region is unfavorable. We obtained different frequencies of congenital anomalies propagation.

Higher frequencies of the epidemiological prevalence of congenital anomalies were recorded here, in contrast to those in the Navoi region [ 2 ].

It should be emphasized that in the Bukhara region, unfavorable environmental impacts in the complex negatively affect the health of the population living here [3, 6] and make a certain "contribution" to the frequency of birth of children with congenital anomalies.

We believe that the high frequency of congenital anomalies occurrence in this region is due to environmental, hereditary and social factors.

## **REFERENCES.**

- [1]. Chernenkov Yu. V.. Nechaev V. N. Diagnosis, prevention and correction of congenital malformations. Saratov Scientific and Medical Journal, 2009; 3 (5) : 379-83..
- [2]. Inoyatov A. S. Characteristics of the frequency of occurrence of congenital

pathologies in the Bukhara region. //V1snik problemy blogs i meditsina. 2010; 1 : 238-40.

[3]. Kudryavtsev I. Yu. Analysis of the state of the oncological service in the Navoi region and the organization of anti-cancer control. Autoref. on the job. uch.step. Doctor of Medical Sciences. - Moscow. - 2008. 48 p.

[4]. Neonatology (national research)/Edited by N. N. Volodin. - M.: Academy, 2008. - 848 p.

[5]. Silbet S. M., Kronen K. Secrets of emergency pediatrics. // Translated from English; Edited by N. P. Shabalov M.: MEDpress-inform, 2006. - 480 p.

[6]]Худайкулова, Н. И., & Жумаева, Ш. Б. (2020). О стимуляции иммунитета на гиалуронидазу-фактор патогенности паразитов. In *Университетская наука: взгляд в будущее* (pp. 106-108).

[7]Аминжонова, Ч. А., & Мустафаева, М. И. (2017). Биоэкологическая Характеристика Водорослей Биологических Прудов г. Бухары. In *Экологические проблемы промышленных городов* (pp. 387-389).

[8]Жарылкасымова Г.Д. и др. Встречаемость полидефицитных состояний в пожилом и старческом возрасте // Врач-аспирант, 2007. Т. 18. № 3. С. 179-182.

[9]Наврузова Н.О. и др. Современные диагностические методы для раннего выявления заболеваний шейки матки // Доктор ахборотномаси, 2019. № 4. С. 77-82.