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Research Article

PROBLEMS AND RELEVANCE OF EARLY DIAGNOSIS AND TREATMENT OF SEVERE HEREDITARY AND ACQUIRED DISEASES IN CHILDREN

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ABSTRACT

It is known that severe diseases that occur in children manifest themselves mainly in the early periods of children's life, which causes profound disorders in the mental and physical development of children. This article discusses the relevance of orphan (rare) diseases with more than 6,000 nosologies, the need to develop timely and accurate instructions for early diagnosis, support research on treatment and prevention measures in case of defects that develop as a result of common infectious diseases, and in rare cases based on the results of scientific research by a number of scientists. It also spoke about scientific achievements in the search and introduction of affordable imported drugs with high activity for the treatment of severe diseases in children.

KEYWORDS

Liver fibrosis, cirrhosis of the liver, orphan (rare) diseases, orphan drugs, heart defects, hepalipin.

INTRODUCTION

Currently, there is an increase in the incidence of severe concomitant diseases of various etiologies in children from year to year, including rare (orphan) reproductive and increased pathologies, viral, toxic and other genital hepatitis, as a result of disorders of the nervous, cardiovascular system and severe liver complaints, which ultimately lead to an increase in cases of disability and even death [1, 2]. The group of rare diseases in children includes diseases established by the legislation of different countries with a congenital (hereditary) or acquired frequency of occurrence not exceeding a certain value, such as phenylketonuria, cystic fibrosis, Gaucher disease, Pompe disease, Prader-Willi syndrome, acute lymphoblastic leukemia, idiopathic pulmonary hypertension, non-Hodgkin's lymphoma, adrenal carcinoma, esophageal cancer, kidney cancer.

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According to the existing requirements, if the disease is assessed as a rare or rare disease in no more than 1 or 500 cases per 1 million inhabitants [3, 4]. Despite the low frequency of occurrence, there are many of them - more than 6000 nosologies - these pathologies were first adopted in 1983 in the USA in relation to diseases and medicines, the term "orphan" (orphan or rare) was adopted in the legislation "orphan preaparats Law" ("Orphan Drug Act"), and about 1600 were registered certain diseases and syndromes, as well as about 300 medicines belonging to the World Health Organization (WHO), currently about 30-40 million Europeans, 25 million North Americans suffer from rare diseases [2, 5]. Taking this into account, in our country, in accordance with the law of the Republic of Uzbekistan "On medicines and pharmaceutical activities": orphan medicines intended for the treatment of rare diseases, as well as additives, in accordance with the application, are included in the list of medical devices intended for the diagnosis and treatment of rare (orphan) diseases, and all are described in detail necessary funds. Along with the nosologies described above, the number of diseases of the cardiovascular system and liver is also growing. According to WHO, almost every year in the world, a potentially life-threatening liver infection affects 2 million people with chronic hepatitis V (HBV), and many of them are young children. This is due to the fact that the social and medical significance of this disease is due to the high prevalence of infection, the inability to extract the virus from liver tissue, accurate and complete diagnosis at early stages, as well as the presence of a latent form of infection in the human body, which has existed for a long time [6, 7]. In this regard, in particular, in our country, in order to find and put into practice cheap medicines that replace imports with high activity, research work is being carried out by scientists of the Republican Specialized Scientific and Practical Medical Center of Pediatrics on early diagnosis of various congenital and acquired severe diseases using modern diagnostic methods, as well as scientists of the Institute chemistry of plant substances to study the biological activity of substances isolated on the basis of local plants and synthetic compounds.

Periods of occurrence of various serious diseases that occur in children. While many rare diseases, such as spinal muscular atrophy, osteogenesis imperfecta, mucopolysaccharidosis, achondroplasia, begin to manifest themselves in

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the early postpartum periods or in childhood, Huntington's disease, spinal ataxia, Kaposi's sarcoma and thyroid cancer can manifest themselves in adulthood. although many diseases begin at an early age, the correct diagnosis can take many years. The severity levels of the disease also differ from patient to patient, and orphan diseases significantly worsen the quality of life of patients and shorten life expectancy. The life expectancy of a patient varies greatly depending on the nosology, i.e. some degenerative and life-threatening diseases lead to death almost immediately after birth, while others, such as phenylketonuria, with timely diagnosis and proper treatment, patients will be able to return to a normal rhythm of life [2]. Also, diseases of infectious etiology, in particular, in endemic regions, HBV infection is most often transmitted from mother to child during childbirth (perinatal transmission) or as a result of horizontal transmission (through contact with damaged blood) in the first 5 years of life. HBV infection can take acute and chronic forms, the severity of which varies from asymptomatic infection or mild disease to severe or lightning hepatitis. Infectious disease in children under the age of 5, 95% of cases of mother-to-child transmission develop chronic HBV infection [6, 7]. Thus, severe diseases that occur in children are mainly manifested in the early periods of children's life, which causes profound disorders in the mental and physical development of children.

Priorities in the field of health, as well as the main economic and social aspects. The costs of treating rare diseases decrease in different amounts depending on the size of the population and the frequency of occurrence in different mammals. This encourages pharmaceutical companies to develop new drugs. But the number of medicines produced is very small, and their cost is considered too high. For example, the cost of treating a child with mucopolysaccharidosis alone (a hereditary disease associated with a metabolic disorder that manifests itself as a "disease of accumulation or accumulation" and leads to various defects of bones, cartilage, connective tissue) in Russia will be equal to about 32.5 million rubles per year [8]. Even in our country, a presidential decree was adopted "On measures to further improve medical and social care for children with rare (ARVI) and other hereditary and genetic diseases." A registry of patients with rare hereditary and genetic diseases will be created in Uzbekistan. The resolution defines the main priorities for

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providing medical and social assistance to children with rare (orphan) and other hereditary and genetic diseases. A program of measures to organize the provision of medical and social assistance to children with rare (ARVI) and other hereditary and genetic diseases for 2019-2024 has also been approved. In accordance with the program, only 424.6 billion rubles were allocated for the diagnosis and treatment of these diseases in children. soums and 548.0 thousand soums. It has been established that the import of medical devices used for the treatment of rare (orphan) diseases is carried out without state registration in accordance with the list approved by the Ministry of Health. Until January 1, 2020, a National Registry of Patients with Rare (Orphan) and Other Hereditary genetic diseases was created. Social aspects, on the other hand, are determined: hereditary rare and increased severe diseases and the complications caused by them, as well as the need for lifelong treatment, cause inequality and vulnerability of people. It is important that health authorities, as well as interested persons working as specialists in patient health management, develop information networks that would provide them with reliable and useful information as timely and continuous care.Economic and social aspects of the above-mentioned diseases or liver diseases, as well as severe diseases of increased infectivity or other etiology, various individual assessments of the effectiveness and cost of treatment of liver diseases, which are leaders in frequency of occurrence, in particular, this imposes the need for discussion in terms of factors related to social and economic consequences.

Achievements in the study and treatment of severe diseases that occur in children. The importance of research into hereditary rare or aggravated severe diseases is to obtain valuable information about the causes of their development, which will expand the possibilities of not only their treatment, but also prevention. In recent decades, thanks to various state and public organizations, interest in such severe pathologies as orphan diseases that occur in children, infectious diseases that lead to negative consequences has increased. All this, based on scientific research, contributes to the growth of the possibilities of various programs to study the features of the development of the disease in patients with rare and severe diseases, in particular clinics, treatment and rehabilitation. Many state and public organizations support research projects that are planned to be

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implemented in this regard [9-12]. In our country, by the decisions of the President and the Government in this regard, the Ministry of Innovative Development announced scientific, practical and innovative competitions for the development of new drugs for the early diagnosis, treatment and prevention of severe hereditary and infectious diseases and funded projects that are recognized as justified. Based on this, scientists from the Institute of Plant Chemistry have been conducting scientific research on the basis of allapinin and its metabolites for many years in order to find and put into practice promising drugs against arrhythmias that develop independently or as a complication of cardiovascular pathologies in patients of different ages. Based on the conducted studies, clinical preliminary studies of the activity of the main metabolite of allapinin N-deacetylappaconitin in relation to arrhythmia, pharmacodynamic properties, effects on organs and systems with acute administration in therapeutic doses and chronic in various doses, as well as general pharmacotoxicological properties were completed [13-17]. At the same time, a comprehensive research work is being carried out to study the biological activity of substances isolated from local plant raw materials and synthetic compounds, as well as to study drugs with hepatoprotective activity against neurodegenerative diseases, epilepsy [18]. In the Republican Specialized Pediatric Scientific and Practical Medical Center, scientists intensively carry out measures for the early diagnosis of various diseases of different genera and increased severity using modern diagnostic methods. In addition, in-depth and comprehensive research work is underway to identify models of hepatitis in experimental animals, as well as combinations of hepalipine and yantazine with high hepatoprotective activity, which are obtained on the basis of local plant raw materials and synthetic compounds, including ecdystene, plant lycopene and fofolipids [19-24].

CONCLUSION

Thus, first of all, the rights of patients with these pathologies should be supported through various programs and political views. Orphan diseases are currently an urgent problem on the scale of modern healthcare and society as a whole, and understanding this problem will consist in taking care of patients and their families with rare diseases of the first group of queue, as well as carrying out the prevention of rare pathology.

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Monitoring the accuracy and regularity of criteria for prenatal and neonatal screening and various preventive tests for the purpose of early diagnosis or prevention of pathologies with a high probability of occurrence of unique diseases based on the knowledge gained about the causes of the disease, support groups should be able to inform patients in order to prevent them. constantly pondering.

Also, the result of scientific achievements in the search and practical application of promising affordable imported drugs with high activity for the treatment of severe diseases that occur in children today can be considered as a debauch of such a period as the use of local original drugs in the near future in the elimination of severe pathologies.

All of the above aspects allow us to conclude that a serious illness provides a significant improvement in the outcome of the disease and the quality of life of children.

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