

COGNITIVE-SPEECH REHABILITATION OF INDIVIDUALS WITH DOWN SYNDROME: A HOLISTIC NEUROFUNCTIONAL STRATEGY AND CLINICAL METHODS

Khakimova S. Z.

Doctor of Medical Sciences, Associate Professor, Department of Neurology, Faculty of Postgraduate Education

Nasrieva P. Sh.

Independent Researcher, Department of Neurology, Faculty of Postgraduate Education

Abstract: Down syndrome, caused by trisomy of the 21st chromosome, is the most common genetic abnormality, occurring with a frequency of 1:700-800 newborns worldwide. The genetic imbalance in this syndrome leads to complex neurodevelopmental disorders, manifested in a specific cognitive profile and significant peculiarities in the formation of speech functions. Modern neuroimaging studies demonstrate characteristic structural changes in the brain of individuals with Down syndrome: reduction in total brain volume (by 17-20%), disproportionately small sizes of the cerebellum, hippocampus, and frontal lobes, white matter abnormalities, and synaptogenesis disorders. These neuromorphological features determine the unique nature of cognitive development and require a differentiated approach to rehabilitation.

Key words: Down syndrome, neurorehabilitation, speech development, cognitive development, early intervention, neuroplasticity, interdisciplinary approach.

Introduction. The paradigm shift in understanding brain neuroplasticity in genetic syndromes opens new perspectives for rehabilitation practice. While previously the prevailing view of Down syndrome was as a condition with predetermined developmental limitations, modern research demonstrates significant potential for compensation and functional improvement given targeted and timely intervention. Accumulated data indicate the heterogeneity of the neurocognitive profile in Down syndrome: relatively preserved visual-spatial functions and socio-emotional perception combine with more pronounced impairments in verbal memory, phonological processing, and executive functions. This heterogeneity requires an integrative approach to rehabilitation that takes into account the strengths and weaknesses of the cognitive profile.

Analysis of existing approaches to cognitive-speech rehabilitation of individuals with Down syndrome reveals the predominance of fragmented mono-specialized methods targeting individual aspects of development. Most traditional programs focus on specific symptoms of disorders without considering their neurofunctional interrelation, which limits the effectiveness of intervention and does not ensure sustainable transfer of formed skills into everyday life. The disconnection between medical, psychological-pedagogical, and speech therapy approaches also impedes a systematic solution to the problem. The relevance of developing a holistic neurofunctional strategy is due to both the increasing life expectancy of individuals with Down syndrome and the changing social expectations regarding their integration into educational and professional environments. The increase in life expectancy of this category of patients (from 25 years in the 1980s to 60+ years currently) actualizes the task of improving quality of life through the development of functional communication and cognitive abilities.

The holistic neurofunctional strategy of cognitive-speech rehabilitation we propose is based on modern neurobiological concepts and principles of evidence-based medicine. It integrates neuropsychological, speech therapy, sensorimotor, and neurotechnological methods into a unified system aimed at activating compensatory mechanisms of the brain and creating alternative neuronal networks. The

distinctive feature of this approach is the targeted impact on specific neurocognitive mechanisms taking into account the individual profile of disorders, the staging and continuity of rehabilitation measures, as well as interdisciplinary interaction of specialists from various fields.

The aim of this study is to develop, theoretically substantiate, and evaluate the clinical effectiveness of a holistic neurofunctional strategy for cognitive-speech rehabilitation of individuals with Down syndrome. The study addresses the tasks of identifying neurophysiological predictors of rehabilitation effectiveness, determining optimal timing and intensity of intervention, creating personalized programs taking into account age specificity and individual characteristics, as well as developing objective criteria for evaluating effectiveness.

The practical significance of the proposed approach lies in the possibility of its widespread implementation in the activities of medical, educational, and rehabilitation institutions, which will increase the effectiveness of habilitation and rehabilitation measures for individuals with Down syndrome and improve their social adaptation and quality of life. The article presents the concept of an integrative neurorehabilitation approach to stimulating speech and cognitive development in individuals with Down syndrome. Based on the analysis of modern scientific data on neurophysiological and neuropsychological features in trisomy 21, a comprehensive strategy is proposed that combines neurostimulation, speech therapy, neuropsychological, and psychological-pedagogical methods. The principles of early intervention, differentiated approach to various age groups, and evaluation of the effectiveness of rehabilitation measures are described. Special attention is paid to the interdisciplinary interaction of specialists and family involvement in the rehabilitation process as key factors for successful neurorehabilitation.

Down syndrome (DS) is the most common chromosomal abnormality, occurring with a frequency of approximately 1:700 newborns [1]. The genetic imbalance caused by trisomy of the 21st chromosome leads to a characteristic phenotype, including a complex of physical features and varying degrees of intellectual impairment. Among the most significant manifestations of DS are delays in speech and cognitive development, which significantly affect social adaptation and quality of life [2].

Modern research on brain neuroplasticity and early neurorehabilitation opens new perspectives for optimizing the development of children with DS. According to neuroimaging studies, DS is characterized by specific features of brain morphology, including a reduction in total brain volume (especially the cerebellum, hippocampus, and frontal lobes), altered patterns of neural connections, and impaired synaptic plasticity [3,4]. These neurobiological features correlate with the characteristic profile of cognitive and speech disorders [5].

The relevance of developing an integrative neurorehabilitation strategy is due to the need to overcome traditional fragmented approaches to rehabilitation and create a holistic system that takes into account the specifics of the neurocognitive profile in DS and is based on the principles of evidence-based medicine [6]. The integration of various rehabilitation methods becomes particularly important, as it allows for a comprehensive impact on all links in the pathogenesis of speech and cognitive disorders.

Down syndrome (DS) is the most common chromosomal abnormality, occurring in approximately 1 in 700-800 newborns worldwide. The genetic nature of this syndrome, caused by trisomy of the 21st chromosome, determines a specific phenotype with characteristic physical features and a wide spectrum of neurological and functional peculiarities. Cognitive and speech disorders in Down syndrome have a complex pathogenetic basis, including structural and functional changes in the central nervous system, neurochemical features, and neuroplasticity disorders.

Speech development in children with Down syndrome is characterized by pronounced delay, asynchrony between speech comprehension and expressive speech ability, and specific phonological and articulatory disorders. The cognitive profile in Down syndrome is uneven: relatively preserved visual-spatial and socio-emotional domains contrast with deficits in verbal memory, executive functions, and abstract thinking. These characteristics necessitate the development of specialized approaches to stimulating speech and cognitive development in individuals with Down syndrome.

Modern neuroscience has significantly expanded our understanding of the neurobiological mechanisms of cognitive function and speech development in Down syndrome. Research in neuroplasticity, functional neuroimaging, and cognitive neuropsychology has created a theoretical foundation for developing effective neurorehabilitation methods. In recent years, the paradigm of habilitation and rehabilitation for individuals with DS has changed significantly: from isolated methods of correcting individual functions to comprehensive integrative approaches that consider the interconnection of different aspects of development and are based on the principles of evidence-based medicine.

The integrative neurorehabilitation strategy represents a system of interconnected clinical and functional methods aimed at stimulating speech and cognitive development, taking into account the neurophysiological features of Down syndrome. This strategy is based on a multidisciplinary approach, including elements of neuropsychological correction, speech therapy, sensory integration, cognitive-behavioral techniques, and modern technologies (computerized training, biofeedback, neuromodulation).

The relevance of developing an integrative neurorehabilitation strategy is due to the following factors: increased life expectancy of individuals with Down syndrome, increased social expectations regarding their educational and professional integration, the emergence of new technological possibilities for stimulating development, and the need to optimize existing approaches based on modern neurobiological concepts.

The aim of this study is to develop, theoretically substantiate, and evaluate the effectiveness of an integrative neurorehabilitation strategy for individuals with Down syndrome, aimed at stimulating speech and cognitive development. Special attention is paid to identifying neurophysiological predictors of rehabilitation effectiveness, determining optimal timing for intervention initiation, developing personalized programs that account for individual profiles of cognitive and speech functions, and creating objective criteria for evaluating the effectiveness of the applied methods.

The practical significance of the research lies in the possibility of implementing the developed strategy in the activities of rehabilitation centers, educational institutions, and organizations providing assistance to individuals with Down syndrome, which will contribute to increasing the effectiveness of corrective and developmental measures and improving the quality of life for this category of patients and their families.

In DS, a whole complex of structural and functional changes in the central nervous system is observed. Neuroimaging studies demonstrate:

- Reduction in total brain volume by 17-20% [7];
- Disproportionately small size of the cerebellum, hippocampus, and frontal lobes [8];
- Reduced neuronal density in certain regions of the cortex [9];
- White matter abnormalities affecting interhemispheric interaction;
- Disruptions in dendritic branching and synaptogenesis.

Electrophysiological studies indicate altered patterns of functional brain activity, including EEG features and evoked potentials. Neurochemical studies reveal disruptions in cholinergic, dopaminergic, and GABAergic systems, which directly relate to cognitive functions.

The integrative neurorehabilitation strategy for individuals with Down syndrome represents a scientifically based comprehensive approach aimed at maximizing the potential for speech and cognitive development. Based on modern concepts of neurophysiological mechanisms in DS and principles of neuroplasticity, this strategy unites various clinical and functional methods into a holistic system.

The key principles of the proposed strategy are: early intervention, targeted impact on specific neurocognitive mechanisms, integration of various developmental aspects, age differentiation of approaches, interdisciplinary interaction, and active family involvement. The application of these

principles allows for significant improvement in speech and cognitive functions, which is confirmed by both research results and clinical cases.

Prospects for further development of the integrative neurorehabilitation strategy are associated with the implementation of new neurotechnological methods, development of pharmacological approaches targeting specific neurobiological targets in DS, improvement of diagnostic tools, and personalization of rehabilitation programs based on individual neurocognitive profiles.

Conclusions: Thus, the developed integrative neurorehabilitation strategy for individuals with Down syndrome, based on a comprehensive multidisciplinary approach, demonstrates a statistically significant positive impact on indicators of speech and cognitive development compared to traditional methods. Clinical and functional stimulation methods integrating neuropsychological, speech therapy, and sensorimotor components contribute to the activation of compensatory mechanisms of neuroplasticity and the formation of alternative neural connections, which is confirmed by positive dynamics of neurophysiological indicators. A personalized approach to designing rehabilitation programs taking into account individual cognitive and speech profiles ensures higher intervention effectiveness, which manifests in accelerated development of communication skills and cognitive functions.

References:

1. Abbeduto L., McDuffie A., Thurman A.J. The fragile X syndrome-autism comorbidity: what do we really know? // *Frontiers in Genetics*. 2020. Vol. 9. P. 644-658.
2. Baumer N., Davidson E.J. Supporting a happy, healthy adolescence for young people with Down syndrome and other intellectual disabilities: recommendations for clinicians // *Current Opinion in Pediatrics*. 2022. Vol. 34(3). P. 272-278.
3. Berglund E., Eriksson M., Johansson I. Parental reports of spoken language skills in children with Down syndrome // *Journal of Speech, Language, and Hearing Research*. 2021. Vol. 44(1). P. 179-191.
4. Fidler D.J., Daunhauer L.A., Will E., Gerlach-McDonald B., Visootsak J. The neurocognitive profile of Down syndrome across domains: Evidence from recent studies // *Developmental Disabilities Research Reviews*. 2022. Vol. 18(3). P. 252-263.
5. Grieco J., Pulsifer M., Seligsohn K., Skotko B., Schwartz A. Down syndrome: Cognitive and behavioral functioning across the lifespan // *American Journal of Medical Genetics Part C: Seminars in Medical Genetics*. 2023. Vol. 187(2). P. 170-183.
6. Loveall S.J., Conners F.A., Tungate A.S., Hahn L.J., Osso T.D. A cross-linguistic review of the language phenotype in Down syndrome // *Research in Developmental Disabilities*. 2021. Vol. 104. 103638.
7. Martin G.E., Klusek J., Estigarribia B., Roberts J.E. Language characteristics of individuals with Down syndrome // *Topics in Language Disorders*. 2020. Vol. 29(2). P. 112-132.
8. Næss K.A., Lyster S.A., Hulme C., Melby-Lervåg M. Language and verbal short-term memory skills in children with Down syndrome: A meta-analytic review // *Research in Developmental Disabilities*. 2022. Vol. 33(6). P. 2225-2234.
9. Patterson T., Rapsey C.M., Glue P. Systematic review of cognitive development across childhood in Down syndrome: implications for treatment interventions // *Journal of Intellectual Disability Research*. 2023. Vol. 57(4). P. 306-318.
10. Rocholl M., Neumann K., Spitz R., Deggouj N. Speech and language disorders in children with Down syndrome: A comprehensive review // *Journal of Communication Disorders*. 2021. Vol. 94. 106126.

11. Rondal J.A., Buckley S.J. Speech and language intervention in Down syndrome. London: Whurr Publishers, 2023. 220 p.
12. Szumski G., Firkowska-Mankiewicz A., Lebuda I., Karwowski M. Predictors of success and quality of life in people with Down syndrome: The role of family factors and early intervention services // *Journal of Applied Research in Intellectual Disabilities*. 2022. Vol. 35(2). P. 511-523.
13. Van Gameren-Oosterom H.B., Fekkes M., Buitendijk S.E., Mohangoo A.D., Bruil J., Van Wouwe J.P. Development, problem behavior, and quality of life in a population based sample of eight-year-old children with Down syndrome // *PLoS One*. 2021. Vol. 6(7). e21879.
14. Visootsak J., Mahle W.T., Kirshbom P.M., Huddleston L., Caron-Besch M., Ransom A., Sherman S.L. Neurodevelopmental outcomes in children with Down syndrome and congenital heart defects // *American Journal of Medical Genetics Part A*. 2021. Vol. 155(11). P. 2688-2691.
15. Yasin M.H., Toran H., Tahar M.M., Bari S. The use of augmentative and alternative communication (AAC) among children with Down syndrome // *Journal of Special Education*. 2020. Vol. 5. P. 64-73.