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8.1 Using neuro-headsets to diagnose genetic diseases

A neuroheadset (Fig. 1) is a portable device that records brain activity or related physiological signals for the analysis of cognitive state, emotional reactions, neuromuscular activity or for interaction with digital systems (brain-computer interfaces). It usually consists of sensors (electrodes or optical sensors), a transmitting module and software that processes signals in real time. Such devices are used in medicine, rehabilitation, sports training, psychophysiological monitoring, game interfaces and research.

Fig. 1 External view of the neuroheadset [165]

The most common are EEG neuroheadsets [166-177] , which record the electrical activity of the cerebral cortex using dry or gel electrodes; they are both consumer (4–16 channels) and professional (32–256 channels). Optical neuroheadsets fNIRS use near-infrared radiation to measure changes in cerebral blood flow and assess cognitive load in real time. A separate group is EMG and EOG headsets , which record peripheral neurosignals - muscle activity or eye movements, and are used in combined control systems or for users with motor disorders. Invasive neuroheadsets are also used in medical and research centers , which read impulses directly from the cerebral cortex through implanted electrodes; they provide the highest accuracy, but require surgical intervention. In recent years, hybrid neuroheadsets have been rapidly spreading , combining EEG, fNIRS, EMG, and other sensors to improve signal reliability. Commercial and gaming BCI headsets are focused on interaction with digital content,

meditation, concentration training and use in VR/AR environments. All these types form a modern spectrum of technologies that are gradually moving from experimental devices to tools for practical medicine and everyday use.

The global neuroheadset market in 2024–2025 is showing stable growth within 12–15% annually , which is associated with the development of BCI, neurotraining and remote medicine. According to open analytical reviews, the total volume of the global neurotechnology market in 2024 is estimated at 17–19 billion dollars , and the segment of portable neuroheadsets is about 1.5–2 billion dollars with a forecast of growth to 5 billion by 2030. EEG devices have the largest share (over 55%) , while fNIRS and hybrid systems are growing the fastest (20–25% annually). Key manufacturers - Emotiv, NeuroSky, OpenBCI, NextMind (Snap), g.tec, Bitbrain, Muse - actively expand product lines, adding multi-channel systems and AI algorithms. The consumer segment (gaming, meditation, fitness) covers over 40% of sales, but the medical direction is growing most dynamically - neurorehabilitation, monitoring of cognitive disorders, diagnostics of neurodegenerative and genetic diseases.

Over the past decade, biomedical engineering has undergone a significant technological shift associated with the development of portable sensor devices, in particular neuroheadsets [165-169], capable of recording electrical, magnetic and optical brain activity. Until now, such systems have been mostly associated with brain-computer interfaces, cognitive function studies, rehabilitation of patients after strokes or neuromarketing. However, the gradual complication of sensor architecture, increasing the accuracy of biosignal registration and integration of deep learning algorithms have opened up the possibility of using neuroheadsets in areas that were previously considered exclusively the prerogative of laboratory genetic methods. One such area is the early diagnosis of genetic diseases, in which the analysis of brain activity can reveal characteristic patterns associated with mutations, disorders of neuronal myelination, changes in synaptic transmission and deficiency of neurotransmitters.

Traditionally, genetic diagnostics is based on molecular approaches - DNA sequencing, the study of chromosomal aberrations, the search for mutations, the

analysis of parental genomes or biochemical markers. These approaches are highly accurate, but they do not answer the question of how exactly a genetic defect manifests itself in the functioning of the nervous system at the level of electrophysiology . That is why neuro-headsets are gradually being integrated as an auxiliary tool for clarifying the clinical picture, monitoring the dynamics of the disease and searching for phenotypic markers that are difficult to observe by other methods. The emergence of highly sensitive dry electrodes and optical sensors has made it possible to regularly monitor brain activity without the need to use bulky laboratory equipment.

Among the genetic conditions that can exhibit specific patterns of brain activity, Down syndrome, Rett syndrome, fragile X syndrome, phenylketonuria, and some variants of epileptic encephalopathies are most often considered. Many of these disorders are accompanied by changes in the formation of the cortex, impaired neuronal plasticity, and reduced levels of certain enzymes, which directly affect electrical processes in the brain. Therefore, neuroheadsets can serve as an additional tool for identifying functional biomarkers that reflect the genetic defect not as a molecular phenomenon, but as a consequence of its systemic action.

Current consumer and clinical neuroheadset models vary in sensitivity, but they all rely on several traditional methods: electroencephalography (EEG), functional near-infrared spectroscopy (fNIRS), portable magnetoencephalography, and combined approaches where electrical signals are supplemented with data from motion sensors, oculography, or cardiac activity. The use of multimodal systems reduces error and allows for clearer markers. For example, generalized slow waves are characteristic of Rett syndrome. and a pronounced decrease in alpha rhythm, which can be detected even by small-channel devices. In fragile X syndrome, typical changes are manifested in the form of increased theta activity and asynchrony between the hemispheres, which is also recorded by portable EEG.

Special attention is paid to early diagnostic technologies in newborns. Not all genetic diseases manifest themselves immediately after birth, and some of them can be detected only by indirect signs - sleep disorders, frequent myoclonus, deviations in sensory reactivity. Portable neuro-headsets for infants, which use soft electrodes and

low-intensity optical sensing, practically do not cause discomfort and can be used for long-term monitoring at home. The use of algorithms for analyzing rhythm variability, assessing channel synchronization, spectral density and nonlinear characteristics allows detecting abnormal neural patterns associated with genetic disorders of metabolism, myelin sheath or synaptic transmission.

A powerful direction is the use of neuro-headsets in combination with machine learning. Neural networks trained on large samples of biosignals are able to notice complex and subtle changes that a human expert cannot assess without high-precision equipment. For example, deep convolutional networks detect atypical ratios of alpha and theta rhythms, asymmetry of temporal patterns, or imbalances in neuronal ensembles that are inherent in certain genetic mutations. In some experiments, classification accuracy reached 85–92%, which is a significant indicator for non-invasive methods. Although this level does not yet allow replacing molecular tests, it enhances comprehensive screening and significantly accelerates the start of therapy.

In practice, neuro-headsets are valuable for dynamic monitoring of patients in whom genetic disorders affect cognitive development. Doctors can record changes in the response to sensory stimuli, assess the stability of attention, the speed of formation of neural responses and patterns of brain excitation. For example, in children with phenylketonuria, impaired metabolism of phenylalanine causes changes in the structure of white matter, which is reflected on the EEG as an increase in slow waves. Regular measurements using neuro-headsets allow us to assess how effective dietary therapy is and whether there is an improvement in neurophysiological indicators.

In addition to diagnosis, certain types of genetic diseases can manifest themselves in the form of specific patterns of emotional regulation, sleep disorders, or stress reactions. Neuroheadsets that simultaneously record EEG and cardiac variability allow you to capture complex correlations between the central and autonomic nervous systems. Some chromosomal abnormalities are characterized by sleep phase disorders, shifts in REM periods, and a decrease in the depth of the delta phase. Such features can help doctors differentiate genetic pathologies from purely behavioral or psychoemotional conditions.

Research in the field of digital medicine shows that neuro-headsets can serve as an important tool for early diagnosis , when identifying risk patterns is carried out even before the appearance of external symptoms. This is especially important for conditions that develop gradually: neurodegenerative disorders, mitochondrial diseases, hereditary epilepsies. In such cases, early detection of characteristic changes can help to start therapy at a preclinical stage, when the brain structure has not yet suffered significant damage.

A separate direction has been the use of portable neuro-headsets in studies of behavioral phenotypes associated with genetic mutations. Many genetic conditions have hidden, mild manifestations that are not always monitored by doctors. Some mutations affect the regulation of emotions, the speed of neural adaptation, and the formation of motor circuits. Neuro-headsets help collect quantitative data that allow us to assess how pronounced certain features are compared to normative values. This approach provides more objective diagnostics and helps to justify the need for genetic counseling.

The effectiveness of neuro-headsets depends on the precise modeling of test protocols. For each genetic disease, special cognitive tasks, sensory stimuli or motor responses can be created that evoke a characteristic neural response. For example, in fragile X syndrome, adaptation to repetitive sound stimuli is impaired - this can be detected by changes in the amplitude of sound-evoked potentials. In Down syndrome, changes in the speed of sensory integration are typical, which are also recorded on the EEG. Thus, the neurophysiological profile can act as a marker of the genetic condition.

Modern neuro-headsets also allow for the analysis of nonlinear characteristics of signals: entropy, fractal indices, mutual information, correlation dimensions. Such parameters are especially valuable, since many genetic diseases cause dysfunctions of neural networks at the level of organization, which is not always manifested in the classical spectrum of rhythms. For example, disorders associated with myelin disruption can cause chaotic changes in the duration of neural impulses and asymmetries between channels. Nonlinear analysis allows for the detection of such changes before they become noticeable at the behavioral level.

Engineering innovations in the creation of neuro-headsets contribute to improving the quality of diagnostics. Thanks to flexible electrodes with nanostructured surfaces, it has become possible to record even weak potentials without using gel or paste. New generation optical sensors provide greater accuracy in the analysis of blood supply to the brain, which is important for studies of metabolic disorders associated with genetic changes. Multi-channel systems are now equipped with real-time processors, which allows for primary signal processing directly on the device, reducing the amount of data transmitted to the cloud.

An important factor in development is the availability of portable devices, thanks to which neurophysiological studies can be conducted in schools, rehabilitation centers, behavioral genetics laboratories, as well as at home. Comfortable neuroheadsets for the user allow for long-term data collection, which significantly expands the possibilities of creating predictive models. For example, for some genetic syndromes it is important to assess how brain activity changes during the day or during fatigue. Long-term monitoring helps to detect functional fluctuations that cannot be recorded with a single examination in the clinic.

A particularly promising direction is the integration of neuro-headsets with personalized biomedical platforms, in which genetic data, metabolic test results, and neurophysiological indicators are analyzed in a complex. This approach allows the creation of digital phenotypes, which are detailed individual profiles that reflect the interaction of genotype and neural dynamics. In the future, such profiles may help predict the risks of developing diseases, assess the effectiveness of therapy, and develop personalized recommendations for each patient.

Given the rapid development of technologies, the emergence of hybrid neuroheadsets based on a combination of EEG, ECG modeling, optical sensors and micronavigation systems capable of recording spatial characteristics of brain activity is expected in the coming years. This will allow obtaining even more accurate data on the operation of neural networks and identifying specific patterns associated with rare genetic mutations.

EEG (electroencephalography) is a non-invasive method of recording the electrical activity of the brain, which measures fluctuations in electrical potentials that occur in cortical neurons.

EEG can be recorded using a neuroheadset, since most modern portable systems are built on the principle of electroencephalography and contain dry or gel electrodes that record the electrical activity of the cerebral cortex. Such devices usually have fewer channels than clinical equipment, but they are able to qualitatively record the main rhythms - delta, theta, alpha, beta and gamma, as well as track spectrum changes associated with attention, stress, cognitive loads or certain neurophysiological abnormalities. Neuroheadsets allow for long-term home monitoring, are used in BCI systems, neurotraining, rehabilitation and preliminary diagnosis of certain disorders. The limitation is lower accuracy and spatial resolution due to a smaller number of electrodes and sensitivity to artifacts, however, for most applied tasks, including general screening, training, research, and assessment of functional brain states, a portable neuroheadset provides a sufficient level of information and is an effective alternative to stationary EEG systems.

Table 1. Characteristic EEG frequencies in genetic diseases

Genetic disease	Characteristic frequency changes in EEG	Typical reasons for changes
Rett syndrome (MECP2)	• Delta 1–3 Hz ↑ • Theta 4–6 Hz ↑ • Alpha 8–10 Hz ↓ • Periodic slow waves	Impaired synaptic plasticity and cortical integration
Down syndrome (Trisomy 21)	• Slow waves 2–6 Hz ↑ • Alpha 8–12 Hz ↓ • Late sensory responses 3–7 Hz	Delayed myelination, impaired neural integration
Fragile X syndrome (FMR1)	• Theta 4–8 Hz ↑ • Delta 1–3 Hz ↑ • Beta 13–20 Hz ↓ • Hemispheric asymmetry	Disturbances in GABAergic transmission and neuronal homeostasis
Phenylketonuria (PKU)	• Delta/theta 1–7 Hz ↑ • Alpha 8–12 Hz ↓ • Decrease in alpha peak frequency	Phenylalanine neurotoxicity, white matter degeneration

Continuation table 1

MELAS/MERRF (mitochondrial)	• Delta 1–4 Hz ↑ • Episodes of high-frequency rhythms 20–30 Hz • Alpha disorganization	Neuronal energy deficiency, damage to the cortex and subcortex
Tuberous sclerosis (TSC1/TSC2)	• Local delta 0.5–4 Hz ↑ • Slow background 5–8 Hz • Theta synchronization	Cortical tubers, developmental disorders of neural tissue
Duchenne/Becker Dystrophy	• Theta 5–7 Hz ↑ • Beta 13–20 Hz ↓ • Alpha variable	Dystrophin deficiency in the CNS, changes in subcortical structures
Dravet (SCN1A mutation)	• Theta 4–7 Hz ↑ • Delta 1–3 Hz in drowsiness • High-frequency spikes 30–80 Hz	Sodium channel dysfunction, epileptic hyperexcitability
CDKL5-encephalopathy	• Deep delta 0.5–2 Hz ↑ • Theta 4–6 Hz ↑ • Rhythm disorganization	Impaired formation of cortical networks in infants
Lennox–Gastaut (often genetic in nature)	• Slow spikes 1–2.5 Hz • Hypersynchronous beta 20–30 Hz	Severe diffuse cortical disorders, polygenic mutations
Wilson's disease (ATP7B)	• Slowed alpha rhythm 6–8 Hz • Theta 5–7 Hz ↑ • Discoordination of hemispheres	Basal ganglia damage due to copper accumulation
Lysosomal diseases (GM1, Niemann–Pick)	• Delta 1–3 Hz ↑ • High frequencies 10–30 Hz ↓ • Slow recovery after stimulus	Neurodegeneration due to enzyme defects

An increase in delta activity in the range of 1–3 Hz indicates the dominance of the slowest brain oscillations, which in a healthy adult appear mainly during deep sleep. When such waves are recorded in a state of wakefulness, this indicates a slowdown in neural transmission, insufficient maturity of cortical networks or their functional disorders. An increase in the theta rhythm of 4–6 Hz usually means a decrease in the level of cortical activation and the predominance of immature or insufficiently organized neural networks, since theta waves are characteristic of childhood, drowsiness and transitional states between wakefulness and sleep. Together with delta, increased theta activity indicates a reduced ability of the cortex to effectively integrate information and maintain sustained attention. A decrease in the power of the alpha

rhythm of 8–10 Hz, which is normally the leading background rhythm of the brain during a calm state with closed eyes, means a violation of synchronization between neuronal populations and a deterioration in the functional organization of the cortex. This condition is observed in various genetic and metabolic abnormalities that affect neuronal plasticity and interhemispheric communication. The presence of periodic slow waves, which are regularly repeated in the range of 1–4 Hz, indicates generalized cortical dysfunction, reduced speed of interneuronal communication and possible structural or metabolic damage to brain tissue. The combination of these signs reflects a slowed, poorly organized brain function and is characteristic of a number of genetic, neurometabolic and neurodegenerative disorders.

Neuroheadsets are actively used in the study and monitoring of genetic diseases, as they allow non-invasive recording of brain activity and detection of characteristic disorders of the cortical networks. For example, in Rett syndrome, neuroheadsets record a pronounced increase in slow waves, a decrease in alpha rhythm, and uneven synchronization between hemispheres, which makes it possible to detect deterioration even before the appearance of external symptoms. In children with Down syndrome, portable EEG helps to assess the delay in sensory response and the level of cortical maturity, which is important for planning early development programs. In phenylketonuria, such devices allow you to regularly monitor changes in brain activity at home and observe how the body responds to diet therapy - a decrease in slow waves on the EEG is often a sign of improved metabolism. In patients with fragile X syndrome, neuroheadsets are able to detect excessive theta activity and impaired brain synchronization during attention tests, which helps to more accurately assess the degree of cognitive impairment. In mitochondrial diseases such as MELAS or MERRF, portable EEG records episodes of sharp slowing of brain activity, which allows for long-term monitoring and determination of the frequency of crisis states. In the case of tuberous sclerosis, neuroheadsets help to localize areas of excessive slow activity associated with cortical tubers, which is useful for rehabilitation programs. In Wilson's disease, changes in theta and alpha rhythms recorded by the neuroheadset may indicate early damage to subcortical structures, even before the onset of severe symptoms. In

combination with cognitive tests, neuroheadsets allow to assess how brain activity changes during task performance, forming an individual neurophysiological profile of a person with genetic disorders. In addition, they are used for remote monitoring, when parents can record EEG at home and transmit data to a doctor, which is especially important for children with rare pathologies. In scientific research, neuroheadsets are used to create digital phenotypes - large databases of brain activity for specific mutations, which helps to discover new biomarkers and develop personalized medicine. Collectively, these examples demonstrate that portable neuroheadsets are already a valuable tool for understanding and controlling genetic diseases and continue to play an important role in modern neuromedicine.

The prospects for further development of neuro-headsets for the diagnosis of genetic diseases are associated with the complex evolution of sensor technologies, signal processing algorithms and digital medical platforms. In the coming years, a significant expansion of the functionality of portable systems is expected, which will allow them to be used not only as a tool for primary monitoring, but also as part of full-fledged clinical protocols.

The use of neuro-headsets for analyzing pathological fluctuations of brain activity encompasses a much wider spectrum of indicators than basic frequency characteristics. At the current stage, clinical research focuses on microstate phenomena, phase synchronization, cross-frequency interactions (CFC), spectro-temporal shifts, and local perturbations that may be sensitive to the mutational profile of specific genes.

In patients with myelination disorders typical of many rare genetic syndromes, a decrease in alpha-rhythm coherence between parietal and occipital regions is observed. This phenomenon is explained by the dispersion of impulse conduction velocity in demyelinated axons. In response to simple cognitive stimuli, extended latencies of P1, N1, and P300 potentials are also recorded, indicating slowed sensory integration. Even low-channel neuro-headsets are capable of detecting such changes when recognition algorithms operate with high temporal resolution.

Cross-frequency correlations are of particular importance, as they are frequently disrupted in cases of mutations affecting synaptic transmission. For example, patients

with SCN2A or CACNA1A mutations demonstrate abnormal interaction between gamma oscillations (30–80 Hz) and theta rhythms — a phenomenon that may be informative for early prediction of epileptic crises. New-generation neuro-headsets can already detect these patterns through increased sampling rates.

Expanding the functionality of portable systems allows their use in various clinical scenarios. Screening protocols are applied for the primary assessment of newborns and young children. Here, the ability to perform continuous long-term recording is essential, as it enables the detection of spontaneous myoclonic episodes or sleep abnormalities. Studies show that in the case of metabolic genetic disorders, the first deviations in EEG may appear precisely during transitions between sleep phases.

Monitoring disease progression is crucial for conditions such as phenylketonuria, MELAS, tuberous sclerosis, or Wilson disease. Regular measurements reveal changes in spectral power or interhemispheric synchronization in response to dietary therapy, medication protocols, or cognitive rehabilitation. Portable neuro-headsets make it possible to create individualized dynamic curves of a patient's neurophysiological state — something that was previously achievable only in laboratory settings.

Assessment of therapeutic effectiveness is also important. For example, when L-DOPA is administered to patients with dystonia or certain neurometabolic disorders, it is possible to evaluate the reduction of pathological theta activity. During physiotherapy or cognitive-behavioral treatment, neuro-headsets enable detection of restored alpha-rhythm stability and improved brain reactivity to sensory stimuli.

The development of artificial neural networks drives a transition from classical spectral analysis to complex computational models. Among the most effective approaches are:

• deep convolutional neural networks (CNN) for recognizing spatial-frequency spectrum maps;

• recurrent networks (LSTM, GRU) for identifying temporal dependencies;

• graph neural networks (GNN) for modeling inter-channel synchrony;

• autoencoders and variational autoencoders for detecting latent patterns unobservable through classical analysis.

Recent studies demonstrate that combining deep CNN architectures with GNNs enables classification accuracy of 90–94% in tasks involving recognition of rare genetic pathologies based on EEG profiles. These results pave the way for fully automated screening platforms that can operate in primary-care clinical settings.

Special attention is paid to developing models capable of functioning on small datasets, since many genetic syndromes are rare. Transfer-learning techniques are applied in such cases: a neural network pretrained on general EEG data is fine-tuned on a small cohort of patients with a specific disorder. This significantly improves accuracy while minimizing the number of real clinical recordings required.

In the coming years, multifunctional systems combining the following are expected to appear:

• high-frequency EEG electrodes with nanostructured coating;

• hybrid optoelectrical modules with high sensitivity to changes in cerebral blood flow;

• micro-inertial sensors for analyzing subtle head micro-movements and coordination;

• graphene and polymer electrodes providing high conductivity and low impedance;

• edge processors for local signal processing and reduced dependence on cloud systems.

The combination of different sensing channels will enable the construction of three-dimensional models of brain activity in real time. Integrated with sLORETA methods and neural inverse modeling, such systems will bring portable neuro-headsets closer to the capabilities of laboratory-grade neurophysiology.

A technically challenging yet highly promising task is the creation of completely wireless systems with autonomous energy support — for example, using nanogenerators that convert micromovements or body heat into electrical energy. This will allow long-term monitoring for patients with severe genetic pathologies without interruption.

The expanding capabilities of neuro-headsets in diagnosing genetic disorders raise important ethical questions. Specifically, the storage of large biosignal databases requires specialized confidentiality protocols; EEG pattern analysis may reveal abnormalities even before a genetic diagnosis is confirmed, necessitating a careful approach when informing families; autonomous systems must comply with international safety standards, including IEEE as well as FDA/EMA medical regulations.

A particularly important aspect is avoiding misinterpretation, since EEG is a functional rather than a genetic test. Therefore, the results obtained using neuro-headsets should be applied only as part of a comprehensive model, and not as a standalone diagnostic tool.

One of the prospects is the creation of high-precision hybrid neuro-headsets that will combine EEG, fNIRS, EOG, EMG, micro-accelerometers and optical sensors. Combining such channels will provide more reliable identification of specific neurophysiological patterns characteristic of genetic mutations, as well as reduce the influence of motion artifacts and environmental noise.

Such systems can become the basis for remote clinical monitoring, when patients with rare genetic syndromes are under constant observation of doctors without the need to frequently visit medical institutions.

A promising direction is the development of machine learning algorithms capable of analyzing large arrays of biosignals and forming individual digital phenotypes. The growth of databases of EEG patterns in various genetic disorders will allow the creation of prediction models that can identify disease risks even before clinical manifestation.

Of particular importance will be the use of deep neural networks capable of recognizing complex nonlinear dependencies that often accompany mutations associated with neuronal myelination or disruption of synaptic activity.

Another promising direction is the development of neuro-headsets for use in young children, including newborns. The use of soft electrodes, sensors with low-intensity optical radiation and flexible materials makes long-term monitoring possible without discomfort for the child. This opens the way to the creation of programs for

early diagnosis of hereditary disorders of metabolism, myelination and development of neural networks at stages when traditional methods are still ineffective.

Technologies based on the integration of bioelectrical, optical and behavioral data into single multi-structural models have significant potential. Combining information on brain activity, sleep characteristics, motor responses, emotional responses and metabolic indicators can provide a deeper understanding of how a particular genetic defect affects the functioning of the nervous system. This will contribute to more accurate patient stratification and selection of personalized treatments.

In the long term, neuro-headsets could become part of comprehensive biomedical platforms that integrate genetic data, biochemical markers, and neurophysiological profiles of each patient, allowing for dynamic models of disease progression and prediction of the body's response to therapeutic interventions.

In general, the prospects for the development of neuro-sets in the diagnosis of genetic diseases indicate the possibility of forming a new level of personalized medicine, where monitoring of brain activity will become an integral part of clinical practice, and timely detection of pathological changes will be the basis for effective treatment and improving the quality of life of patients.