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ABSTRACT

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GENOME SEQUENCING IN NEWBORNS: NEW HORIZONS IN NEONATAL SCREENING IN UKRAINE

The rapid evolution of genomic technologies has fundamentally revolutionized the landscape of neonatal screening, offering unprecedented opportunities for the early detection of a wide range of hereditary and rare diseases. This transformative shift moves beyond traditional screening methods, which are often limited to a small number of conditions, to offer expansive diagnostic capabilities. Genome sequencing allows for the simultaneous analysis of hundreds, if not thousands, of genes, enabling the detection of a vast array of genetic disorders from a single test.

This article examines the current status and future prospects of integrating this advanced technology into neonatal screening programs, with a particular focus on the unique challenges and opportunities within the Ukrainian healthcare system. It provides a comprehensive overview of the clinical advantages, including the ability to provide early and precise diagnoses. This early insight is crucial for implementing personalized medical interventions and beginning targeted treatments immediately after birth, which can significantly improve health outcomes and the quality of life for affected children.

While the clinical benefits are immense, their implementation is not without significant ethical, legal, and social challenges. Key concerns include ensuring genuine informed consent, as parents must make complex decisions about their child's future genetic information, and guaranteeing comprehensive data privacy to protect highly sensitive genomic data from misuse. Furthermore, addressing equitable access is paramount to prevent the creation of a two-tiered system where only a privileged few can benefit from these life-saving technologies. Consequently, the advancement of the genetic component in neonatal screening must be accompanied by the establishment of a clear ethical and legal framework. This framework

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should not only address technical and clinical standards but also ensure a careful balance between the interests of the child, the parents, and society at large.

The article underscores the urgent need for a comprehensive regulatory framework, robust infrastructure development, and specialized medical education to support the responsible and effective use of genome sequencing in newborns. By analyzing international experience and highlighting current gaps in Ukrainian practice, the study provides a detailed roadmap for implementing genomic technologies to improve early diagnosis, reduce the burden of genetic diseases on families and the healthcare system, and ultimately align Ukraine's neonatal care with the global standards of precision medicine.

Keywords: neonatal screening, genome sequencing, whole genome sequencing, whole exome sequencing, hereditary diseases, rare diseases.

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СЕКВЕНУВАННЯ ГЕНОМУ НОВОНАРОДЖЕНИХ: НОВІ ГОРИЗОНТИ НЕОНАТАЛЬНОГО СКРИНІНГУ В УКРАЇНІ

Швидкий розвиток геномних технологій докорінно змінив ландшафт неонатального скринінгу, пропонуючи безпрецедентні можливості для раннього виявлення широкого спектру спадкових та рідкісних захворювань. Цей трансформаційний зсув виходить за рамки традиційних методів скринінгу, які часто обмежуються невеликою кількістю станів, пропонуючи розширені діагностичні можливості. Секвенування геному дозволяє одночасний аналіз сотень, якщо не тисяч, генів, що дозволяє виявляти широкий спектр генетичних порушень за допомогою одного тесту.

У цій статті розглядається поточний стан та майбутні перспективи інтеграції цієї передової технології в програми неонатального скринінгу, з особливим акцентом на унікальні виклики та можливості в українській системі охорони здоров'я. Вона надає всебічний огляд клінічних переваг, включаючи можливість ранньої та точної діагностики. Це раннє розуміння має вирішальне значення для впровадження персоналізованих медичних втручань та початку цілеспрямованого лікування одразу після народження, що може значно покращити результати здоров'я та якість життя уражених дітей.

Хоча клінічні переваги є величезними, їх впровадження не позбавлене значних етичних, правових та соціальних викликів. Ключові проблеми включають забезпечення справжньої інформованої згоди, оскільки батьки повинні приймати складні рішення щодо майбутньої генетичної інформації своєї дитини, та гарантування повної конфіденційності даних для захисту високочутливих геномних даних від неправомірного використання. Крім того, вирішення питання рівного доступу має першочергове значення для запобігання створенню дворівневої системи, де лише привілейовані групи можуть скористатися цими рятівними технологіями. Отже, розвиток генетичного компонента неонатального скринінгу має супроводжуватися створенням чіткої

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етичної та правової бази. Ця база повинна не лише враховувати технічні та клінічні стандарти, але й забезпечувати ретельний баланс між інтересами дитини, батьків та суспільства в цілому.

У статті підкреслюється нагальна потреба в комплексній регуляторній базі, розвитку надійної інфраструктури та спеціалізованій медичній освіті для підтримки відповідального та ефективного використання секвенування геному у новонароджених. Аналізуючи міжнародний досвід та висвітлюючи поточні прогалини в українській практиці, дослідження пропонує детальну дорожню карту впровадження геномних технологій для покращення ранньої діагностики, зменшення тягаря генетичних захворювань на сім'ї та систему охорони здоров'я, а також, зрештою, узгодження неонатальної допомоги в Україні зі світовими стандартами прецизійної медицини.

Ключові слова: неонатальний скринінг, секвенування геному, повне секвенування геному, повне секвенування екзому, спадкові захворювання, рідкісні захворювання.

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INTRODUCTION

From the moment of birth, the first days of a newborn's life are critically important for the detection and prevention of serious congenital and genetic disorders. This is why neonatal screening has become one of the key elements of child healthcare policy in many countries around the world [1]. Traditional screening methods, based on the biochemical analysis of several metabolites, have proven effective but remain limited in their ability to detect a wide range of conditions, especially rare or genetically determined disorders [2, 3].

The rapid development of biotechnology and genomics over the past decade has opened new possibilities for transforming approaches to neonatal screening [4]. Whole Genome Sequencing (WGS) and Whole Exome Sequencing (WES) allow for a much broader and more accurate analysis of an infant's hereditary information. These technologies enable the identification of hundreds of genetic disorders before the onset of clinical symptoms, providing a personalized approach to patient management, early initiation of therapy, and, in some cases, the prevention of disease development [5, 6].

This review article explores recent advancements in genomic sequencing in newborns, analyzes the advantages and challenges of implementing such methods in clinical practice, and places particular emphasis on the prospects of their application in Ukraine. In view of global trends and national needs, the article discusses the potential of genomic screening as a new frontier in neonatal medicine within the context of improving early diagnostic systems and personalized child healthcare.

MATERIALS AND METHODS

This article is based on a narrative review of contemporary scientific literature on genome sequencing in the context of neonatal screening. The information search was conducted using international electronic scientific databases, including PubMed, Scopus, and Web of Science. A systematic analysis was carried out on scientific publications published mainly between 2015 and 2025, including original studies, meta-analyses, and review articles describing clinical approaches, results of pilot programs, and practical experience with genome sequencing in newborns.

The review focused on articles addressing the accuracy, diagnostic value, clinical implementation, ethical considerations, and potential risks associated with the use of genome sequencing in early life. Special attention was paid to studies comparing traditional neonatal screening methods with WGS (whole genome sequencing) and WES (whole exome sequencing) technologies, as well as works that explored the capabilities and limitations of these technologies in the context of population-based screening.

The conducted analysis allowed for the systematization of current scientific approaches and the evaluation of the potential for integrating genome sequencing into the practice of early diagnosis of hereditary diseases.

Current State of Neonatal Screening

Neonatal screening is an organized medical examination program for newborns aimed at the early

detection of a range of hereditary, metabolic, and endocrine diseases that may have severe health consequences for the child if timely treatment is not provided. Newborn screening using dried blood spots (NBS — Newborn Bloodspot Screening) is one of the most effective public health preventive measures developed over the past 60 years. Since the introduction of the first screening program for phenylketonuria in the 1960s, the scale and technological capabilities of neonatal screening have significantly expanded, covering an increasingly wide spectrum of diseases.

Recently, DNA-based technologies have begun to be integrated into neonatal screening, allowing not only an expansion of the list of diseases that can be detected at the preclinical stage but also improving diagnostic accuracy, identifying carriers of genetic mutations, predicting disease progression, and personalizing approaches to treatment and monitoring of newborns. [7].

In many countries worldwide, neonatal screening is a mandatory part of the national healthcare system. For example, in the United States, according to recommendations from the American Academy of Pediatrics and the Centers for Disease Control and Prevention (CDC), screening covers more than 30 diseases, including phenylketonuria, congenital hypothyroidism, sickle cell anemia, cystic fibrosis, adrenogenital syndrome, as well as certain organic acidemias and defects of β-oxidation of fatty acids [8]. In European Union countries, the approach is more variable: some states, such as the Netherlands and Italy, have broad programs including 25–30 conditions, while in other countries, particularly some in Eastern Europe, programs are more limited. [9,10].

In Ukraine, the neonatal screening program remained limited for a long time. Until 2021, it covered only four conditions: phenylketonuria, congenital hypothyroidism, cystic fibrosis, and adrenogenital syndrome. Only in 2022, with the support of international partners, the Ministry of Health of Ukraine announced the expansion of screening to include 21 conditions using tandem mass spectrometry [11]. The implementation of this update is an important step toward the modernization of pediatric care and aligns with global trends in the field of rare diseases.

The emergence of WGS and WES technologies has opened a new era in neonatal screening, enabling the detection of genetic variations associated with rare diseases even before the onset of clinical symptoms. [12] However, genome-based methods — particularly WGS and WES — are not yet part of standard screening practice in Ukraine and are currently used only within research projects or selected clinical cases. At the same time, the integration of genomic technologies into routine neonatal screening faces a number of challenges, including ethical, social, and clinical concerns. Additional challenges include the lack of bioinformatics infrastructure, a shortage of clinical genetics specialists, and limited funding.

Genome Sequencing: Approaches and Potential

Genome sequencing is a technique used to determine the nucleotide sequence of DNA, providing comprehensive genetic information about an organism. In the context of neonatal screening, two main approaches are applied: WGS and WES [13, 14].

WGS decodes virtually the entire genetic information of a newborn—approximately 3 billion base pairs—covering both coding and non-coding regions of DNA. In contrast, WES focuses only on exons—the coding regions of genes—which make up about 1–2% of the genome but contain approximately 85% of known pathogenic mutations. [15] Both approaches have their own advantages and limitations depending on the research objectives and clinical context. [16]

The potential of genome sequencing in neonatal screening lies in significantly expanding the range of diagnosable hereditary and rare diseases. Traditional screening methods can detect only a limited number of metabolic and endocrine disorders, while WGS and WES make it possible to identify thousands of monogenic diseases, including those that previously remained undiagnosed or were diagnosed only after clinical symptoms appeared [17, 18].

The implementation of genome sequencing in neonatal practice also enables:

- establishing a molecular diagnosis in complex cases with nonspecific symptoms;
- identifying carriers of inherited diseases, which is important for further genetic counseling;
- predicting future disease risks;
- tailoring therapeutic strategies based on the patient's genetic profile.

However, the use of WGS and WES in newborns is associated with a number of challenges. One of the main issues is the interpretation of variants of unknown or uncertain clinical significance, which requires highly qualified expertise and the development of comprehensive databases. Additionally, there are ethical concerns related to obtaining information about conditions that may manifest in adulthood, as well as incidental findings unrelated to the primary screening purpose [19, 20, 21, 22].

From an economic standpoint, although the cost of sequencing has significantly decreased in recent years, it remains considerable, especially when factoring in the need for high-quality bioinformatics analysis and subsequent clinical support.

Despite these challenges, international projects and pilot programs clearly demonstrate the effectiveness and promise of integrating genome sequencing into neonatal screening, opening new opportunities for early diagnosis and prevention of inherited diseases.

One of the first and most well-known initiatives is BabySeq, launched by Harvard University and Boston Children's Hospital (USA) [23,24]. As part of this study, WES was performed on newborns with clinical symptoms as well as on healthy infants. The results showed that clinically significant genetic variants were detected in approximately 10% of cases, potentially impacting patient management—underscoring the potential of sequencing for early prevention and treatment. Moreover, the project explored ethical and psychological aspects in detail, particularly how parents perceive genetic information, which is crucial for the clinical adoption of such technologies.

In the United Kingdom, the national genomic service Genomics England launched a pilot project to include WGS in neonatal screening. This initiative aims to evaluate the effectiveness, safety, and acceptability of WGS implementation within the National Health Service (NHS). Participants in the program receive results that facilitate early diagnosis of rare diseases, significantly improving clinical outcomes.

In Canada, several pilot projects are underway in the provinces of Ontario and Alberta, aiming to integrate genome sequencing into routine neonatal screening protocols. A key component of these initiatives is the development of ethical guidelines, algorithms for genetic data interpretation, and public engagement in open dialogue about new technologies.

Similar efforts are observed in Australia, Israel, and several European countries, where genomic sequencing in neonatal screening is viewed as a key direction for healthcare modernization and the advancement of personalized medicine.

This growing body of experience provides a foundation for the further expansion of sequencing in neonatal screening worldwide and serves as a model for other countries planning to implement similar programs. [18].

Ethical, Legal, and Social Challenges

The integration of genome sequencing into neonatal screening opens up broad opportunities for early diagnosis and personalized medicine. However, it also raises a range of complex ethical, legal, and social issues that require careful consideration and regulation. [25, 26, 27].

One of the main ethical concerns is obtaining informed consent from parents. Since genome sequencing can reveal not only information about conditions that manifest in early childhood but also risks of adult-onset diseases or unexpected incidental findings (e.g., genetic predisposition to cancer), parents must be made aware of the full range of potential results and their implications. A major challenge is the extent to which parents are able to make informed decisions regarding such complex information during the critical period immediately after a child's birth. A related dilemma is determining which results should be disclosed — only those relevant to the neonatal period or also information about future health risks.

Another important issue is the child's right to genetic privacy and the ability to make autonomous decisions in the future about whether to receive information about their own genome. The dissemination of genetic data may potentially affect a person's private life and create risks of discrimination, for instance in insurance or employment [28, 29, 30].

The legal regulation of the use of genetic data remains inadequate in many countries. Not all jurisdictions have clearly defined norms regarding the storage, processing, and transfer of genomic information. The lack of unified standards creates risks related to privacy, data security, and secondary use of information without the consent of patients or their legal representatives. A particularly sensitive issue is the long-term storage of sequencing data and its potential future use for purposes beyond the initial screening [31].

An additional challenge is that, in the process of obtaining consent for genomic studies within neonatal screening, a key stakeholder is often overlooked — the child undergoing testing. While respect for parental authority, consent, and responsibilities is essential in both clinical and research settings, the primary goal of newborn screening remains the identification of asymptomatic children at high risk, for whom further investigation or early intervention may be offered. This approach may be viewed as a legally relevant duty of the state in light of the provisions of the United Nations Convention on the Rights of the Child (1989), which has been signed and ratified by 196 countries [32].

Specifically, Article 3 of the Convention states that "the best interests of the child shall be a primary consideration" in all actions concerning children. Other key provisions guarantee the child's right to express their views (Article 12), the right to privacy (Article 16), and the right to the highest attainable standard of health, including pre- and postnatal healthcare services (Article 24). It is important to emphasize that the latter article does not guarantee the right to be born healthy, but rather establishes the obligation of the state to promote this right as much as possible — in particular through the development of preventive healthcare [31].

In the context of newborn sequencing, one of the most challenging issues is ensuring that parents are

properly informed about the nature and implications of genetic testing, as well as the capabilities and limitations of new technologies. This reinforces the need for an effective communication system and the protection of the child's interests not only as a patient today but also as a future adult.

Furthermore, in many countries, legal regulation of the responsibilities of physicians and medical institutions for the accuracy of interpretation and disclosure of sequencing results remains underdeveloped. This creates potential legal risks in cases of erroneous or incomplete interpretation of genetic data.

The social dimensions relate to the psychological impact of genetic information on parents and the family as a whole. Identifying genetic risks — especially those for which no effective treatment exists — can lead to anxiety, stress, and changes in family dynamics. This highlights the importance of providing adequate psychological support and counseling.

Another social challenge is inequality in access to genomic technologies. The high cost of sequencing and limited healthcare resources may exacerbate disparities between social groups, where some families have access to state-of-the-art diagnostic tools while others do not.

In light of these concerns, a targeted approach to genomic screening may be particularly valuable — for example, for newborns with severe medical conditions, unclear clinical presentations, or known familial genetic risks [33].

Prospects for Genome Sequencing in Neonatal Screening in Ukraine

The introduction of genome sequencing into the neonatal screening system in Ukraine presents broad prospects for significantly improving the early diagnosis of hereditary and rare diseases, enhancing the quality of medical care for newborns, and transforming the healthcare system as a whole. However, realizing this potential requires a comprehensive approach, coordinated efforts at the national level, and active involvement of the medical community and society.

Firstly, the use of WGS and WES technologies would substantially expand the range of conditions that can be identified at an early stage, compared to traditional neonatal screening in Ukraine, which currently covers only four conditions. This creates the potential to detect hundreds of monogenic disorders, including those requiring urgent treatment or close monitoring. It also allows for the reduction of undiagnosed cases and shortens the time to diagnosis. This approach supports personalized treatment and prevention strategies, enhancing the effectiveness of medical interventions and improving patients' quality of life.

Secondly, the implementation of genome sequencing will stimulate the development of interdisciplinary clinical genetics and bioinformatics in Ukraine. The establishment of a modern laboratory infrastructure, training of highly qualified specialists, and creation of national electronic databases of genetic information in line with international standards will contribute to raising the country's scientific and medical profile and open new opportunities for participation in global genomic research initiatives.

Thirdly, integrating genome sequencing into the healthcare system aligns with global trends and WHO recommendations on the development of personalized medicine. Ukrainian patients will gain access to advanced, more accurate diagnostic tools, which is critically important for the timely detection and effective treatment of rare and genetic disorders. This will also help reduce the socio-economic burden associated with chronic diseases and disability.

At the same time, achieving these prospects will require overcoming a number of challenges. Foremost, it is essential to develop a national strategy for implementing genome sequencing in neonatal screening, including the creation of a regulatory framework, establishment of quality standards, introduction of ethical norms, and financing mechanisms. It is critically important to ensure transparent and well-defined procedures for obtaining informed parental consent, along with standardized protocols for interpreting and communicating results to families.

Ensuring equitable access to the technology across all regions of the country is another key issue, given existing geographical, economic, and infrastructural disparities. Addressing this requires investment in modernizing laboratory facilities, expanding telemedicine, and developing digital healthcare systems that enable centralized data processing and delivery of high-quality consultations.

One of the key success factors is increasing awareness among healthcare professionals and the general public about the possibilities and limitations of genome sequencing, as well as fostering a culture of responsible use of genetic information. This can be achieved by integrating relevant educational programs into the training of physicians, medical geneticists, counselors, and related specialists.

Finally, strengthening international collaboration with organizations, foundations, and research centers experienced in genomic screening will be a promising direction. Such partnerships will facilitate the adoption of best practices, improve the quality of scientific research and clinical protocols, and attract investment into Ukraine's healthcare system.

In conclusion, genome sequencing in neonatal screening has the potential to become a driving force for the modernization of the national healthcare system, significantly improve the diagnosis and prevention of hereditary diseases, and promote the development of personalized medicine. However, this requires the formulation of comprehensive national policies, effective coordination among stakeholders, and consistent implementation of initiatives with consideration of ethical, social, and economic dimensions.

CONCLUSIONS

1. The integration of DNA-based technologies into neonatal screening significantly enhances the ability to detect hereditary diseases at an early stage; however, it also introduces new ethical and legal challenges related to the use of genomic information.
2. Legislative regulation in the field of genetic research in newborns remains fragmented in many countries. The lack of unified standards for the storage, processing, and secondary use of genetic data threatens patients' rights, particularly with regard to confidentiality and informed consent.
3. Special attention must be given to ensuring the rights of the child in the process of neonatal screening. This includes maintaining a balance between parental representation and adherence to international legal instruments, particularly the UN Convention on the Rights of the Child.
4. It is necessary to develop and implement clear legal mechanisms that define the responsibility of healthcare professionals and institutions for interpreting the results of genomic testing, while also guaranteeing the child's right to the highest attainable standard of health.
5. The advancement of the genetic component in neonatal screening should be accompanied not only by technical and clinical improvements but also by the establishment of a clear ethical and legal framework that ensures a balance between the interests of the child, the parents, and society.

AUTHOR CONTRIBUTIONS

All authors substantively contributed to the drafting of the initial and revised versions of this paper. They take full responsibility for the integrity of all aspects of the work.

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CONFLICT OF INTEREST

The authors declare no conflict of interest.

REFERENCES

1. Schielen PCJI. Historical Appreciation of World Health Organization's Public Health Paper-34: Principles and Practice of Screening for Disease, by Max Wilson and Gunnar Jungner. International Journal of Neonatal Screening. 2025; 11(3):56. URL: https://doi.org/10.3390/ijns11030056.
2. Stark Z, Scott RH. Genomic newborn screening for rare diseases. Nature Reviews Genetics. 2023; 24:755–766. URL: https://doi.org/10.1038/s41576-023-00621-w
3. Gold NB, Harrison SM, Rowe JH, Gold J, Furutani E, Biffi A, Duncan CN, Shimamura A, Lehmann LE, Green RC. Low frequency of treatable pediatric disease alleles in gnomAD: An opportunity for future genomic screening of newborns. HGG Adv. 2021 Sep 25;3(1):100059. URL: https://doi.org/10.1016/s1096-7192(21)00585-0
4. Goldenberg AJ, Ponsaran R, Gaviglio A, Simancek D, Tarini BA. Genomics and Newborn Screening: Perspectives of Public Health Programs. Int J Neonatal Screen. 2022 Jan 28;8(1):11. URL: https://doi.org/10.3390/ijns8010011
5. Remec ZI, Trebusak Podkrajsek K, Repic Lampret B, Kovac J, Groselj U, Tesovnik T, Battelino T, Debeljak M. Next-Generation Sequencing in Newborn Screening: A Review of Current State. Front Genet. 2021 May 26;12:662254. URL: https://doi.org/10.3389/fgene.2021.662254
6. Woerner AC, Gallagher RC, Vockley J, Adhikari AN. The Use of Whole Genome and Exome Sequencing for Newborn Screening: Challenges and Opportunities for Population Health. Front Pediatr. 2021 Jul 19;9:663752. URL: https://doi.org/10.3389/fped.2021.663752
7. Bennetts B, Ho G, Shin S, Cheong PL, Wotton T, Ranieri E, Pirreca S. Newborn Genomic Sequencing Needs Confirmation but Not Repeating. Children (Basel). 2024 Oct 25;11(11):1287. URL: https://doi.org/10.3390/children11111287
8. Shah N, Brlek P, Bulić L, Brenner E, Škaro V, Skelin A, Projić P, Shah P, Primorac D. Genomic sequencing for newborn screening: current perspectives and challenges. Croat Med J. 2024 Jun 13;65(3):261-267. URL: https://doi.org/10.3325/cmj.2024.65.261

9. Therrell BL, Padilla CD, Borrajo GJC, Khneisser I, Schielen PCJI, Knight-Madden J, Malherbe HL, Kase M. Current Status of Newborn Bloodspot Screening Worldwide 2024: A Comprehensive Review of Recent Activities (2020-2023). Int J Neonatal Screen. 2024 May 23;10(2):38. URL: https://doi.org/10.3390/ijns10020038
10. Loeber JG, Platis D, Zetterström RH, Almashanu S, Boemer F, Bonham JR, Borde P, Brincat I, Cheillan D, Dekkers E, Dimitrov D, Fingerhut R, Franzson L, Groselj U, Hougaard D, Knapkova M, Kocova M, Kotori V, Kozich V, Kremezna A, Kurkijärvi R, La Marca G, Mikelsaar R, Milenkovic T, Mitkin V, Moldovanu F, Ceglarek U, O'Grady L, Oltarzewski M, Pettersen RD, Ramadza D, Salimbayeva D, Samardzic M, Shamsiddinova M, Songailiené J, Szatmari I, Tabatadze N, Tezel B, Toromanovic A, Tovmasyan I, Usurelu N, Vevere P, Vilarinho L, Vogazianos M, Yahyaoui R, Zeyda M, Schielen PCJI. Neonatal Screening in Europe Revisited: An ISNS Perspective on the Current State and Developments Since 2010. Int J Neonatal Screen. 2021 Mar 5;7(1):15. URL: https://doi.org/10.3390/ijns7010015
11. On Ensuring The Expansion Of Neonatal Screening Programs For Newborns To Create A New System Of Neonatal Mass Screening in Ukraine: Order of the Ministry of Health of Ukraine dated 01.10.2021 No. 2142: as of November 1, 2022. URL: https://zakon.rada.gov.ua/laws/show/z1403-21#Text
12. Ding S, Han L. Newborn screening for genetic disorders: Current status and prospects for the future. Pediatr Investig. 2022 Oct 24;6(4):291-298. URL: https://doi.org/10.1002/ped4.12343
13. Berg JS, Agrawal PB, Bailey DB Jr, Beggs AH, Brenner SE, Brower AM, Cakici JA, Ceyhan-Birsoy O, Chan K, Chen F, Currier RJ, Dukhovny D, Green RC, Harris-Wai J, Holm IA, Iglesias B, Joseph G, Kingsmore SF, Koenig BA, Kwok PY, Lantos J, Leeder SJ, Lewis MA, McGuire AL, Milko LV, Mooney SD, Parad RB, Pereira S, Petrikin J, Powell BC, Powell CM, Puck JM, Rehm HL, Risch N, Roche M, Shieh JT, Veeraraghavan N, Watson MS, Willig L, Yu TW, Urv T, Wise AL. Newborn Sequencing in Genomic Medicine and Public Health. Pediatrics. 2017 Feb;139(2):e20162252. URL: https://doi.org/10.1542/peds.2016-2252
14. Nurchis MC, Radio FC, Salmasi L, Heidar Alizadeh A, Raspolini GM, Altamura G, Tartaglia M, Dallapiccola B, Pizzo E, Gianino MM, Damiani G. Cost-Effectiveness of Whole-Genome vs Whole-Exome Sequencing Among Children With Suspected Genetic Disorders. JAMA Netw Open. 2024 Jan 2;7(1):e2353514. URL: https://doi.org/10.1001/jamanetworkopen.2023.53514
15. Adhikari AN, Gallagher RC, Wang Y, Currier RJ, Amatuni G, Bassaganyas L, Chen F, Kundu K, Kvale M, Mooney SD, Nussbaum RL, Randi SS, Sanford J, Shieh JT, Srinivasan R, Sunderam U, Tang H, Vaka D, Zou Y, Koenig BA, Kwok PY, Risch N, Puck JM, Brenner SE. The role of exome sequencing in newborn screening for inborn errors of metabolism. Nat Med. 2020 Sep;26(9):1392-1397. URL: https://doi.org/10.1038/s41591-020-0966-5
16. Stevenson DK, Wong RJ, Reiss JD, Shaw GM, Aghaeepour N, Mahzarnia A, Marić I. Pediatr Res. 2025 Jan 20;97(4):1258-1260. URL: https://doi.org/10.1038/s41390-025-03874-9
17. Knoppers BM, Bonilha AE, Laberge AM, Ahmed A, Newson AJ. Genomic sequencing in newborn screening: balancing consent with the right of the asymptomatic at-risk child to be found. Eur J Hum Genet. 2025 Mar;33(2):182-188. URL: https://doi.org/10.1038/s41431-024-01677-w
18. Ben-Moshe Y, Reid Sutton V. Newborn Screening-Advances, Challenges, and Future Directions. Clinics in Perinatology. 2025 Jul 14;52(3):449-459. URL: https://doi.org/10.1016/j.clp.2025.06.001
19. Lynch F, Best S, Gaff C, Downie L, Archibald AD, Gyngell C, Goranitis I, Peters R, Savulescu J, Lunke S, Stark Z, Vears DF. Australian Public Perspectives on Genomic Newborn Screening: Risks, Benefits, and Preferences for Implementation. Int J Neonatal Screen. 2024 Jan 17;10(1):6. URL: https://doi.org/10.3390/ijns10010006
20. Pereira S, Smith HS, Frankel LA, Christensen KD, Islam R, Robinson JO, Genetti CA, Blout Zawatsky CL, Zettler B, Parad RB, Waisbren SE, Beggs AH, Green RC, Holm IA, McGuire AL; BabySeq Project Team. Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project: A Randomized Clinical Trial. JAMA Pediatr. 2021 Nov 1;175(11):1132-1141. URL: https://doi.org/10.1001/jamapediatrics.2021.2829
21. Pichini A, Ahmed A, Patch C, Bick D, Leblond M, Kasperaviciute D, Deen D, Wilde S, Garcia Noriega S, Matoko C, Tuff-Lacey A, Wigley C, Scott RH. Developing a National Newborn Genomes Program: An Approach Driven by Ethics, Engagement and Co-design. Front Genet. 2022 May 30;13:866168. URL: https://doi.org/10.3389/fgene.2022.866168
22. Horton R, Wright CF, Firth HV, Turnbull C, Lachmann R, Houlston RS, Lucassen A. Challenges of using whole genome sequencing in population newborn screening. BMJ. 2024 Mar 5;384:e077060. URL: https://doi.org/10.1136/bmj-2023-077060
23. Armstrong B, Christensen KD, Genetti CA, Parad RB, Robinson JO, Blout Zawatsky CL, Zettler B, Beggs AH, Holm IA, Green RC, McGuire AL, Smith HS, Pereira S; BabySeq Project Team. Parental Attitudes Toward Standard Newborn Screening and Newborn Genomic Sequencing: Findings From the BabySeq Study. Front Genet. 2022 Apr 27;13:867371. URL: https://doi.org/10.3389/fgene.2022.867371
24. Wojcik MH, Zhang T, Ceyhan-Birsoy O, Genetti CA, Lebo MS, Yu TW, Parad RB, Holm IA, Rehm HL, Beggs AH, Green RC, Agrawal PB; BabySeq Project Team. Discordant results between conventional

newborn screening and genomic sequencing in the BabySeq Project. Genet Med. 2021 Jul;23(7):1372-1375. URL: https://doi.org/10.1038/s41436-021-01146-5
25. Vears DF, Savulescu J, Christodoulou J, Wall M, Newson AJ. Are We Ready for Whole Population Genomic Sequencing of Asymptomatic Newborns? Pharmgenomics Pers Med. 2023 Jul 1;16:681-691. URL: https://doi.org/10.2147/pgpm.s376083
26. Ulph F, Bennett R. Psychological and Ethical Challenges of Introducing Whole Genome Sequencing into Routine Newborn Screening: Lessons Learned from Existing Newborn Screening. New Bioeth. 2023 Mar;29(1):52-74. URL: https://doi.org/10.1080/20502877.2022.2124582
27. Mori M, Chaudhari BP, Ream MA, Kemper AR. Promises and challenges of genomic newborn screening (NBS) - lessons from public health NBS programs. Pediatr Res. 2025 Mar;97(4):1327-1336. URL: https://doi.org/10.1038/s41390-024-03689-0
28. Newson AJ. The promise of public health ethics for precision medicine: the case of newborn preventive genomic sequencing. Hum Genet. 2022 May;141(5):1035-1043. URL: https://doi.org/10.1007/s00439-021-02269-0 .
29. Van der Burg S., Oerlemans A. Fostering caring relationships: Suggestions to rethink liberal perspectives on the ethics of newborn screening. Bioethics. 2018. Vol. 32, no. 3. P. 171–183. URL: https://doi.org/10.1111/bioe.12425
30. Morrissey C., Walker R. L. The Ethics of General Population Preventive Genomic Sequencing: Rights and Social Justice. The Journal of Medicine and Philosophy: A Forum for Bioethics and Philosophy of Medicine. 2018. Vol. 43, no. 1. P. 22–43. URL: https://doi.org/10.1093/jmp/jhx034
31. Whither Health Research / D. Patrinos et al. The International Journal of Children's Rights. 2023. Vol. 31, no. 4. P. 865–889. URL: https://doi.org/10.1163/15718182-31040005
32. Knoppers BM, Bonilha AE, Laberge AM, Ahmed A, Newson AJ. Genomic sequencing in newborn screening: balancing consent with the right of the asymptomatic at-risk child to be found. Eur J Hum Genet. 2025 Mar;33(2):182-188. URL: https://doi.org/10.1038/s41431-024-01677-w
33. Jeanne M, Chung WK. DNA Sequencing in Newborn Screening: Opportunities, Challenges, and Future Directions. Clin Chem. 2025 Jan 3;71(1):77-86. URL: https://doi.org/10.1093/clinchem/hvae180

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