The American Journal of Medical Sciences and Pharmaceutical Research
ISSN 2689-1026 | Open Access

TYPE Original Research
PAGE NO. 65-68
DOI 10.37547/tajmspr/Volume07Issue08-10

Expanded Newborn Screening for Inborn Errors of Metabolism: Perspectives for Uzbekistan

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OPEN ACCESS

SUBMITTED 22 June 2025
ACCEPTED 18 July 2025
PUBLISHED 20 August 2025
VOLUME Vol.07 Issue08 2025

CITATION
Akhmedova Diloram Ilkhamovna, Khadjimukhamedova Shokhina Bakhodirovna, & Artikova Magina Akmalovna. (2025). Expanded Newborn Screening for Inborn Errors of Metabolism: Perspectives for Uzbekistan. The American Journal of Medical Sciences and Pharmaceutical Research, 7(8), 65–68. https://doi.org/10.37547/tajmspr/Volume07Issue08-10

Abstract: Expanded newborn screening (ENS) for inborn errors of metabolism represents a crucial advancement in modern neonatology and public health. While many developed countries have already integrated tandem mass spectrometry into national screening programs, several low- and middle-income countries, including Uzbekistan, are still at the initial stages of implementation [1,6]. This review summarizes the global experience with ENS, highlights the clinical and economic benefits of early detection, and discusses the current state and future perspectives for Uzbekistan [20-22]. Strengthening laboratory capacity, training medical professionals, and developing sustainable policies will be essential for establishing an effective ENS program in the country.

Keywords: -Expanded newborn screening, neonatology, pediatrics, inborn errors, tandem mass spectrometry, public health.

Introduction: Expanded newborn screening (ENS) has become an indispensable component of preventive pediatrics and public health, allowing the presymptomatic detection of inborn errors of metabolism and endocrine disorders. The early identification of affected infants and timely initiation of treatment can significantly reduce infant morbidity and

mortality, prevent irreversible complications, and improve long-term outcomes [1]. Globally, more than 50 disorders can be detected through ENS, including aminoacidopathies, organic acidemias, fatty acid oxidation disorders, and endocrine diseases [2,4,5].

In Uzbekistan and others countries of the Commonwealth of Independent States (CIS), neonatal screening programs have historically been limited to congenital hypothyroidism and phenylketonuria [20-22]. Despite these achievements, the lack of advanced laboratory infrastructure and limited awareness among healthcare providers pose significant challenges to expanding ENS. At the same time, the growing interest in public health modernization and integration with international practices creates a unique opportunity to introduce ENS as part of national health policy.

Global Experience with Expanded Newborn Screening. The concept of newborn screening originated in the early 1960s, when Robert Guthrie developed a bacterial inhibition assay to detect PKU [1]. The introduction of tandem mass spectrometry (TMS) in the 1990s revolutionized newborn screening by enabling the simultaneous detection of dozens of metabolic conditions from a single dried blood spot sample [6]. Today, most high-income countries screen for 20 to 40 conditions as part of ENS. Countries such as the United States, Germany, Italy, UK, South Korea, China and Japan have established comprehensive programs supported by strong healthcare infrastructure and legislation [2,3,19].

In contrast, low- and middle-income countries face challenges in adopting ENS, including high initial costs of TMS equipment, shortage of trained specialists, and limited public health budgets. However, successful pilot programs in countries such as Turkey, Brazil, and China demonstrate that gradual expansion is feasible and yields significant benefits [7].

Metabolic Disorders Included in Expanded Newborn Screening. Expanded newborn screening panels typically cover:

- Aminoacidopathies: phenylketonuria (PKU), maple syrup urine disease (MSUD), tyrosinemia type I [1,6].
- Organic acidurias: methylmalonic acidemia, propionic acidemia, isovaleric acidemia [6].
- Fatty acid oxidation disorders: medium-chain acyl-CoA dehydrogenase deficiency (MCADD), very long-chain acyl-CoA dehydrogenase deficiency (VLCADD) [6,7].
- Endocrine disorders: congenital hypothyroidism (CH), congenital adrenal hyperplasia (CAH) [15].
- Other: cystic fibrosis (CF), spinal muscular atrophy (SMA), severe combined immunodeficiency (SCID) [11–14].

Current Situation in Uzbekistan and the CIS. In Uzbekistan, national newborn screening programs currently include only congenital hypothyroidism and phenylketonuria [20]. Other inherited metabolic disorders remain undetected, leading to delayed diagnosis and poor outcomes. The Ministry of Health has expressed interest in expanding screening, but challenges remain, including limited laboratory infrastructure, shortage of qualified personnel, and financial constraints [21,22].

Similar situations exist in other CIS countries, where ENS is either absent or only partially implemented. Regional differences in healthcare funding, infrastructure, and training contribute to uneven progress. Nevertheless, increasing collaboration with international partners and regional pilot projects offer opportunities for gradual expansion [4,19].

Future Perspectives and Recommendations. The future of expanded newborn screening (ENS) in Uzbekistan and other CIS countries is closely linked to the adoption of modern diagnostic technologies, international collaboration, and the development of sustainable healthcare policies. Several key directions can be outlined:

1. Implementation of Tandem Mass Spectrometry (TMS): Establishing centralized laboratories equipped with TMS for broad-spectrum screening [6].
2. Capacity Building and Training: Developing education and continuous professional development programs for pediatricians, neonatologists, geneticists, and laboratory staff [7].
3. Integration with Digital Health Systems: Creating national registries for metabolic diseases and linking ENS results to electronic health records [17,18].
4. International Collaboration: Partnering with established programs and participating in international networks such as ISNS [4,19].
5. Expansion to Genetic and Genomic Screening: Introducing next-generation sequencing (NGS) for precision medicine approaches [16,17].
6. Health Economics and Policy Development: Demonstrating cost-effectiveness and engaging policymakers in sustainable program financing [7,8].

CONCLUSION

Expanded newborn screening (ENS) constitutes a critical advancement in modern pediatrics and public health, offering the opportunity to detect a broad spectrum of

metabolic and endocrine disorders at a presymptomatic stage. The early identification and timely initiation of treatment not only reduce infant morbidity and mortality but also prevent irreversible disability, thus improving long-term health outcomes and alleviating the socioeconomic burden on families and the healthcare system [1,7].

In Uzbekistan, neonatal screening has thus far been limited to a narrow range of conditions, such as congenital hypothyroidism and phenylketonuria. While these efforts have provided measurable benefits, the experience of countries with well-established ENS programs demonstrates that broadening the scope of screening is both clinically justified and cost-effective [20,21]. The adoption of advanced laboratory technologies, particularly tandem mass spectrometry, combined with capacity building of healthcare professionals, development of national registries, and integration into digital health platforms, represents a necessary foundation for the expansion of ENS.

Furthermore, the implementation of ENS should be regarded as a strategic public health priority, aligning with global trends and international recommendations. Strengthening interdisciplinary collaboration, fostering partnerships with international networks, and adapting best practices to the local healthcare context will be pivotal [4,6].

Thus, the expansion of newborn screening in Uzbekistan is not merely a technical enhancement of existing programs but a comprehensive investment in the nation's future health, equity, and sustainable healthcare development [7,8].

References

Therrell BL, Padilla CD, Borrajo GJC, et al. Current Status of Newborn Bloodspot Screening Worldwide 2024: A Comprehensive Review of Recent Activities (2020–2023). Int J Neonatal Screen. 2024;10(2):38. doi:10.3390/ijns10020038

Health Resources & Services Administration (HRSA). Recommended Uniform Screening Panel (RUSP). 2025. Available at: https://newbornscreening.hrsa.gov/about-newborn-screening/recommended-uniform-screening-panel (accessed Aug 28, 2025).

HRSA. Previously Nominated Conditions—Conditions Added to the RUSP (includes Infantile Krabbe disease, 2024). 2025. Available at: https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/previous-nominations (accessed Aug 28, 2025).

International Society for Neonatal Screening (ISNS). Guidelines and Global Overview of Newborn Screening. 2023. Available at: https://www.isns-neoscreening.org/guidelines/

MetabERN. Newborn screening—Early detection of metabolic disorders. 2024. Available at: https://metab.ern-net.eu/newborn-screening-2/

Kononets V, Babenko A, Pankov VV, et al. Tandem mass spectrometry in screening for inborn errors of metabolism: a bibliometric analysis (1991–2024). Front Pediatr. 2025;13:1463294. doi:10.3389/fped.2025.1463294

Yu M, Li J, Liu L, et al. Cost-effectiveness analysis of newborn screening by tandem mass spectrometry in Shenzhen, China. BMC Health Serv Res. 2022;22:1323. doi:10.1186/s12913-022-08394-4

Rana R, Keramat SA, Ahmed M. Cost-effectiveness of newborn screening for severe combined immunodeficiency: a systematic review. Clin Exp Pediatr. 2025;Epub ahead of print. doi:10.3345/cep.2025.00052

Shih STF, Keller E, Wiley V, Wong M, Farrar MA, Chambers GM. Economic evaluation of newborn screening for severe combined immunodeficiency. Int J Neonatal Screen. 2022;8(3):44. doi:10.3390/ijns8030044

Yoo N, Kim S, Kim J, Ahn JG, Kang I, Shin JJ. The clinical and socioeconomic aspects of T-cell receptor excision circle–based newborn screening for severe combined immunodeficiency in Southeast and East Asia. Front Immunol. 2025;16:1549768. doi:10.3389/fimmu.2025.1549768

Cooper K, Dudding T, Gration D, et al. Systematic review of newborn screening programmes for spinal muscular atrophy. Int J Neonatal Screen. 2024;10(3):49. doi:10.3390/ijns10030049

Wong KN, Cook S, Hart K, et al. A Five-Year Review of Newborn Screening for Spinal Muscular Atrophy in the State of Utah: Lessons Learned. Int J Neonatal Screen. 2024;10(3):54. doi:10.3390/ijns10030054

Lin H-C, Lee N-C, Weng W-C, et al. Newborn screening facilitates early treatment and improved outcomes for spinal muscular atrophy: five-year real-world evidence from Taiwan. Orphanet J Rare Dis. 2025;20:197. doi:10.1186/s13023-025-03697-1

McGarry ME, Raraigh KS, Farrell P, et al. Cystic Fibrosis Newborn Screening: A Systematic Review–Driven Consensus Guideline from the United States Cystic Fibrosis Foundation. Int J Neonatal Screen. 2025;11(2):24. doi:10.3390/ijns11020024

Arrigoni M, Nucci N, Moleti M, et al. Newborn screening for congenital hypothyroidism: worldwide

implementation, methods and coverage. Eur Thyroid J. 2025;14(1):1–14. doi:10.1530/ETJ-24-0327

Shen G, Li T, Chen X, et al. Newborn screening in the era of next-generation sequencing: history, recent advances, and future perspectives. Curr Opin Genet Dev. 2024;85:102111. doi:10.1016/j.gde.2024.102111

Shah N, Lewis R, Kingsmore SF, et al. Genomic sequencing for newborn screening. Mol Genet Genomic Med. 2024;12(6):e2331. doi:10.1002/mgg3.2331

Chan K, Goldberg JD, Brower AM, et al. NBSTRN tools to advance newborn screening research and translation. Int J Neonatal Screen. 2023;9(3):58. doi:10.3390/ijns9030058

Loeber JG, Platis D, Zetterström RH, et al. Neonatal Screening in Europe Revisited: An ISNS Perspective on the Current State and Developments Since 2010. Int J Neonatal Screen. 2021;7(1):15. doi:10.3390/ijns7010015

Republic of Uzbekistan. Presidential Decree PP-3440 (Dec 25, 2017): State program for early detection of congenital and hereditary diseases (includes neonatal screening for CH and PKU). Available at: https://lex.uz/docs/3471753

Republic of Uzbekistan. Presidential Resolution PQ-296 (Sep 8, 2023): On measures to protect maternal and child health and strengthen reproductive health (includes provision of cystic fibrosis screening kits 2024–2028). Available at: https://www.lex.uz/uz/docs/-6598641

Republican Center for Mother and Child Screening (Uzbekistan). Neonatal screening information page. Available at: https://med.uz/medgen/