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The role of the biomedical scientist in the laboratory diagnosis of the expanded newborn screening test.

The role of the biomedical in the laboratory diagnosis of the extended heel prick test
The role of biomedicine in the laboratory diagnosis of prolonged heel puncture testing

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SUMMARY

INTRODUCTION: Neonatal screening, known as the heel prick test, is an essential strategy for the early detection of congenital diseases, allowing for timely interventions and prevention of sequelae. With technological advances, the expanded model has incorporated more sensitive methods, increasing its diagnostic capacity. METHODOLOGY: This is an integrative literature review, conducted in the PubMed, SciELO, Virtual Health Library, and CAPES Portal databases , from 2015 to 2025. Controlled descriptors and Boolean operators were used, including articles available in full and excluding studies that did not meet the proposed objective. In the end, 10 articles were selected for analysis. RESULTS: The studies showed that expanded neonatal screening contributes significantly to the early diagnosis of metabolic, genetic, and endocrine diseases. The use of technologies, such as tandem mass spectrometry, which provides greater diagnostic accuracy, was highlighted . It was also observed that quality control and the correct execution of laboratory steps are fundamental to the reliability of the results. DISCUSSION: The analysis of the studies reinforces that the role of the biomedical professional is essential in all stages of the process, from laboratory analysis to the interpretation of results. Furthermore, factors such as regional inequality and structural limitations still represent challenges to the effectiveness of neonatal screening in Brazil. CONCLUSION: It is concluded that the biomedical professional plays a fundamental role in guaranteeing diagnostic quality in expanded neonatal screening, contributing to the early detection of diseases and the promotion of public health.

Descriptors: Neonatal Screening; Heel Prick Test; Laboratory Diagnosis; Biomedical; Congenital Diseases.

INTRODUCTION

Newborn screening, popularly known as the heel prick test, is one of the key prevention strategies in public health, by enabling the early identification of Genetic, metabolic, and endocrine diseases in newborns. In Brazil, its consolidation occurred with the implementation of the National Neonatal Screening Program (PNTN), responsible for to establish guidelines for screening for these conditions, with the goal of reducing the infant morbidity and mortality and prevent irreversible sequelae (Brazil, 2023; Silva; Gonçalves, 2020). However, the effectiveness of this program depends not only on its existence, but also on... quality of the process steps and timely access to diagnosis and treatment.

With the advancement of laboratory technologies, especially in the field of molecular biology and From clinical biochemistry, neonatal screening was expanded through the expanded heel prick test. This

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This modality incorporates techniques such as tandem mass spectrometry (MS/MS), allowing it to...
Simultaneous analysis of multiple metabolites from dried blood samples, which expands
significantly increased the number of diseases screened (Lima et al., 2022; Fiocruz, 2022). Despite this
Technological advancement, by itself, does not guarantee better clinical outcomes, being
Proper interpretation of the results and integration with effective clinical protocols are necessary .

Among the detectable diseases, phenylketonuria (PKU) stands out, an inborn error of
metabolism characterized by a deficiency of the enzyme phenylalanine hydroxylase, which leads to the accumulation
Phenylalanine in the body can cause irreversible neurological damage if not absorbed.
diagnosed early (Souza et al., 2020; Santos et al., 2020). Neonatal screening allows for
Identifying this condition while it is still asymptomatic allows for early intervention through...
dietary control, which highlights the direct impact of laboratory diagnosis on the prevention of
complications and in improving clinical prognosis.

In this context, the role of the biomedical professional is essential to guarantee quality and...
reliability of laboratory diagnosis. This professional works in all stages of the process,
from the correct collection and packaging of samples on filter paper to the execution of the analyses and the
Interpretation of results. In addition, it is responsible for quality control of the tests and for...
Validation of the methods used, ensuring diagnostic accuracy (Silva; Gonçalves, 2020).
In this way, the biomedical scientist assumes a strategic role in linking laboratory technology and practice.
clinical, contributing directly to the effectiveness of neonatal screening.

Despite the progress observed, the implementation of the expanded newborn screening test in Brazil...
It still faces significant challenges, especially regarding regional inequality and limitations.
structural factors and the need for professional qualifications. Expanding the exam coverage depends
investments in laboratory infrastructure, sample transport logistics, and training
continuous involvement of the professionals involved (Brazil, 2023; Fiocruz, 2022). In this sense, the discussion about
The role of the biomedical professional becomes even more relevant, since the quality of the diagnosis is...
directly related to the technical and scientific competence of this professional, which reinforces the
The importance of its recognition in the context of public health.

Given this scenario, the need to understand, in a more in-depth way, becomes evident.
The role of the biomedical professional in the context of expanded neonatal screening, especially with regard to
ensuring analytical quality, interpreting results, and contributing to diagnosis.
Early detection of congenital diseases. Thus, this study aims to analyze the role of the biomedical scientist.
in the laboratory diagnosis of the expanded newborn screening test, highlighting its relevance for effectiveness.
of neonatal screening strategies and for the promotion of public health.

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METHODOLOGY

This is a descriptive study, conducted through an integrative literature review, which It allowed for the gathering, evaluation, and synthesis of already published scientific knowledge. The choice of this This approach was adopted to allow for a broad and critical analysis of the biomedical professional's role in diagnosis. laboratory analysis of the expanded newborn screening test, highlighting its importance for the early detection of congenital diseases and for the promotion of neonatal health.

The literature review was conducted between September 2025 and June 2026. including national and international publications. The PubMed database was consulted. Central, SciELO, Virtual Health Library (BVS) and CAPES Periodicals Portal , recognized by credibility and comprehensiveness. The descriptors used, based on The Health Sciences Descriptors (DeCS) used were: " newborn screening" (heel prick test). "neonatal screening" , " biomedical scientist", "congenital diseases" (congenital diseases) and " laboratory diagnosis". The searches combined Boolean operators, such as ("heel prick test" AND "neonatal screening") AND ("biomedical" OR "laboratory diagnostics"), To refine the results and ensure relevance.

Articles published between 2015 and 2025, available in full and with free access, were included. and written in Portuguese, English, or Spanish. Undergraduate theses, dissertations, theses, and articles were excluded. Duplicates, research involving animals, and studies that did not directly address the role of the biomedical professional. or laboratory diagnosis.

After the initial selection based on titles and abstracts, the selected articles were analyzed. Critically, this allowed us to identify the main contributions regarding the role of the biomedical professional in testing. of the enlarged footprint and its clinical, technological, and social implications. This approach made it possible to understand how the biomedical professional's actions influenced the effectiveness of the diagnosis and contributed to Prevention and early management of congenital diseases.

The selection of studies occurred through the stages of identification, screening, eligibility and Inclusion, according to previously established criteria. The study selection process is... shown in Figure 1.

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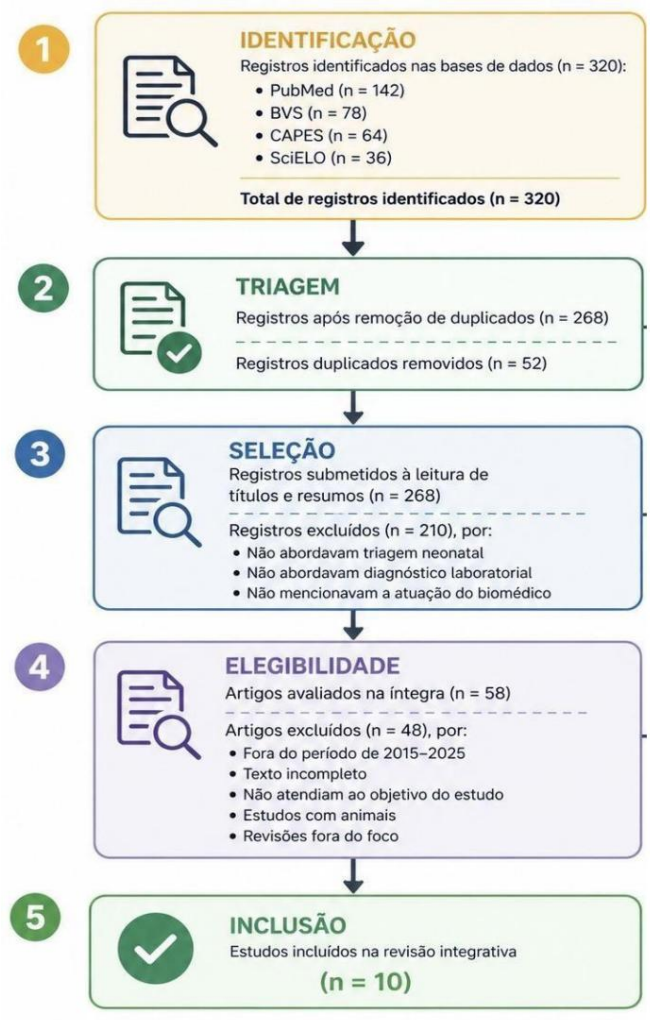
Figure 1 – Flowchart of the selection process for studies included in the review.



Source: Prepared by the author, 2026.

RESULTS

Ten studies were included in this review, as shown in Figure 1. The characterization

The composition of the included studies is presented in Table 1. The analyzed articles generally addressed the

The importance of neonatal screening as a fundamental strategy for the early diagnosis of diseases.

congenital conditions, as well as the role of the biomedical professional in ensuring the quality of laboratory tests.

Studies have shown that the relevance of using laboratory technologies is highlighted.

Advanced technologies, such as tandem mass spectrometry, allow for a wider range of diseases to be diagnosed.

tracked diseases contribute to greater diagnostic accuracy. Furthermore, it has been shown that monitoring...

Quality and proper execution of laboratory steps are fundamental to avoiding false results.

Positive or false negative results.

Another recurring point in the studies refers to the importance of the biomedical professional's role in...

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Interpretation of results, validation of tests, and integration with the multidisciplinary team.

which directly contributes to the effectiveness of newborn screening.

Despite technological advancements, the articles also highlight challenges, such as inequality.

Access to the exam is a challenge, as are structural limitations and the need for ongoing professional training.

Table 1 – Characterization of the scientific articles included in the review, according to authors, year of publication. publication, objective and main findings

AUTHOR/YEAR	OBJECTIVE OF STUDY	MAIN TYPES OF STUDY	FINDINGS
Botler et al., 2010	Assess the coverage	Epidemiological study: Neonatal screening coverage in Brazil	shows regional inequalities, impacting access to early diagnosis.
Calvo-Gonzalez, 2016	Analyze the aspects social aspects of neonatal screening.	Qualitative study of the	Newborn screening involves social and cultural factors in addition to laboratory diagnosis.
Pilar; Manfredini, 2018	Analyze clinical aspects and	Revision	The integration between laboratory diagnosis and
	neonatal from the screening laboratory		Clinical practice is essential for effective screening.
Santos et al., 2020	Studying inborn errors review	Metabolism	Expanded screening promotes the early detection of these diseases.
Silva; Gonçalves, 2020	Evaluate the role of review of in neonatal screening	Biomedical	Biomedical professionals are essential in the execution, quality control, and interpretation of tests.
Souza et al., 2020	Analyzing the phenylketonuria node in the context of neonatal screening.	Clinical study	Early diagnosis prevents irreversible neurological damage.

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Carvalho et al., 2021	Describe the laboratory diagnosis of metabolic diseases.	Clinical study	Early diagnosis contributes to a better clinical prognosis.
Lima; Rodrigues, 2022	Evaluate indicators review in neonatal screening	Quality	Quality control is fundamental for reliable results.
Oliveira et al., 2022	Discussing progress and the challenges of newborn screening.	Reviewing	Structural and logistical challenges persist in the healthcare system.
Brazil, 2023	Presenting guidelines for Program National Newborn Screening	Official document: Expanding newborn screening strengthens prevention and early diagnosis.	

Source: Prepared by the author, 2026.

As shown in Table 1, the studies analyzed show convergence regarding...
the importance of neonatal screening for the early diagnosis of congenital diseases and for the improvement of the quality of healthcare.

DISCUSSION

The studies analyzed demonstrate a consensus regarding the relevance of newborn screening. expanded for the early diagnosis of congenital diseases, especially by enabling
Early therapeutic interventions and reducing clinical complications. However, the authors present different perspectives on the factors that influence the effectiveness of this diagnostic process.

Botler et al. (2010) highlight that, despite the progress observed in the National Program of Newborn screening: coverage of the test is still uneven across Brazilian regions. According to the Authors, socioeconomic factors, and structural limitations directly impact access to
The population is unable to receive early diagnosis, compromising the effectiveness of neonatal care.

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Conversely, Brazil (2023) emphasizes the progress resulting from the expansion of the National Program of Newborn screening, especially the incorporation of newly screened diseases and the gradual expansion of Diagnostic coverage in the country.

In the laboratory setting, Pilar and Manfredini (2018) emphasize that the integration between the Laboratory diagnosis and clinical practice constitute one of the main factors for the success of screening. neonatal. The authors argue that the correct interpretation of laboratory results should occur coordinated with the clinical assessment of the newborn, which provides greater safety. diagnostic. This perspective complements the findings of Silva and Gonçalves (2020), which reinforce the The role of the biomedical professional is essential in the technical execution of examinations and in the control of laboratory quality and validation of results.

Furthermore, Santos et al. (2020) and Souza et al. (2020) agree in highlighting the importance of Expanded neonatal screening for the early detection of inborn errors of metabolism. Santos et al. (2020) They emphasize that expanding the newborn screening test has made it possible to identify a greater number of diseases. Metabolic disorders that are still in the asymptomatic phase, favoring better clinical outcomes. (Souza et al.) (2020) direct the discussion to phenylketonuria, highlighting that early diagnosis, associated With appropriate dietary intervention, it is possible to prevent irreversible neurological damage and improve significantly improves the quality of life of patients.

Similarly, Carvalho et al. (2021) show that laboratory diagnosis Early detection of metabolic diseases allows for more effective therapeutic interventions and reduces complications. Clinical findings contribute to a better prognosis for patients. These findings reinforce the importance the role of biomedical professionals in ensuring diagnostic accuracy and the reliability of the tests performed.

Another aspect widely discussed in the studies refers to quality control. laboratory. Lima and Rodrigues (2022) highlight that failures in the collection, storage, Transportation and processing of samples may compromise significantly alters the results of newborn screening, increasing the risk of false positives and false negatives. negative. In this context, the authors emphasize the need for rigorous control protocols of quality and continuous training of professionals involved in the laboratory process.

On the other hand, Calvo-González (2016) expands the discussion by addressing the social aspects and cultural issues related to newborn screening. The author argues that the diagnostic process should not to be understood not only from a biomedical perspective, but also considering the impacts emotional and social challenges faced by families after a diagnosis of congenital diseases. This perspective This complements the findings of Oliveira et al. (2022), who indicate that, despite technological advances Despite scientific advancements, structural, logistical, and administrative challenges persist, hindering the... Effectiveness of newborn screening in different regions of Brazil.

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Thus, it can be observed that the authors agree on the importance of neonatal screening. expanded and the role of the biomedical professional in ensuring diagnostic quality. However, they present different approaches to the subject, ranging from laboratory and technological aspects to Social and structural factors , as well as aspects related to public health management. Thus, It is evident that the effectiveness of neonatal screening depends not only on the incorporation of advanced technologies, but also professional qualification, multi-professional integration and Strengthening public health policies.

FINAL CONSIDERATIONS

This review showed that expanded newborn screening constitutes a strategy. essential for the early detection of congenital diseases, enabling therapeutic interventions timely and contributing significantly to the reduction of infant morbidity and mortality and to improving the quality of life for patients.

The studies analyzed demonstrated that the incorporation of laboratory technologies Advanced technologies, such as tandem mass spectrometry, have expanded the diagnostic capabilities of Newborn screening programs, allowing for the early identification of various metabolic diseases. genetic and endocrine factors. However, the authors also highlighted that the effectiveness of these Programs depend directly on the quality of the laboratory steps and the proper interpretation of the data. results and the integration between laboratory diagnosis and clinical practice.

In this context, the fundamental role of the biomedical scientist in the laboratory diagnosis of [disease/condition] stands out. expanded newborn screening test, acting in an integrated manner across the stages of collection, processing, and control. quality control, analysis, and interpretation of exams. In this way, this professional contributes directly for the diagnostic reliability and effectiveness of neonatal screening.

Furthermore, the literature reviewed demonstrated that, despite technological advances and Despite scientific advancements, significant challenges related to regional inequalities persist. Structural limitations and the need for continuous professional training. These factors may to compromise the scope and effectiveness of newborn screening programs in different contexts of the Brazilian healthcare system.

Given this, continuous investment in laboratory infrastructure becomes essential. professional qualification and strengthening of public policies aimed at neonatal screening, with the goal of To expand the population's access to early diagnosis and ensure greater equity in care. health.

Finally, the importance of developing new studies on the role of [the institution/organization] is highlighted.

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biomedical professionals in expanded neonatal screening, especially those related to clinical impacts, laboratory and social aspects of this process, contributing to the improvement of diagnostic practices and for the promotion of neonatal health.

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