

# PARENT OR GUARDIAN KNOWLEDGE OF BIOLOGICAL NEWBORN SCREENING

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**Abstract:** The objective of this study was to analyze the level of knowledge of parents or guardians about the biological newborn screening test. An integrative literature review study was conducted using the secondary databases Virtual Health Library, Latin American and Caribbean Health Sciences Literature, Scientific Electronic Library Online and Online System for Search and Analysis of Medical Literature based on the specific descriptors retrieved in the Health Sciences Descriptors. In general, it was identified that parents or guardians have limited knowledge about biological newborn screening, restricting themselves to some diseases, resulting in delays or risk of not performing the exam, also causing risks to the child's health. Efforts are needed from health professionals so that, throughout the prenatal period, pregnant women and family members are exposed to up-to-date and evidence-based information about the test, both in clinical consultations and in systematic health education actions in collective groups and also in the waiting room and other spaces of the unit and territory.

**Keywords:** newborn screening; knowledge; triage.

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## INTRODUCTION

Currently, the Neonatal Screening Program of Minas Gerais enables the Unified Health System to screen for six diseases through the biological neonatal screening test, they are: Sickle Cell Anemia and other Hemoglobinopathies, Biotinidase Enzyme Deficiency, Phenylketonuria, Cystic Fibrosis, Congenital Adrenal Hyperplasia and Congenital Hypothyroidism (Brasil, 2016). An increase made in May 2021, ratified by Law 14,154, raised to 50 the number of diseases and conditions to be screened by this test in the Unified Health System (Brazilian Society of Pediatrics, 2019).

The expansion of the test brings positive outcomes to society in the sense of providing a greater perspective on the knowledge of these diseases. However, for its effectiveness, it must continue to achieve high coverage rates and must improve its implementation in some regions of the national territory that have limited infrastructure (Bomfim et al., 2022; Perígolo et al., 2022).

The Guthrie test, popularly known as the heel prick test, is part of the National Neonatal Screening Program and has a preventive character, whose main purpose is to investigate metabolic and genetic diseases that may be asymptomatic in the early days of the newborn's life. The diseases screened by the exam have effective treatment, but some of them, when not identified and treated early, can lead to mental retardation or even death. Thus, early diagnosis directly interferes with the prognosis and improvement of the quality of life of affected individuals (Carvalho et al., 2020).

However, despite the impact on neonatal health, the lack of information from parents, family members and health professionals due to the lack of understanding of the importance of the test compromises the effectiveness of biological neonatal screening. In addition, the lack of preparation of the health team and the lack of dissemination about the test in the main media with easy access to the population by the competent bodies, hinders the success of the program (Vasconcelos et al., 2021). It should be added that the moment in which this information is offered can influence the way the family understands the subject, being more effective when provided throughout pregnancy, in order to produce maternal health literacy (Carvalho et al., 2020).



Maternal health literacy can be defined as the incorporation of cognitive and social skills required to enable pregnant women to access, understand, analyze and use the information necessary to preserve and improve their health conditions. The World Health Organization suggests health literacy and the empowerment of pregnant women as two key components of maternal health improvement programs. In this sense, providing pregnant women with education and training in various scenarios is a significant point for their empowerment, success, and well-being (Döndü et al., 2021).

In this context, important sources of information for pregnant and puerperal women about biological neonatal screening are the community and the media, in view of the attributes of primary health care, especially community orientation. A significant strategy is to carry out health education in the community spaces available in the area covered by the UBS so that information about the test is disseminated and increases the community's exposure to it. When pregnant and postpartum women do not have access to adequate knowledge, the hunch of laypeople can make them vulnerable and influence the decision-making process (Silva et al., 2021). There is a need for further studies on the subject in order to improve care, expand care measures, and improve the scope of public policies (Miranda et al., 2020). Thus, the present study sought to analyze the level of knowledge of parents or guardians about the biological newborn screening test.

## **MATERIALS AND METHODS**

An integrative literature review was conducted. This approach was adopted because it allows the combination of data from investigative and theoretical research that can thus be directed to conceptualizations, registration of gaps in the areas of investigation, theoretical review and methodological analysis of studies on a specific subject, allowing the analysis of the literature (SOUZA; SILVA; CARVALHO, 2010).

In this sense, six interdependent and interrelated phases were considered: elaboration of the guiding question, search or sampling in the literature, data collection, critical analysis of the included



studies, discussion of the results and presentation of the integrative review. How the guiding question was defined: What is the level of knowledge of the country or guardians about the biological newborn screening test? (SOUZA; SILVA; CARVALHO, 2010).

Studies were collected through an electronic search in the following databases available in the Virtual Health Library (VHL), Latin American and Caribbean Literature on Health Sciences (LILACS), the Scientific Electronic Library Online (SciELO) and the Analysis of Medical Literature (MEDLINE).

The inclusion criteria included complete articles available electronically, in Portuguese, English or Spanish and that presented the theme proposed in the title, abstract or descriptors. Regarding the eligibility criteria, letters to the editor, editorials, duplicate articles, and those that did not unequivocally address the theme under study were considered.

The survey of studies was conducted during the months of May to August 2024. As research strategies, the Health Sciences Descriptors (DECs) were used, retrieved through the website: <https://decs.bvsalud.org/>, which were neonatal screening, knowledge and screening, for the refinement of the search and better selection of data for analysis, the Boolean and Boolean was used to combine the selected descriptors.

For data collection, an instrument validated by Ursi (2005) for integrative reviews was developed, covering the following categories of analysis: identification code, title of the publication, author and author's education, source, year of publication, type of study, region in which the research was carried out and the database in which the article was published. After selecting the articles, the information that would be extracted from the studies was defined. To enable the apprehension of the information, a database developed in the Microsoft Office Excel 2010 software was used, composed of the following variables: title of the article, year of publication, study design, and main outcomes. The data obtained were grouped in a table and in thematic approaches and interpreted according to specific literature.



## FINDINGS

Initially, 20 studies were identified in the databases when the descriptors were combined, after the systematic reading of the titles and abstracts, duplicate studies (n<sup>o</sup>4) and 4 studies were excluded because they were not related to the central theme of the present research, thus, the final sample was 12 articles. The following table delimits the title, design, and main outcomes of the studies analyzed in the final composition of the present study (Chart 1).

Table 1. Studies included in the integrative literature review.

Parents' knowledge about newborn screening	Cross-sectional study with qualitative and quantitative approach	The results also showed that 100% of parents. They claim to recognize the importance of performing neonatal screening, but do not understand how the test is performed and which diseases can be screened.
Neonatal screening: maternal knowledge in a maternity hospital in the interior of Paraná, Brazil	This is a descriptive, cross-sectional study of character quantitative	The purpose of neonatal screening was known by 65% (n = 26) of the participants, and it was observed that most knew the purpose and knew about the importance of performance of the "heel prick test", a fact that denotes the importance of the guidelines that must be dispensed.
Analysis of parents' understanding of the Guthrie Test	Descriptive and cross-sectional study	Most participants. I did not have a good understanding of the importance of the test as a resource to promote prevention in child health.
Guthrie test: perception of pregnant women in prenatal guidance	This is an exploratory-descriptive, cross-sectional study with a quantitative approach	75% of the pregnant women did not know how to report which pathologies were screened, 16.3% mentioned that the diseases detected by the Guthrie Test have a genetic etiology, 82% see the need for more information about the Guthrie Test, 60% refer to the need to focus on which diseases were diagnosed
Knowledge about neonatal screening: discourses of mothers and fathers of newborns	This is a descriptive, qualitative study	Mothers and fathers understand the purpose of the heel prick test, had access to knowledge about neonatal screening, evidencing potentialities in the learning process and health education, through various sources, with emphasis on the professional performance of nurses in prenatal care and weaknesses regarding diseases detected in screening



Guthrie Test: evaluation of knowledge and importance for health	Cross-sectional observational research	Most of the interviewees had some information about the Guthrie Test. However, it was observed that a significant portion of pregnant and puerperal women did not know its importance and, on average, 52% of them did not receive it Information about prenatal testing
Knowledge of puerperal women about neonatal screening	A descriptive study with a quantitative approach	The results indicate a low understanding of the puerperal women regarding the importance of screening, despite the fact that most of them are aware of the performance of the test in their children; They did not know the ideal age to perform the screening and received information from nurses about the test at the time of hospital discharge.
What do mothers know about biological newborn screening?	This is a descriptive-exploratory study	The results showed little or no knowledge of the interviewed postpartum women about the theme, demonstrated through Testimonials acquired in the interview
Parents' knowledge about neonatal screening, contribution of the Portal dos Bebés - Guthrie Test website	Descriptive study	Although the mothers interviewed knew that their children have the right to take the test, they did not demonstrate knowledge about the diseases that can be prevented, the time of diagnosis, the sequelae arising from the lack of diagnosis and early treatment
Knowledge of puerperal women about the heel prick test	Descriptive and exploratory study	Of the 75 puerperal women, 47 (62.7%) would like to receive further clarification about the heel prick test, with emphasis on the appropriate collection period, followed by the screened diseases. Most participants (n=55; 85.9%) took their child for the test between the third and seventh day of life, as recommended by the Ministry of Health. Of the puerperal women, 54 (72%) were unable to name which diseases were screened by the heel prick test in Minas Gerais, nor did they know that most of them have a genetic etiology.
Mothers' knowledge about the heel prick test in a basic health unit	The research is descriptive, quantitative in nature	The results indicate that women's knowledge is superficial. The importance of the nurse's role in health education is highlighted.
Guthrie test: unveiling mothers' knowledge about the test	This is a descriptive, exploratory study with a qualitative approach	It was evidenced that only 3% of the deponents received guidance considered complete, 62% received guidance classified as incomplete.



Source: study data.

## DISCUSSION

In this study, the level of knowledge of parents or guardians about the biological neonatal screening test was evaluated, in general, it was identified that parents or guardians have limited knowledge about biological neonatal screening, being restricted to some diseases, resulting in delays or risk of not performing the exam, also entailing risks to the child's health.

The knowledge of fathers and mothers about the need to perform the Guthrie Test is a preventive action, “with solid knowledge on the subject, mothers will contribute to the effectiveness of neonatal screening” (Arduini et al., 2017), and “will be protecting their children from diseases that can aggressively affect health or even lead to death” (Gomes et al., 2019). The promotion of this knowledge can go beyond prenatal information, in a continuous way in the hospital environment and complemented through the media. Strategies that disseminate knowledge about Neonatal Screening can have a positive impact on the family's knowledge, empowering parents to care for the baby (Gomes et al., 2019).

It is believed that the information is not offered satisfactorily to mothers at the time the newborn screening test is being performed, or sometimes it does not happen at all, since the postpartum women have not appropriated the essence of biological newborn screening (Vasconcelos et al., 2021).

In a study conducted in a state in northeastern Brazil with parents and guardians of newborns, traditional knowledge about the test (Brazil et al., 2016; Gomes et al., 2019). Another study, carried out with mothers of newborns who underwent the test, showed that they had a perception of the relevance of the test, however, they reported that they were not properly informed about the purpose of the test (Vasconcelos et al., 2021). Thus, superficial knowledge about biological neonatal screening may be associated with the instruction received, which often corresponds only to the need to perform



the test, without detailing the phases involved in the process, the advantages and outcomes if it is not performed (Carvalho et al., 2020).

A study demonstrates the fragility of the information on Neonatal Screening provided to mothers in prenatal care, as only 50% of the mothers in this study received guidance on the Guthrie Test, and 75% did not know how to report on diseases screened by the test (Silva et al., 2017). Research carried out in a public maternity hospital corroborates the study, as it identified that 89% of the mothers who answered the proposed questionnaire were unable to indicate the purpose of the heel prick test and 30% did not know the test collection period (Mendes et al., 2017). This lack of information makes us reflect on whether this is the reason why mothers do not take their children to undergo the test, or go outside the recommended period (Miranda et al., 2020).

The diseases that are screened from the exam, because they are less frequent, may be little known, so there is a need for guidance on the signs and symptoms and lethality among the population (Takemoto et al., 2020). Thus, in prenatal care, it is essential to clarify and guide pregnant women and their families on how and in which place to perform the test, according to the collection network available in the family's state of residence, indicating the need for collection to be carried out by the 5th day of the newborn's life (Brasil, 2016).

Another study points out that the lack of information and superficial knowledge of parents about the Guthrie Test is notorious, that postpartum women would like to receive more information about the collection period and the diseases screened (Miranda et al., 2020; Silva et al., 2017). These findings are in agreement with a study carried out in Primary Care “in a municipality in the interior of Minas Gerais, which reports that 63% of puerperal women would like to receive more clarification about the heel prick test, such as the appropriate collection period and the diseases screened” (Arduini et al., 2017).

Another study carried out in the state of Bahia, with 18 mothers and two fathers, identified that “mothers and fathers had an understanding of the Guthrie Test, information acquired in actions during the prenatal period, through the waiting room and a group of pregnant women, continued in





hospital care, as well as provided by neighbors and the media” (Gomes et al., 2019). A study carried out in Minas Gerais identified that 57% of postpartum women received guidance on the test at prenatal consultations and 43% at hospital discharge (Arduini et al., 2017).

Consequently, in many cases, parents are faced with professionals immersed in the routine of services and in the execution of technical procedures that are absent from humanization and the educational dimension of care (Pedrosa et al., 2021). In addition, it is pertinent to use soft care technologies, which make use of intersubjective processes, such as therapeutic listening and therapeutic communication, which allow the focus of care to be the person and not the disease.

## **CONCLUSION**

The level of knowledge of parents or guardians about biological neonatal screening, in general, was limited, being restricted to some diseases, resulting in delays or risk of not performing the test, also entailing risks to the child’s health. Efforts by health professionals are necessary so that throughout the prenatal period, pregnant women and family members are exposed to up-to-date and evidence-based information about the test, both in clinical consultations and in systematic health education actions in collective groups and also in the waiting room and other spaces of the unit and territory.

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