

Assessment of knowledge and awareness levels of healthcare professionals involved in newborn heel prick screening

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ABSTRACT

Aims: The aim of this study is to evaluate the knowledge levels of healthcare workers involved in newborn heel prick screening regarding the screening process, sample collection steps, and the diseases screened for.

Methods: This descriptive and cross-sectional study was conducted with 202 healthcare workers employed at Family Health Centres in Malatya province between 07.11.2024 and 07.02.2025. Data were collected using a structured questionnaire consisting of 35 knowledge questions covering the newborn screening process, heel prick blood sampling procedures, and the clinical characteristics and management of the screened diseases.

Results: 75.7% of participants were physicians, 12.9% were nurses, and 11.4% were midwives. The level of knowledge regarding the heel prick blood collection process was generally found to be high. A significant difference was found in the level of knowledge regarding the practice among the professional groups, with midwives and nurses being more successful than physicians ($p=0.021$). The level of knowledge regarding the screened diseases was found to be higher among physicians ($p=0.003$). However, general knowledge about rare diseases such as biotinidase deficiency, congenital adrenal hyperplasia, and spinal muscular atrophy was found to be low.

Conclusion: In this study, while the level of knowledge regarding the steps of heel prick blood collection was found to be adequate knowledge gaps were identified regarding the clinical characteristics and management of the screened diseases. It is recommended that up to date, disease-focused and profession-specific training programmes for healthcare professionals be promoted.

Keywords: Newborn screening, mass screening, blood specimen collection, healthcare professionals, metabolic diseases

INTRODUCTION

Congenital metabolic disorders (CMDs) are a group of diseases that are mostly autosomal recessive and rare, but they are a significant public health issue because they encompass a wide range of different metabolic disorders. The prevalence of CMD in our country is reported to range between 1:1500 and 1:3000.^{1,2} If these diseases are not diagnosed early they can lead to severe neurological sequelae, developmental delay, and life-threatening complications. The incidence of genetically transmitted diseases increases, particularly in communities where consanguineous marriages are common. The rate of consanguineous marriage in Türkiye varies between 12.8% and 35% depending on the region, and is reported to be approximately 21% nationwide.^{3,4}

The idea of detecting diseases before symptoms appear has pioneered the development of newborn screening

programmes.⁵ Under the National Newborn Screening Programme (UYTP) in Türkiye, heel blood samples are taken from all live-born babies; screening for phenylketonuria (PKU), biotinidase deficiency (BD), congenital hypothyroidism (CH), cystic fibrosis (CF), congenital adrenal hyperplasia (CAH) and spinal muscular atrophy (SMA) is carried out.⁶

The success of newborn screening largely depends on the heel blood sample being collected using the correct technique, stored appropriately, and delivered to the laboratory in a timely manner. Errors made during the sample collection process can lead to incorrect test results and the need for repeat sampling. This causes delays in diagnosis and treatment, increasing the risk of damage caused by the diseases. However, early diagnosis and rapid treatment are

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of vital importance in newborn screening programmes. Although the scope of newborn screening programmes has been expanded in recent years, the limited number of studies evaluating healthcare professionals' knowledge of newborn screening means that potential shortcomings in practice cannot be adequately identified. This situation makes it difficult to determine training needs and develop screening programmes. This study was designed to assess the knowledge and awareness levels of healthcare professionals involved in newborn heel prick screening.

METHODS

Ethical approval for the study was obtained from the İnönü University Non-interventional Clinical Researches Ethics Committee (Date: 22.10.2024, Decision No: 2024/5378). All procedures were carried out in accordance with the ethical rules and the principles of the Declaration of Helsinki. Written informed consent was obtained from all participants before completing the questionnaire.

This study was designed as a descriptive and cross-sectional study. The study was conducted between 07 November 2024 and 07 February 2025 among healthcare professionals working at Family Health Centres in Malatya province. The study population consisted of 405 healthcare professionals (250 physicians and 155 nurses/midwives) working in Family Health Centres in the province during the study period. No sampling method was applied, and it was aimed to reach the entire population. A total of 202 healthcare professionals (153 physicians, 26 nurses and 23 midwives) who voluntarily agreed to participate were included in the study. This corresponds to approximately 49.8% of the target population. Data were collected using a structured questionnaire consisting of sociodemographic questions and 35 knowledge questions developed based on the literature.^{6-8,18,19} The knowledge questions were grouped into three domains:

- Knowledge about the newborn screening programme and heel prick blood sample collection procedures
- Knowledge about the diseases included in the national newborn screening panel
- Knowledge about the clinical characteristics, diagnosis and treatment approaches of these diseases.

The diseases covered in the questionnaire included PKU, CH, CF, BD, CAH, and SMA. Each correct answer was scored as 1 point, while incorrect answers and "I do not know" responses were scored as 0 points. The total knowledge score ranged from 0 to 35 points. Knowledge levels were classified according to the percentage of correct answers as follows: low knowledge (0–11 points, ≤33%), moderate knowledge (12–23 points, 34–66%), and high knowledge (24–35 points, ≥67%).

Statistical Analysis

The data analyses were performed using SPSS version 22.0. Descriptive statistics were expressed as frequency and percentage values. The Chi-square test was used to compare knowledge levels between occupational groups. A p-value <0.05 was considered statistically significant.

RESULTS

The sociodemographic characteristics of the participants are presented in Table 1. Of the participants, 75.7% were

physicians, 12.9% were nurses, and 11.4% were midwives. The mean age of the participants was 40.4±8.6 years. Knowledge levels regarding the heel prick blood sampling process are presented in Table 2. The percentage of correct answers exceeded 85% for several items, particularly those related to completing the sample form and the appropriate timing of sample collection. A statistically significant difference was observed between occupational groups in terms of knowledge regarding the heel prick blood sampling procedure (p=0.021). Midwives and nurses demonstrated higher practical knowledge levels than physicians (Table 3).

Table 1. Sociodemographic characteristics of participants (n=202)

Characteristic	n	%
Gender		
Female	102	50.5
Male	100	49.5
Marital status		
Married	164	81.2
Single	38	18.8
Having children		
Yes	157	77.6
No	45	22.4
Occupation		
Physician	153	75.7
Nurse	26	12.9
Midwife	23	11.4
Professional experience		
0-5 years	25	12.4
6-10 years	48	23.8
≥11 years	129	63.9
Mean age (years)	40.4±8.6	

Table 2. Knowledge level regarding the heel prick blood collection process

Question/application step	Correct n (%)	Incorrect n (%)
Sample collection time (48-72 hours)	181 (89.6)	21 (10.4)
Removal of the first drop of blood	167 (82.7)	35 (17.3)
Complete filling of the sample form	188 (93.1)	14 (6.9)
Warming the foot	159 (78.7)	43 (21.3)
Baby order in multiple pregnancies	172 (85.1)	30 (14.9)
Proper drying of the sample	176 (87.1)	26 (12.9)

Table 3. Knowledge level regarding the heel blood collection process by profession and comparison

Occupation	High n (%)	Moderate n (%)	Low n (%)	p
Physician	98 (64.1)	41 (26.8)	14 (9.1)	
Nurse	21 (80.8)	4 (15.4)	1 (3.8)	
Midwife	19 (82.6)	3 (13.0)	1 (4.4)	
Total	138 (68.3)	48 (23.8)	16 (7.9)	0.021

Regarding knowledge of the screened diseases, physicians were found to have significantly higher knowledge levels than nurses and midwives (p=0.003) (Table 4).

Table 4. Level of knowledge regarding screened diseases by profession and comparison

Profession	High n (%)	Moderate n (%)	Low n (%)	p
Physician	109 (71.2)	33 (21.6)	11 (7.2)	0.003
Nurse	13 (48.1)	9 (34.6)	4 (17.3)	
Midwife	10 (43.5)	9 (39.1)	4 (17.4)	
Total	132 (65.3)	51 (25.2)	19 (9.5)	

Disease-specific correct response rates regarding the conditions included in the newborn screening panel are presented in [Table 5](#).

Table 5. Knowledge level regarding diseases included in the newborn screening panel

Disease	Correct n (%)	Incorrect n (%)
Phenylketonuria	159 (78.7)	43 (21.3)
Congenital hypothyroidism	69 (34.2)	133 (65.8)
Biotinidase deficiency	127 (62.9)	75 (37.1)
Cystic fibrosis	152 (75.2)	50 (24.8)
Congenital adrenal hyperplasia	180 (89.1)	22 (10.9)
Spinal muscular atrophy	161 (79.7)	41 (20.3)

DISCUSSION

The success of newborn screening largely depends on the heel blood sample being taken using the correct technique, stored appropriately, and delivered to the laboratory in a timely manner. Errors made during the sampling process can lead to incorrect test results and the need for repeat sampling. In this study, healthcare workers' technical knowledge regarding the heel prick blood sampling process was generally found to be adequate. However, significant knowledge gaps were observed regarding the clinical characteristics and treatment processes of the screened diseases.

This study found that healthcare workers involved in newborn heel prick screening generally had sufficient practical knowledge, but there were gaps in their knowledge regarding certain congenital metabolic and genetic diseases. Notably, awareness of rarer diseases such as BD, CAH, and SMA was limited.

When disease-specific knowledge levels were analysed, the highest correct response rate was observed for CAH (89.1%), while knowledge regarding CH was relatively lower (34.2%). These findings indicate that awareness among healthcare professionals may vary considerably depending on the disease included in the screening panel ([Table 5](#)).

The collection of heel blood samples at the right time and using the appropriate technique is critical to the success of newborn screening programmes. In our study, the high level of knowledge among healthcare workers regarding the steps involved in sample collection suggests that practices in the field are largely carried out in accordance with standards. Similarly, Beyzadeoğlu and colleagues⁸ reported a significant increase in the knowledge levels of healthcare workers who received in-service training on newborn screening. Erdim and İnal¹⁸ also emphasised that nurses' responsibilities in the sample collection and dispatch process directly affect the accuracy of test results. However, the fact that the level

of knowledge about the clinical characteristics and long-term outcomes of screened diseases is lower than that of the implementation steps indicates that theoretical knowledge is not as strong as practical knowledge in the field.¹⁰ Bayrak's¹¹ study also reported that health care workers' knowledge levels about newborn screening programmes varied. Gürbüz¹² noted that family physicians require more training, particularly regarding rare metabolic diseases. These findings are consistent with the results of our study.

In a comparison between occupational groups, it was observed that midwives and nurses had a higher level of practical knowledge than doctors, whereas doctors were more knowledgeable about the theoretical aspects of diseases. This situation can be attributed to midwives and nurses taking a more active role in the sampling process in the field, while doctors focus more on the diagnosis and treatment processes. The literature also emphasises that nurses and midwives play a fundamental role in newborn screening.^{9,13} With the expansion of the scope of newborn screening programmes in recent years, the variety of diseases encountered by healthcare professionals has also increased. In particular, the addition of SMA to the screening panel has created a new need for knowledge in the field. However, the relatively low level of knowledge about SMA in our study suggests that training in this area is not yet sufficiently widespread. Demirtaş and colleagues also reported that family physicians may experience a lack of knowledge about CF and other rare diseases.¹⁴

It should not be forgotten that newborn screening is not merely a technical process, but also involves informing, guiding, and psychosocially supporting families. Having a sick child places a serious psychological burden on families, and the communication skills of healthcare personnel are of great importance during this process.¹⁵⁻¹⁷ Therefore, healthcare workers need to be supported not only with technical knowledge but also with communication and counselling skills. In addition, the correct interpretation of newborn screening results and their appropriate communication to the family are also important in preventing unnecessary anxiety. Failure to properly guide families in cases of suspicious results can lead to delays in the diagnostic process and a loss of trust among families. Therefore, healthcare professionals must also have sufficient knowledge about the clinical significance of screening results and the roadmap to be followed.

The content of training programmes should not be limited to sampling techniques; it should also cover the clinical characteristics of the diseases screened for, treatment approaches, and long-term follow-up processes. Sharing up-to-date information, particularly on rare diseases, will reduce knowledge gaps in the field. Regular in-service training, updating guidelines, and using digital training materials can support this process.

One of the strengths of this study is that it evaluated both the practical knowledge related to heel prick blood sample collection and the theoretical knowledge regarding the diseases included in the newborn screening programme. In addition, the study included different professional groups such as physicians, nurses, and midwives working in primary healthcare settings. This provided a comprehensive assessment of healthcare professionals' knowledge and awareness regarding newborn screening practices.

Limitations

This study has some limitations. The research was conducted in a single province, and the results cannot be generalised to the entire country. Furthermore, the data were collected through self-reported questionnaires. Nevertheless, our study provides important data in terms of revealing the knowledge levels of healthcare professionals involved in newborn screening programmes.

Consequently, in order to increase the effectiveness of newborn heel prick screening, regular, up-to-date and disease-focused training programmes for healthcare professionals need to be widely implemented. Raising awareness, particularly regarding rare metabolic and genetic diseases, will directly impact the success of early diagnosis and treatment processes.

CONCLUSION

This study found that healthcare professionals involved in newborn heel prick screening generally had sufficient practical knowledge, but there were gaps in their knowledge regarding certain congenital metabolic and genetic disorders. Notably, awareness of rarer disorders such as BD, CAH, and SMA was limited. The high level of practical knowledge among midwives and nurses involved in this process indicates that field practices are largely conducted in accordance with standards. In contrast, physicians were found to have stronger theoretical knowledge of diseases. This suggests that educational content should be differentiated for different professional groups. With the expansion of the scope of newborn screening programmes, the variety of diseases encountered by healthcare professionals has also increased. Therefore, training programmes should not be limited to sampling techniques alone; they should also cover the clinical characteristics of the diseases screened for, treatment approaches, follow up processes, and family information steps.

Consequently, it is recommended that regular, up to date, and disease focused in-service training programmes for healthcare professionals be promoted to increase the effectiveness of newborn heel prick screening. Raising awareness, particularly regarding rare metabolic and genetic diseases, will directly impact the success of early diagnosis and treatment processes and significantly contribute to newborns leading healthy lives.

ETHICAL DECLARATIONS

Ethics Committee Approval

Ethical approval for the study was obtained from the İnönü University Non-interventional Clinical Researches Ethics Committee (Date: 22.10.2024, Decision No: 2024/5378).

Informed Consent

Written informed consent was obtained from all individual participants prior to their inclusion in the study. Participants were fully informed about the study's aims, procedures, potential risks and benefits, and their rights—including the right to withdraw at any time without consequence. All participants voluntarily signed a written informed consent form.

Peer Review Process

This manuscript was subject to external peer review.

Conflict of Interest

The authors declare no conflicts of interest related to this study.

Financial Disclosure

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Author Contributions

Concept: BKT, MEB; Design: BKT, MEB; Control: BKT, MEB; Resources: BKT, MEB; Materials: MEB; Data Collection and/or Processing: MEB; Analysis and/or Interpretation: MEB, BKT, EU; Literature Review: MEB, HYB, ŞY; Writing the Article: MEB, HYB, ŞY; Critical Review: MEB, HYB, ŞY, BKT, EU.

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