ORIGINAL ARTICLE DOI: 10.20515/otd.1983178

Osmangazi Journal of Med, 2026;48(6):1042-1052

Osmangazi Journal of Medicine

e-ISSN: 2587-1579

Knowledge and Awareness of Inherited Metabolic Disorders and The Newborn Screening Program Among Pregnant Women: A Health Literacy Perspective

Gebelerin Kalıtsal Metabolik Hastalıklar ve Yenidoğan Tarama Programı Konusundaki Bilgi ve Farkındalık Düzeyleri: Sağlık Okuryazarlığı Perspektifi

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Received : 01.07.2026

Accepted : 07.08. 2026

Abstract: Newborn screening is a cornerstone of preventive child health, and understanding its rationale is one of the most important factors for its effectiveness. This study aimed to evaluate pregnant women's attitudes and opinions regarding the newborn heel-prick screening program and to examine the relationship between these attitudes and opinions and maternal health literacy. Pregnant women aged ≥18 years who did not require emergency intervention were included. Participants' sociodemographic and pregnancy-related characteristics and awareness of inherited metabolic diseases (IMDs) were assessed using a questionnaire, and the Maternal Health Literacy Inventory in Pregnancy was used to assess health literacy. 127 pregnant women with a mean age of 28.9 ± 5.1 years were included. 29.1% of participants rated their knowledge of the NBS as adequate, and 23.6% rated their knowledge of IMDs as adequate. Participants most often described the main purpose of NBS as early disease diagnosis. The total MHELIP score ranged from 44.7 to 100.0, with a median of 77.6, and 107 were considered to have desired health literacy. Participants with desired health literacy reported family physicians and nurses as sources of information and were more likely to identify the NBS's main purpose. Support for expanding NBS was reported by 89 participants. Higher maternal health literacy was associated with more frequent reliance on healthcare professionals as sources of information and more accurate identification of the main purpose of NBS. The findings cannot be generalized to the overall population and were not associated with an understanding of IMDs.

Keywords: health literacy, inherited metabolic disorders, MHELIP, newborn screening programs

Özet: Yenidoğan taraması, koruyucu çocuk sağlığının temel taşlarından biridir ve gerekçesinin anlaşılması, etkinliğinin en önemli unsurlarından biridir. Bu çalışma, gebe kadınların yenidoğan topuk kanı tarama programına ilişkin tutum ve görüşlerini değerlendirmeyi ve bu tutum ile görüşler arasındaki ilişkiyi maternal sağlık okuryazarlığı ile incelemeyi amaçlamıştır. Acil müdahale gerektirmeyen, 18 yaş ve üzeri gebeler çalışmaya dahil edildi. Katılımcıların sosyodemografik ve gebelikle ilişkili özellikleri ile kalıtsal metabolik hastalıklar (KMH) hakkındaki farkındalıkları bir anket aracılığıyla değerlendirildi ve gebelikte maternal sağlık okuryazarlığı, Gebelikte Maternal Sağlık Okuryazarlığı Envanteri (MHELIP) kullanılarak değerlendirildi. Ortalama yaşı 28.9 ± 5.1 yıl olan toplam 127 gebe kadın çalışmaya dahil edildi. Katılımcıların %29.1'i yenidoğan taraması (NBS) hakkında yetersiz bilgiye sahip olduğunu ifade ederken, %23.6'sı KMH'lar hakkında bilgi sahibi olduğunu kabul etti. Katılımcıların çoğu, NBS'nin erken hastalık tanısı için olduğunu belirtti. Toplam MHELIP puanları 44.7 ile 100.0 arasında olup, medyan değer 77.6 idi. Katılımcıların bilgi ediniminde en güvenilir sağlık kaynağı olarak aile hekimleri ve hemşireler gösterildi ve NBS'nin temel amacını doğru belirleme olasılıkları daha yüksekti. NBS'nin yaygınlaştırılmasına destek, katılımcıların 89'u tarafından bildirildi. Daha yüksek maternal sağlık okuryazarlığı düzeyi, bilgi kaynağı olarak sağlık profesyonellerine daha sık başvurulması ve yenidoğan tarama programının temel amacının daha doğru şekilde tanımlanması ile ilişkili bulunmuştur. Bulgular genel popülasyona genellenemez ve kalıtsal metabolik hastalıkların anlaşılmasıyla ilişkili bulunmamıştır.
Anahtar Kelimeler: sağlık okuryazarlığı, kalıtsal metabolik hastalıklar, MHELIP, yenidoğan tarama programı.

Etik Kurul Onayı: Çalışma Etlik Zübeyde Hanım Kadın Hastalıkları Hastanesi Bilimsel Araştırmalar ve Etik Kurulu tarafından onaylanmıştır.

(Karar no:2024/15, Tarih: 09.10.2024

Onam: Tüm yazarlar tarafından Telif Hakkı Devir Formu imzalanmıştır.

Telif Hakkı Devir Formu: Tüm yazarlar tarafından Telif Hakkı Devir Formu imzalanmıştır.

Yazar Katkı Oranları: E.Ö.A.: Conceptualization, study design, data collection, data analysis, manuscript writing and revision. A.G.Y.: Data collection, manuscript review. N.Ç.K.: Data collection, manuscript review. K.T.K.: Data collection. A.K.T.: Data collection. E.P.: Supervision, critical revision of the manuscript.

Hakem Değerlendirmesi: Hakem değerlendirmesinden geçmiştir.

Çıkar Çatışması Bildirimi: Yazarlar çıkar çatışması olmadığını beyan etmişlerdir.

How to cite/ Atıf için: Özsaydı Aktaşoğlu E , Yıldız AG , Çayönü Kahraman N , Tarazan Türk K , Tehçi AK , Polat E , Knowledge and Awareness of Inherited Metabolic Disorders and The Newborn Screening Program Among Pregnant Women: A Health Literacy Perspective, Osmangazi Journal of Medicine, 2026;48(6):1042-1052

1. Introduction

Rare diseases are chronic and often progressive conditions that collectively affect approximately 6–8% of the general population. Many of these conditions present during infancy or childhood and are associated with substantial challenges for affected individuals and their families, including delayed diagnosis, difficulties in disease management, and limited access to reliable information and resources. Although the definition of a rare disease varies across regions, diseases affecting fewer than 1 in 2,000 individuals are generally considered rare in the European Union and Türkiye (1). Inherited metabolic disorders (IMDs) are a large and expanding group of genetic disorders, but can also be classified as a rare disease. According to the International Classification of Inherited Metabolic Disorders, 1,450 inherited metabolic diseases have been identified (2), but since then, the number of identified IMDs has continued to grow. Today, there are 2040 identified IMDs (3).

Newborn screening is a cornerstone of preventive child health, aiming to identify selected inherited conditions early so that timely interventions can prevent irreversible morbidity and mortality. In Turkey, the national newborn screening program (NBS) includes phenylketonuria, congenital hypothyroidism, cystic fibrosis, biotinidase deficiency, congenital adrenal hyperplasia, and spinal muscular atrophy. Only biotinidase deficiency and phenylketonuria are classified as IMDs.

The effectiveness of newborn screening depends not only on the availability of laboratory testing but also on families' understanding of the NBS, appropriate follow-up, and adherence to repeat sampling when needed. A second blood sample is recommended within the first week, and as is known, National public information emphasizes that families are typically contacted only if repeat sampling or referral is required after birth to improve detection. This makes the antenatal period a critical opportunity to strengthen expectant mothers' knowledge and preparedness regarding newborn screening pathways.

In the 1970s, the term 'health literacy' was introduced (4) to refer to the ability to obtain, understand, and use health information to make informed decisions, promote health, and prevent disease (5). Maternal health literacy refers to the cognitive and social competencies that empower women to seek, understand, and use health information to protect and promote maternal and child health (6), and it may play a key role in shaping pregnant women's knowledge and perceptions of newborn screening, including screening for IMDs. Higher health literacy is expected to facilitate better comprehension of screening aims, procedures, and the importance of timely follow-up. Therefore, this study aimed to evaluate pregnant women's knowledge and opinions regarding the NBS using a researcher-developed questionnaire and to examine the relationship between these and maternal health literacy, as assessed by the Turkish version of the Maternal Health Literacy Inventory in Pregnancy (MHELIP) (7).

2. Materials and Methods

2.1. Study participants

The study was initiated at Ankara Etlik Zübeyde Hanım Training and Research Hospital. The study was planned to begin between November 2024 and May 2025, but due to the hospital's closure in January 2025, the target sample size could not be reached at this site. Without any changes to the study protocol, the study was continued in the same manner at the Ankara Etlik City Hospital, Department of Obstetrics and Gynecology, where data collection was completed.

Sample size calculation was performed using G*Power version 3.1.9.7. The required sample size was estimated based on the planned comparison of pregnant women grouped by health literacy level and the examination of between-group differences in knowledge and awareness regarding the NBS, using a one-way ANOVA model. Assuming a Cohen's f effect size of 0.40 (large effect), a Type I error rate (α) of 0.05, and a power of 95%, the minimum required sample size was 112 pregnant women. Accordingly, at least 112 participants were planned to be included. However, since the number of participants in some individual health literacy categories was too small to allow reliable subgroup comparisons, the inadequate and problematic health literacy categories were combined into a "limited" health literacy group, while the adequate and excellent categories were combined into a "desired" health literacy group. This resulted in 20 participants in the limited group and 107 in the desired group. Following dichotomization, a post hoc power analysis was performed using two-sided comparisons of two independent proportions.

Assuming a large effect size (Cohen's h = 0.80), a Type I error rate (α) of 0.05, and the actual group sizes (20 and 107), the achieved statistical power was 92.76%. Therefore, the current sample retained sufficient power to detect large differences between groups; however, unequal group sizes may have limited the detection of small to moderate effects.

Pregnant women aged ≥18 years who were attending antenatal follow-up and provided informed consent were eligible for inclusion. Pregnant women who were under follow-up but required urgent or emergency medical intervention at the time of recruitment, and were unable to complete the questionnaire, were excluded from the study.

The study was planned in accordance with the principles of the Declaration of Helsinki, and ethical approval was obtained from both centers (No: 2024/15 / AEŞH-BADEK1-2025-731). Language editing and manuscript preparation were supported by digital tools; all scientific content was developed, reviewed, and approved by the authors.

2.2 Data Collection Instruments

To assess pregnant women's knowledge and opinions about the national NBS, the researchers developed a structured questionnaire informed by relevant literature and clinical experience. The developed questionnaire included multiple-choice and/or open-ended questions designed to obtain information on participants' sociodemographic characteristics, pregnancy-related characteristics, and awareness of IMDs. All participants completed both the researcher-developed questionnaire and the Turkish-validated Maternal Health Literacy Inventory in Pregnancy (MHELIP).

MHELIP, originally developed by Taheri et al. (8), has Turkish validity and reliability examined by Abay et al. (7) The scale consists of 48 items rated on a 5-point Likert scale. Items 1–21 assess pregnancy-related health knowledge and are scored from 1 ("I do not know at all") to 5 ("I know very well"). Items 22–48 evaluate functional health literacy and are scored from 1 ("never") to 5 ("always"). The scale comprises four subdimensions: (1) Maternal Health Knowledge, (2) Search for Maternal Health Information, (3) Assessment of Maternal Health Information, and (4) Maternal Health Decision-Making and Behavior. Scores on the four MHELIP subscales and the total score were calculated separately. Based on the total score, the group that scored 'inadequate' and 'problematic' was considered to have limited health literacy, while the group that scored 'adequate' and 'excellent' was considered to have the desired level of health literacy. The Turkish version of MHELIP was administered with permission from the scale owner/authors, and total and/or subscale scores were used in the analyses.

2.3 Statistical analyses

Descriptive statistics were presented as mean ± standard deviation, median (minimum–maximum), and frequency (percentage), as appropriate. The normality of continuous variables was assessed using the Kolmogorov–Smirnov and Shapiro–Wilk tests. For comparisons of non-normally distributed independent continuous variables, the Mann–Whitney U test was used. Associations between independent categorical variables were examined using the chi-square test, and the Fisher's exact test was applied when chi-square assumptions were not met. A p-value < 0.05 was considered statistically significant. All analyses were performed using IBM SPSS Statistics version 27.0.

3. Results

A total of 127 pregnant women were included in the study. The mean age of the participants was 28.9 ± 5.1 years (median 28, range 18–46), and that of their spouses was 31.6 ± 5.8 years (median 30, range 19–52) (Table 1). Regarding education, nearly half of the pregnant women were university graduates (48.8%), and a similar distribution was observed among spouses, with the highest proportion being university graduates (44.1%). Most participants reported that their income and expenditure were balanced (62.2%), and the rate of consanguineous marriage was 6.3%. Overall, 68.5% of the participants had at least one child.

Overall, 29.1% of participants rated their knowledge about the NBS as adequate, whereas 28.3% reported having no idea. Most participants (67.7%) had previously heard the names of the diseases included in the NBS. Among the diseases, SMA was the most commonly recognized condition (62.2%). The most frequently reported sources of information were family physicians and the internet (both 40.9%), followed by social media (33.9%) and TV (31.5%) (Table 2). A majority of participants (78.0%) correctly identified that screening samples are obtained by collecting a few drops of blood from the newborn's heel. One participant stated that the specimen collected for the NBS was urine. Participants most commonly endorsed the purpose of the NBS as early diagnosis of a disease (n=99), followed by early initiation of treatment (n=89). Twenty-one participants stated that the collected samples were obtained for future research purposes. (16.5%) (Table 2). Notably, one participant stated, "I

provided a heel-prick blood sample for my first baby, but because I do not trust the purpose of sampling, I will not allow it again".

Overall, 23.6% of participants rated their knowledge about IMDs as adequate. Almost all participants (99.2%) reported having no child diagnosed with an IMD. Most participants agreed that consanguineous marriage increases IMD risk (78.7%) and that having an affected child is important for the likelihood of IMDs in other siblings (67.7%). The majority also stated that IMDs should be investigated in cases of unexplained infant/child death (81.1%). Regarding perceived onset, 52.0% indicated that IMDs may present with findings in utero, and 35.4% reported that IMDs can begin at any age. For diagnostic approaches, 66.1% identified NBS, followed by specialized laboratory techniques (36.2%) and genetic testing (34.6%). Most participants believed that diagnostic tests for IMDs should be interpreted primarily by a pediatric metabolic diseases specialist (46.5%), whereas 22% reported having no opinion. Participants' answers were also summarized in Table 3.

MHELIP scores indicated an overall high level of health literacy among participants. The total MHELIP score ranged from 44.7 to 100.0, with a median of 77.6 and a mean ± SD of 78.3 ± 13.2. Most participants were classified as adequate (65/127; 51.2%) (Table 4). The distribution of subscale scores and the number of participants are also summarized in Table 4 and Figure 1. In this study, health literacy scores were divided into two groups: 'limited' and 'desired'. Among the participants, 20 were classified as having limited health literacy and 107 as having desired health literacy. Maternal age did not differ significantly between the groups, whereas paternal age was significantly higher in the desired health literacy group. No significant between-group differences were observed in maternal or paternal educational level, income status, consanguinity, or having children (Table 5).

Regarding the NBS, self-reported knowledge level, recognition of individual screened conditions, and knowledge of the sampling method were similar in the two groups. However, participants with desired health literacy reported family physicians and nurses as sources of information significantly more often than those with limited health literacy. They were also significantly more likely to identify the main purpose of the NBS as early disease diagnosis (Table 5). No significant differences were found between the groups in self-reported knowledge of IMDs, having a child with an IMD, awareness of the relationship between consanguinity and IMD risk, or opinions regarding recurrence risk within the family.

No significant differences were found between the limited and desired health literacy groups regarding opinions on the possible age of onset of IMDs, available diagnostic methods, or which physicians could evaluate diagnostic tests for these disorders (Table 6). Regarding the NBS, a clear majority supported expanding screening to more diseases (Yes: n=89; 70.1%), but this difference was not statistically significant between the groups.

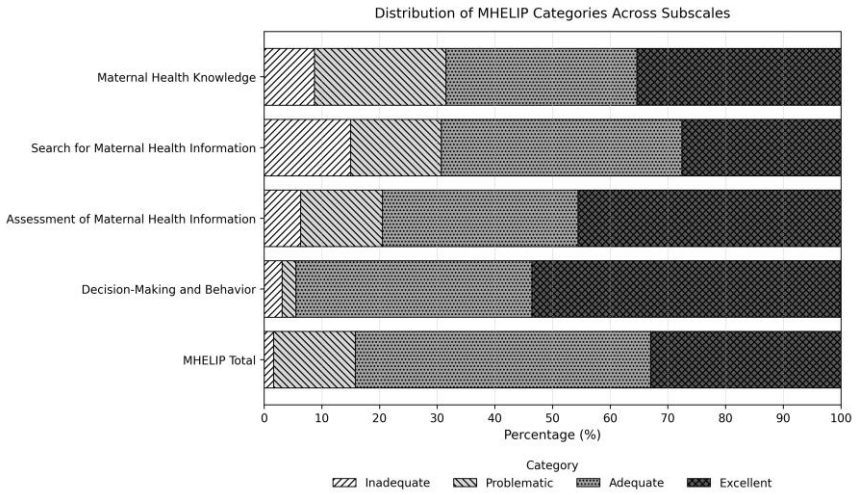

Figure 1. Distribution of Maternal Health Literacy Inventory in Pregnancy (MHELIP) categories across subscales

Table 1. Sociodemographic characteristics of the participants

Variables	**n (%) / Mean±SD**	**Median (Min–Max)**
Age of pregnant women (years)	28.9±5.1	28 (18–46)
Age of spouses (years)	31.6±5.8	30 (19–52)
	n	**(%)**
Educational status of pregnant women		
Cannot read/write	2	1.6
Primary school	7	5.5
Middle school	18	14.2
High school	32	25.2
University	62	48.8
Master's degree	6	4.7
Educational status of spouses		
Cannot read/write	2	1.6
Primary school	9	7.1
Middle school	15	11.8
High school	38	29.9
University	56	44.1
Master's degree	7	5.5
Income status		
Income less than expenses	23	18.1
Income equals expenses	79	62.2
Income greater than expenses	25	19.7

Table 2. Responses to general questions about the Newborn Screening Program

Domain / Question	**Response / Item**	**n (%)**
Self-rated knowledge about the NBS program	No idea	36 (28.3)
	Undecided	20 (15.7)
	Inadequate	34 (26.8)
	Adequate	37 (29.1)
Previously heard the names of diseases included in the NBS program	Yes	86 (67.7)
	No	41 (32.3)
Which NBS diseases have you heard of?*	Biotinidase deficiency	15 (11.8)
	Phenylketonuria	30 (23.6)
	Congenital hypothyroidism	22 (17.3)
	Congenital adrenal hyperplasia	13 (10.2)
	Cystic fibrosis	30 (23.6)

	Spinal muscular atrophy (SMA)	79 (62.2)
	Unanswered	1 (0.8)
Sources of information about the NBS program*	Family physician	52 (40.9)
	Pediatrician	28 (22.0)
	Nurse	36 (28.3)
	Internet	52 (40.9)
	TV	40 (31.5)
	Social media	43 (33.9)
	Relatives	9 (7.1)
	Neighbors	3 (2.4)
	School	1 (0.8)
	Other	13 (10.2)
	Unanswered	18 (14.2)
The purpose of the NBS as*	early diagnosis if a disease is present	99 (78.0)
	initiating treatment early	89 (70.1)
	estimating the number of affected individuals in the country	25 (19.7)
	using collected samples for further research	21 (16.5)
	Other	4 (3.2)
	Missing response	2 (1.6)

NBS; newborn screening.
** Participants were allowed to select more than one option (multiple responses permitted).*

Table 3. Participants' responses regarding inherited metabolic disorders

Item	**Response options**	**n (%)**
How would you rate your level of knowledge about IMDs?	Adequate	30 (23.6)
	Inadequate	41 (32.3)
	Undecided	41 (32.3)
	Heard for the first time	14 (11.0)
	No information	1 (0.8)
Do you have a child with an IMD?	Yes	1 (0.8)
	No	126 (99.2)
Does consanguineous marriage increase the risk of IMDs?	Yes	100 (78.7)
	No	4 (3.1)
	No idea	22 (17.3)

	No information	1 (0.8)
If one child has an IMD, is it important regarding the likelihood in other children?	Yes	86 (67.7)
	No	1 (0.8)
	No idea	39 (30.7)
	"If one child has it, the other will not"	1 (0.8)
If an IMD is present in a relative, is it important regarding the likelihood in other relatives?	Yes	86 (67.7)
	No idea	40 (31.5)
	No	1 (0.8)
In the presence of unexplained infant/child death, should IMDs be investigated?	Yes	103 (81.1)
	No idea	23 (18.1)
	No	1 (0.8)
At what age do IMDs begin (in your opinion)?*	May present with findings in utero	66 (52.0)
	Present at birth	12 (9.4)
	Begin in infancy	21 (16.5)
	Begin in childhood	7 (5.5)
	Begin in adulthood	4 (3.1)
	Can begin at any age	45 (35.4)
How are IMDs diagnosed?*	Amniocentesis	20 (15.7)
	Chorionic villus sampling	27 (21.3)
	Newborn screening programs	84 (66.1)
	Specialized laboratory techniques	46 (36.2)
	Routine laboratory tests	38 (29.9)
	Genetic testing methods	44 (34.6)
	Unanswered	9 (7.1)
Who can evaluate/interpret the tests performed for the diagnosis of IMDs?*	Any physician can evaluate/interpret	10 (7.9)
	A neonatologist can evaluate/interpret	47 (37)
	A pediatric metabolic diseases specialist can evaluate/interpret	59 (46.5)
	A pediatrician can evaluate/interpret	45 (35.4)
	No opinion	28 (22.0)
	Unanswered	3 (2.4)

IMD; Inherited metabolic disorders
**Multiple responses may have been permitted for these items; therefore, totals may exceed 100%.*

Table 4. Maternal Health Literacy Inventory in Pregnancy scores and categorical distribution

Subscale	Min–Max	Median	Mean ± SD	Inadequate n (%)	Problematic n (%)	Adequate n (%)	Excellent n (%)
Maternal Health Knowledge	28.5–100.0	75.0	74.9 ± 18.0	11 (8.7%)	29 (22.8%)	42 (33.1%)	45 (35.4%)
Search for Maternal Health Information	25.0–100.0	70.8	73.0 ± 17.0	19 (15.0%)	20 (15.7%)	53 (41.7%)	35 (27.6%)
Assessment of Maternal Health Information	37.5–100.0	83.3	80.5 ± 15.0	8 (6.3%)	18 (14.2%)	43 (33.9%)	58 (45.7%)
Maternal Health Decision-Making and Behavior	43.3–100.0	85.0	84.3 ± 12.0	4 (3.1%)	3 (2.4%)	52 (40.9%)	68 (53.5%)
MHELIP – Total	44.7–100.0	77.6	78.3 ± 13.2	2 (1.6%)	18 (14.2%)	65 (51.2%)	42 (33.1%)

MHELIP; Maternal Health Literacy Inventory in Pregnancy

Table 5. Summary of key comparisons between limited and desired health literacy groups

Variable	Limited Health Literacy (n=20)	Desired Health Literacy (n=107)	*p* value
Maternal age, mean ± SD	27.4±4.2	29.2±5.2	0.143*
Paternal age, mean ± SD	28.6 ± 5.8	32.1 ± 5.6	0.008*
Previously heard the names of screened diseases	10 (50.0%)	76 (71.0%)	0.065†
Family physician as information source	4 (20.0%)	48 (44.9%)	**0.038**†
Nurse as information source	2 (10.0%)	34 (31.8%)	**0.047**†
Correctly identifying the main aim of newborn screening as early diagnosis	12 (60.0%)	87 (81.3%)	**0.035**†
Identifying the aim as determining the number of patients in the country	7 (35.0%)	18 (16.8%)	0.061†
Self-rated knowledge of inherited metabolic diseases as sufficient	2 (10.0%)	28 (26.4%)	0.333†

SD; Standard deviation
**Mann-Whitney U test; †Fischer test*

Table 6. Detailed comparison of selected knowledge items by health literacy group

Domain	Response/item	Limited health literacy n (%)	Desired health literacy n (%)	*p* value*
Possible age of onset of inherited metabolic diseases	Prenatal period	12 (60.0%)	54 (50.5%)	0.433
	At birth	1 (5.0%)	11 (10.3%)	0.690
	Infancy	2 (10.0%)	19 (17.8%)	0.391
	Childhood	0 (0.0%)	7 (6.5%)	0.595
	Adulthood	0 (0.0%)	4 (3.7%)	1.000
	Any age	5 (25.0%)	40 (37.4%)	0.288

Methods used to diagnose inherited metabolic diseases	Amniocentesis	2 (10.0%)	18 (16.8%)	0.442
	Chorionic villus sampling	2 (10.0%)	25 (23.4%)	0.180
	Newborn screening programs	13 (65.0%)	71 (66.4%)	0.906
	Specialized laboratory techniques	9 (45.0%)	37 (34.6%)	0.373
	Routine laboratory tests	6 (30.0%)	32 (29.9%)	0.993
	Genetic analysis methods	4 (20.0%)	40 (37.4%)	0.134
Physicians who can evaluate diagnostic tests for inherited metabolic diseases	Any physician	2 (10.0%)	8 (7.5%)	0.657
	Neonatologist	6 (30.0%)	41 (38.3%)	0.479
	Pediatric metabolic disease specialist	9 (45.0%)	50 (46.7%)	0.887
	Pediatrician	4 (20.0%)	41 (38.3%)	0.116
	No opinion	4 (20.0%)	24 (22.4%)	0.810

** Fischer test*

4. Discussion

In this study, most pregnant women were classified as having desirable health literacy based on the MHELIP total score; however, their self-reported knowledge of both NBS and IMDs remained limited. Although many participants were familiar with some basic aspects of NBS, such as heel-prick blood sampling and the program's role in early diagnosis and treatment, their knowledge of IMDs, their diagnostic evaluation, and specialist interpretation of test results was less robust. In addition, while desired health literacy was associated with some favorable patterns, including greater reliance on healthcare professionals as information sources and better recognition of the main purpose of NBS, it was not associated with broader knowledge across all IMD-related domains. Taken together, these findings suggest that general health literacy alone may not be sufficient to ensure adequate understanding of rare and complex disorders during pregnancy, such as IMDs.

Among the 107 participants classified as having desired health literacy, 29.9% reported adequate knowledge of the NBS program and 26.4% reported adequate knowledge of IMDs, indicating a discrepancy between overall health literacy and condition-specific knowledge. This suggests that it is not accurate to interpret maternal health literacy solely from a screening context or a specific disease. In a study of 512 mothers with children under 5 years old, higher knowledge and more positive attitudes were associated with higher knowledge of prenatal tests and awareness of NBS diseases (9). In another study, Health literacy was strongly correlated with total NBS knowledge-attitude scores in the Turkish group compared with other ethnic groups (10). However, both studies used the Maternal Knowledge and Attitudes about Newborn Screening Survey (MKANSS), with one using the Health Literacy Scale (Short Form) (9) and the other using the Turkey Health Literacy Scale-32 (10). Furthermore, the samples in both studies did not specifically focus on pregnant women. Awareness of NBS and IMDs likely depends on whether these topics are specifically addressed during antenatal care and on the health literacy scale used. The use of the MHELIP scale, which is more closely associated with pregnancy, in this study may have contributed to this result and is a limitation.

A majority of participants correctly identified that the sample used in the screening program is obtained by collecting a few drops of blood from the newborn's heel, and most also recognized early diagnosis and early initiation of treatment as the program's main goals. A study conducted in Jordan found that mothers' knowledge of newborn screening was moderate, and that this knowledge contributed positively to their attitudes towards the test (11). However, in our study, 21 participants (16.5%) identified sample collection for further research as the purpose of the NBS program, and 1 participant said they would not have a heel prick test again. We cannot argue for a general opposition to screening based solely on this data, but as health professionals, we should question whether sufficient information is being provided about screening. A study from our country also found lower vaccine

hesitancy among participants in NBS (12). This relates to showing that preventive medicine practices concerning public health are interrelated, should be evaluated as a whole, and that we need to inform the public.

In this study, SMA was the most commonly recognized condition in the NBS. Similarly, in a study by Sert et al., SMA was the most frequently identified disease (9). In our country, SMA screening also began in 2022, and as of 2023, SMA screening was being conducted in 31 countries (13). New treatment modalities, greater public visibility, media coverage, or increased public discussion may reflect the awareness of SMA screening, compared with other screened conditions. This suggests that disease recognition may be influenced not only by structured health education but also by contemporary public discourse.

Women with desired health literacy were more likely to identify family physicians and nurses as sources of information and to recognize early diagnosis as the main purpose of the NBS. In a study conducted in our country, healthcare professionals were identified as the most trusted and most commonly cited sources of information (9). However, no significant between-group differences were found in self-reported knowledge of IMDs, perceptions of onset age, awareness of diagnostic approaches, or opinions regarding which physicians should interpret diagnostic tests. It suggests that even when health literacy is relatively high, knowledge about rare conditions may remain limited. On the other hand, the high frequency of internet use (40.9%) and social media use (33.9%) should be considered. A recent U.S.-based study described three educational strategies (online learning modules, a prenatal education pilot program, and a social media awareness campaign) and emphasized that these approaches showed promise in improving families' awareness, knowledge, and self-efficacy regarding newborn screening. The study also proposed a bidirectional family engagement model, highlighting the importance of involving families not only as recipients of information but also as active partners in the newborn screening process (13). In light of technological advancements, healthcare systems and professionals may consider developing and standardizing messages about screening programs and their content, in collaboration with the Ministry of Health or relevant associations, to ensure more consistent information for expectant families during the antenatal period.

The finding that most participants supported expansion of the NBS is also noteworthy. Although this view did not differ significantly between the limited and desired health literacy groups, the overall positive attitude suggests broad acceptance of preventive neonatal health interventions. Despite the widespread implementation of NBS in Europe, studies highlight a significant lack of standardization across countries regarding parental information, screened diseases, sampling timing, laboratory methods, and follow-up processes. The same study also notes that while some countries screen for only one disease, others screen for 30 (15). The ACMG expert panel identified 29 core conditions recommended for mandatory newborn screening and recognized 25 additional secondary conditions that could be detected through screening due to their relevance to the differential diagnosis, potential clinical significance, or incidental detection despite limited treatment options (16). Ongoing efforts in our country aim to expand the scope of the NBS (17).

This study has several limitations, including its cross-sectional design, , reliance on self-reported knowledge, and a single-center, single-city setting with a relatively small, highly educated population, which limits the generalizability of the results to the overall population. Additionally, health literacy was dichotomized into limited and desired categories because the number of participants in some of the original health literacy categories was insufficient for reliable subgroup analyses. However, it is significant because it assesses knowledge of both NBS and IMDs, it uses a standardized and validated scale (MHELIP), and it investigates the relationship between them.

5. Conclusion

This study demonstrates that while most pregnant women possess adequate general maternal health literacy, their knowledge of NBS and IMD remains limited. Desired maternal health literacy was associated with greater use of healthcare professionals as information sources and better recognition of the main purpose of NBS, but it did not translate into a broader understanding of IMDs. These results indicate that maternal health literacy alone is not sufficient to ensure adequate awareness of rare neonatal conditions. Future studies including educational interventions with pre- and post-training assessments may help clarify the effectiveness of antenatal education in improving parental awareness of NBS and IMDs.

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