RESEARCH PAPER

Awareness and Uptake of Newborn Screening in India: A Community-Based Cross-Sectional Study

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ABSTRACT

Background

Newborn screening (NBS) is an important preventive public health strategy for the early detection of congenital disorders and timely intervention. However, community-level awareness and uptake of NBS remain limited in many low- and middle-income countries, including India. This study assessed awareness, knowledge, uptake, and perceptions regarding newborn screening in a community-based population in India.

Methods

A community-based cross-sectional survey was conducted among 500 participants from urban and rural regions of North India between February 2025 and February 2026. Data were collected using a structured questionnaire assessing awareness, knowledge of screening timing, uptake, sources of information, and perceptions regarding NBS. Associations between sociodemographic factors and awareness were evaluated using chi-square tests and multivariable logistic regression analysis.

Results

Overall, 51.4% of participants were aware of newborn screening, while only 20.6% reported that their child had undergone screening. Correct knowledge regarding the recommended screening window of 24–72 hours after birth was observed in 36.0% of participants. Healthcare professionals were the primary source of information regarding NBS. Participants demonstrated highly favorable perceptions toward newborn screening and strong willingness to accept screening if adequately explained. Higher educational attainment and urban residence were independently associated with greater awareness of NBS.

Conclusions

Despite favorable public perceptions, substantial gaps remain in awareness, knowledge, and uptake of newborn screening in the community. Strengthening public education, improving counseling services, and integrating newborn screening into routine maternal and child healthcare programs may improve equitable uptake and support the expansion of newborn screening services in India.

Keywords: Newborn screening; preventive health services; public health; awareness; cross-sectional study

How to cite this article: Sharma A, Dutta AK, Vashist M, Siwach K, Kaushik A, Rani A. Awareness and Uptake of Newborn Screening in India: A Community-Based Cross-Sectional Study. Int J Drug Deliv Technol. 2026;16(59s): 226-233. DOI: 10.25258/ijddt.16.59s.22

Source of support: Nil

Conflict of interest: None

Introduction

Newborn screening (NBS) is a well-established public health strategy aimed at the early identification of serious congenital metabolic, endocrine, hematological, and genetic disorders that are not clinically apparent at birth but may lead to irreversible morbidity, neurodevelopmental impairment, or death if left untreated. Early diagnosis through systematic screening enables timely intervention, including dietary modification, pharmacological treatment, or long-term clinical monitoring, thereby substantially improving survival and quality of life outcomes [1].

In recent years, newborn screening has increasingly been recognized within the broader framework of public health, where biochemical and laboratory-based information is applied at the population level to support early disease detection and targeted interventions. Advances in screening technologies, including tandem mass spectrometry, have expanded the range of conditions detectable through newborn screening programs. Many high-income countries now routinely screen for more than 30–50 conditions and have emphasized harmonization of screening standards, quality assurance, and equitable access to services [1,2]. International public health agencies recognize early detection of congenital disorders as a cost-effective strategy to reduce childhood morbidity

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and long-term healthcare burden, particularly in low- and middle-income countries.

Despite these technological and policy advances, the implementation and effectiveness of newborn screening programs remain highly variable across regions. In many developing countries, challenges related to infrastructure, availability of trained personnel, funding, and follow-up systems persist. Importantly, beyond laboratory capacity, community awareness, parental understanding, and public acceptance are increasingly recognized as critical determinants of screening uptake and continuity of care [3,4]. Without adequate awareness and trust, even well-designed screening programs may fail to achieve population-level health benefits.

India bears a substantial burden of genetic and congenital disorders due to its large population, high birth rate, and sociocultural factors such as consanguinity in certain regions. It has been estimated that more than 1.5 million infants are born annually with birth defects in India, many of which are detectable and manageable through early newborn screening [5]. Although several states and private healthcare institutions have initiated screening programs for select conditions, a comprehensive nationwide newborn screening policy has not yet been uniformly implemented. As a result, screening practices remain fragmented, with marked disparities between urban and rural areas and between public and private healthcare sectors [6].

Recent policy discussions and pilot initiatives supported by national agencies have highlighted the feasibility and long-term benefits of expanding newborn screening services in India. However, these efforts have also underscored that infrastructural expansion alone is insufficient. Studies consistently report that limited public awareness, inadequate counseling, and misconceptions regarding screening procedures and outcomes act as major barriers to parental participation and follow-up adherence [7,8]. Addressing these challenges is central to translating advances in screening into effective public health practice.

Community-level awareness, knowledge, and perception therefore play a pivotal role in shaping attitudes toward newborn screening and its acceptance as a routine public health service. Evidence from community-based and hospital-linked surveys indicates that individuals who are better informed about the purpose, timing, and benefits of newborn screening demonstrate higher acceptance and trust in healthcare recommendations [3]. Conversely, lack of awareness may lead to delayed decision-making, refusal of screening, or poor compliance with confirmatory testing and long-term management protocols.

Although previous studies in India have examined laboratory outcomes, disease prevalence, and policy frameworks related to newborn screening, data assessing population-level awareness and perception across diverse age groups in the community remain limited. Understanding how different segments of the population perceive newborn screening is essential for designing targeted educational strategies, strengthening counseling services, and informing evidence-based policymaking within a public health framework.

Therefore, the present study was undertaken to assess the level of awareness, knowledge, uptake, and perception regarding newborn screening among 500 participants from different age groups. The study further examined awareness of recommended screening timing, sources of information, and associations between awareness and key sociodemographic factors, including age, gender, educational status, occupation, and place of residence. By identifying gaps in awareness and attitudes at the community level, this study aims to generate evidence that can support targeted public health interventions and inform strategies for the effective expansion of newborn screening services in the Indian healthcare context.

MATERIALS AND METHODS

Study Design and Setting

A community-based, cross-sectional survey was conducted to assess awareness, knowledge, and perception regarding newborn screening (NBS) among the general population. The study employed a structured questionnaire and was carried out over a defined study period among volunteers from different age groups. The cross-sectional design was chosen to capture prevailing levels of awareness and attitudes toward newborn screening at a single point in time. The study was conducted in community settings across urban and rural areas of Haryana and adjoining regions of North India between February 2025 and February 2026.

Study Participants

A total of 500 volunteers were included in the study. Participants were recruited using a convenience sampling approach from the community. Individuals belonging to different age groups and sociodemographic backgrounds were invited to participate in the survey.

Inclusion criteria included participants aged 15 years and above who were willing to participate voluntarily and provided informed consent prior to data collection. Participants who were unable to comprehend the questionnaire or declined consent were excluded from the study. Efforts were made to include participants from both urban and rural areas to ensure diversity in representation.

Survey Instrument and Data Collection

Data were collected using a pre-designed, structured questionnaire developed to evaluate awareness, knowledge, and perception related to newborn screening. The questionnaire was developed based on previously published literature and reviewed by subject experts in public health and genetics to ensure content validity. It was pilot tested on 20 participants prior to final data collection to assess clarity and comprehension.

The questionnaire consisted of multiple sections:

- Sociodemographic characteristics, including age group, gender, educational status, occupation, and place of residence.
- Awareness and knowledge of newborn screening, including whether participants had heard of NBS, awareness of recommended timing for screening, and prior exposure to information about NBS.
- Uptake-related information, including whether newborn screening had been performed for their child, where applicable.
- Source of information, such as healthcare professionals, media, or other channels. Perception and attitude toward newborn screening, assessed using Likert-scale statements addressing perceived importance of NBS, willingness to undergo screening if adequately informed, trust in healthcare recommendations, acceptance of government-supported screening, and perceived barriers such as lack of awareness.
- The questionnaire was administered through interviewer-based interactions in a language understandable to the participants to ensure inclusion across different educational levels. Responses were recorded anonymously to ensure confidentiality. Participants were encouraged to respond honestly, and no personal identifiers were collected.

Ethical Considerations

The study was conducted in accordance with the ethical principles of the Declaration of Helsinki. Participation was voluntary, and informed consent was obtained from all participants prior to data collection. The survey was anonymous, and no identifiable personal or clinical information was collected. Confidentiality of participant responses was strictly maintained throughout the study. According to applicable institutional guidelines for anonymous questionnaire-based surveys involving voluntary adult participants, formal ethics committee approval was not required.

Statistical Analysis

Collected data were entered into a spreadsheet and checked for completeness and consistency prior to analysis. Data analysis was performed using IBM SPSS Statistics version 26. Categorical variables were summarized using frequencies and percentages. Associations between sociodemographic variables and awareness or perception of newborn screening were assessed using appropriate statistical tests, including the chi-square test. For perception-related items measured on a Likert scale, descriptive summary measures were calculated, and comparisons across groups were performed where applicable. Variables with $p < 0.20$ in univariate analysis were included in the multivariable logistic regression model to identify factors independently associated with awareness. A p-value of less than 0.05 was considered statistically significant.

The study was reported in accordance with the STROBE guidelines for cross-sectional studies.

RESULTS

Sociodemographic characteristics of the study participants

A total of 500 participants were included in the study. The age distribution indicated that the largest proportion of participants belonged to the 46–55 years age group (163, 32.6%), followed by those aged 15–25 years (142, 28.4%), 26–35 years (119, 23.8%), and 36–45 years (76, 15.2%). Female participants constituted nearly two-thirds of the study population (326, 65.2%), while 174 participants (34.8%) were male.

With regard to educational status, more than half of the participants had attained graduate-level education or higher (276, 55.2%), whereas 88 participants (17.6%) were illiterate. Equal proportions of participants had completed primary education (68, 13.6%) and secondary education (68, 13.6%). In terms of place of residence, the majority of participants were from rural areas (357, 71.4%), while 143 participants (28.6%) resided in urban settings (Table 1).

Table 1. Sociodemographic characteristics of the study participants (n = 500)

Variable	Category	Frequency (n)	Percentage (%)
Age group (years)	15–25	142	28.4
	26–35	119	23.8
	36–45	76	15.2
	46–55	163	32.6
Gender	Male	174	34.8
	Female	326	65.2
Education	Illiterate	88	17.6
	Primary	68	13.6
	Secondary	68	13.6
	Graduate & above	276	55.2
Residence	Urban	143	28.6
	Rural	357	71.4

Awareness and uptake of newborn screening

Overall, 257 participants (51.4%) reported that they were aware of newborn screening, while 243 participants (48.6%) indicated that they had no prior awareness.

Uptake of newborn screening was assessed among participants who reported having children (n = 500). Among them, only 103 participants (20.6%) reported that their child had undergone newborn screening. A large proportion of participants stated that newborn screening had not been performed for their child (342, 68.4%), while 55 participants (11.0%) were unsure about the screening status.

Assessment of knowledge regarding the recommended timing of newborn screening revealed that only 180 participants (36.0%) correctly identified that screening should be conducted within 24–72 hours after birth. In contrast, 54 participants (10.8%) incorrectly believed that screening is performed at birth, and more than half of the participants (266, 53.2%) reported that they did not know the appropriate timing for newborn screening (Table 2).

Table 2. Awareness and uptake of newborn screening among participants

Variable	Response	Frequency (n)	Percentage (%)
Awareness of newborn screening	Yes	257	51.4
	No	243	48.6
Child received newborn screening	Yes	103	20.6
	No	342	68.4
	Not sure / Maybe	55	11.0
Knowledge of recommended timing (24–72 h)	Correct	180	36.0
	Incorrect (at birth)	54	10.8
	Don't know	266	53.2

Sources of information regarding newborn screening

Among the participants who were aware of newborn screening (n = 257), healthcare professionals were the most commonly reported sources of information. Genetic counselors were identified as the primary source by 146 participants (56.8%), followed by media or internet sources reported by 66 participants (25.7%). Information obtained directly from doctors was reported by 43 participants (16.7%), while 24 participants (9.3%) indicated that they had received information from both doctors and genetic counselors. Additionally, 31 participants (12.1%) reported other sources of information. As participants were allowed to select more than one source, the total percentage exceeds 100% (Table 3).

Table 3. Sources of information regarding newborn screening (among participants aware of NBS, n = 257)

Source of information	Frequency (n)	Percentage (%)	Source of information
Doctor	43	16.7	Doctor
Genetic counselor	146	56.8	Genetic counselor
Doctor & genetic counselor	24	9.3	Doctor & genetic counselor
Media / Internet	66	25.7	Media / Internet
Others	31	12.1	Others

Perception toward newborn screening

Overall, participants demonstrated a highly positive perception toward newborn screening. A large majority of respondents strongly agreed (380, 76.0%) or agreed (107, 21.4%) that newborn screening is important for every newborn, with only a small proportion expressing neutral or negative views.

Similarly, most participants indicated willingness to accept newborn screening if it were properly explained, with 377 participants (75.4%) strongly agreeing and 98 participants (19.6%) agreeing with this statement. High levels of trust in healthcare providers were also observed, as 385 participants (77.0%) strongly agreed and 87 participants (17.4%) agreed that they trust healthcare professionals regarding newborn screening.

Support for policy-level intervention was evident, with 389 participants (77.8%) strongly agreeing and 93 participants (18.6%) agreeing that newborn screening should be provided free of cost in government hospitals. Additionally, a substantial proportion of participants perceived lack of awareness as a major barrier to newborn screening uptake, with 409 participants (81.8%) strongly agreeing and 57 participants (11.4%) agreeing with this statement. Overall, neutral and negative responses were minimal across all perception-related statements (Table 4).

Table 4. Perception toward newborn screening among participants (n = 500)

Statement	Strongly Agree n (%)	Agree n (%)	Neutral n (%)	Disagree n (%)	Strongly Disagree n (%)
NBS is important for every newborn	380 (76.0)	107 (21.4)	10 (2.0)	3 (0.6)	0 (0.0)
Would accept NBS if properly explained	377 (75.4)	98 (19.6)	18 (3.6)	7 (1.4)	0 (0.0)
Trust healthcare providers regarding NBS	385 (77.0)	87 (17.4)	20 (4.0)	8 (1.6)	0 (0.0)
NBS should be free in government hospitals	389 (77.8)	93 (18.6)	14 (2.8)	4 (0.8)	0 (0.0)
Lack of awareness is a major barrier	409 (81.8)	57 (11.4)	20 (4.0)	14 (2.8)	0 (0.0)

Factors independently associated with awareness of newborn screening

Multivariable logistic regression analysis was performed to identify factors independently associated with awareness of newborn screening. After adjustment for relevant sociodemographic variables, age, education, and place of residence emerged as significant determinants of awareness.

Compared with participants aged 15–25 years, those in the 36–45 years age group had significantly higher odds of being aware of newborn screening (AOR = 1.92, 95% CI: 1.11–3.33, p = 0.02), as did participants aged 46–55 years (AOR = 1.56, 95% CI: 1.01–2.42, p = 0.04). Participants aged 26–35 years did not show a statistically significant difference in awareness compared with the reference group.

Educational status showed a strong and graded association with awareness of newborn screening. Compared with illiterate participants, those with primary education had higher odds of awareness (AOR = 2.11, 95% CI: 1.10–4.03, p = 0.02), while participants with secondary education (AOR = 3.02, 95% CI: 1.58–5.78, p < 0.001) and graduate-level education or above (AOR = 4.87, 95% CI: 2.88–8.24, p < 0.001) demonstrated progressively greater awareness.Place of residence was also independently associated with awareness, with urban residents showing significantly higher odds of awareness compared with rural residents (AOR = 1.94, 95% CI: 1.29–2.92, p = 0.001). In contrast, gender was not independently associated with awareness of newborn screening (AOR = 1.08, 95% CI: 0.74–1.58, p = 0.69). These findings indicate that educational attainment and urban residence are key predictors of awareness, even after controlling for other sociodemographic factors (Table 5).

Table 5. Factors independently associated with awareness of newborn screening (n = 500)

Variable	Category	Adjusted Odds Ratio (AOR)	95% Confidence Interval	p-value
Age group (years)	15–25	Reference	—	—
	26–35	1.38	0.88–2.17	0.15
	36–45	1.92	1.11–3.33	0.02
	46–55	1.56	1.01–2.42	0.04
Gender	Male	Reference	—	—
	Female	1.08	0.74–1.58	0.69
Education	Illiterate	Reference	—	—
	Primary	2.11	1.10–4.03	0.02
	Secondary	3.02	1.58–5.78	<0.001
	Graduate & above	4.87	2.88–8.24	<0.001
Residence	Rural	Reference	—	
	Urban	1.94	1.29–2.92	0.001

DISCUSSION

The present community-based cross-sectional study assessed awareness, knowledge, uptake, and perceptions regarding newborn screening (NBS) among 500 participants from diverse sociodemographic backgrounds in North India. The findings demonstrate that although perceptions toward newborn screening were highly favorable, important gaps persist in awareness, procedural knowledge, and actual uptake of screening services. These findings highlight continuing public health challenges in the implementation and equitable utilization of newborn screening programs in India.

Newborn screening is increasingly recognized as an essential component of preventive public health because early identification of congenital metabolic,

endocrine, hematological, and genetic disorders enables timely intervention and reduces long-term morbidity and mortality [1,3]. Despite technological advances and expanding screening panels in many countries, successful implementation of NBS depends not only on laboratory infrastructure but also on public awareness, parental understanding, and accessibility of healthcare services [3,7]. The present study contributes important community-level evidence from India, where data on public awareness and uptake of newborn screening remain relatively limited.

In this study, approximately half of the participants reported prior awareness of newborn screening. Although this level of awareness is higher than that reported in some resource-limited settings, it remains substantially lower than in countries where NBS has been integrated into routine neonatal healthcare services [1,3]. Similar findings have been reported in previous studies from low- and middle-income countries, where limited public education, inadequate counseling, and restricted healthcare access were identified as major barriers to newborn screening awareness and participation [3,9]. In India, fragmented implementation of screening programs across states and healthcare sectors may further contribute to inconsistent public understanding of NBS [6,8].

A notable finding of the present study was the low reported uptake of newborn screening, with only one-fifth of participants indicating that their child had undergone screening. This observation suggests that favorable attitudes alone may not translate into utilization of services. Previous Indian studies have similarly reported limited uptake of NBS despite growing recognition of its clinical and public health importance [6,11]. Several factors may contribute to this discrepancy, including lack of routine integration of NBS into public healthcare services, limited counseling during antenatal and postnatal care, financial constraints, and inadequate awareness regarding the benefits of early diagnosis. These findings emphasize the need for strengthening implementation frameworks and ensuring equitable access to screening services, particularly in underserved communities.

Knowledge regarding the recommended timing of newborn screening was also limited in the study population. Only about one-third of participants correctly identified the recommended screening window of 24–72 hours after birth, while more than half were uncertain regarding appropriate timing. Procedural knowledge is critical for effective implementation because delayed or missed screening may reduce the effectiveness of early detection and intervention programs [12]. Similar gaps in practical knowledge have been reported in previous awareness studies involving preventive health services and neonatal care [11,13]. The findings indicate that educational interventions should focus not only on the importance of newborn screening but also on practical aspects such as timing, follow-up testing, and interpretation of results.

Healthcare professionals were identified as the primary source of information regarding newborn screening, with genetic counselors contributing substantially to participant awareness. This finding is consistent with previous evidence highlighting the important role of healthcare providers in influencing parental decision-making and improving acceptance of preventive screening services [4,13]. Counseling provided by trained professionals has been shown to improve understanding, reduce misconceptions, and enhance trust in healthcare recommendations [13]. However, the relatively limited contribution of mass media and digital platforms observed in the present study suggests that public awareness campaigns related to newborn screening remain inadequate. Expanding community-based education through public health programs, digital media, and primary healthcare systems may improve awareness at the population level.

Participants in the present study demonstrated highly favorable perceptions toward newborn screening. Most respondents considered NBS important for newborn health and expressed willingness to undergo screening if adequately informed. High levels of trust in healthcare providers and strong support for free screening services in government hospitals were also observed. Similar positive attitudes toward newborn screening have been reported in previous studies examining parental perceptions and preventive healthcare acceptance [13]. These findings suggest that the major barriers to NBS implementation in India may be related less to public resistance and more to gaps in awareness, counseling, and healthcare delivery systems.

The multivariable logistic regression analysis demonstrated that higher educational attainment and urban residence were independently associated with greater awareness of newborn screening. Participants with graduate-level education showed significantly higher odds of awareness compared with illiterate participants, indicating a strong social gradient in access to health-related information. Similar associations between educational status and awareness of preventive health interventions have been consistently reported in both Indian and international public health literature [9,11]. Urban residents were also more likely to be aware of NBS than rural participants, likely reflecting disparities in healthcare infrastructure, availability of specialized services, and exposure to healthcare information. These findings highlight the need for targeted public health strategies focusing on rural populations and individuals with lower educational attainment.

From a public health perspective, the findings of this study have important implications for the expansion of newborn screening services in India. The coexistence of favorable public perception and low screening uptake suggests that strengthening healthcare delivery systems and community-level education could substantially improve implementation outcomes. Integration of newborn screening counseling into routine antenatal and postnatal healthcare services may improve awareness and facilitate informed decision-

making among parents. In addition, inclusion of NBS within publicly supported maternal and child health initiatives could improve accessibility and reduce disparities in service utilization.

The study has several limitations that should be acknowledged. First, the cross-sectional design limits causal interpretation of the observed associations. Second, the use of convenience sampling may affect generalizability of the findings beyond the study population. Third, responses were self-reported and therefore subject to recall bias and reporting bias. Additionally, the study was conducted primarily in North India, and awareness levels may differ across other geographical regions and healthcare settings. Despite these limitations, the study provides valuable community-based evidence regarding awareness and uptake of newborn screening and contributes important data relevant to preventive public health planning in India.

In conclusion, the present study demonstrates that although community perception toward newborn screening is highly favorable, substantial gaps remain in awareness, procedural knowledge, and actual uptake of screening services. Strengthening public education, improving counseling by healthcare professionals, and integrating newborn screening into routine public healthcare systems may improve equitable uptake and support the effective expansion of newborn screening services in India.

CONCLUSION

This community-based cross-sectional study assessed awareness, knowledge, and uptake of newborn screening among 500 participants from diverse sociodemographic backgrounds. The findings indicate that while community perception toward newborn screening is highly favorable, this positive attitude does not consistently translate into practice, as reflected by the low uptake of screening and limited knowledge regarding recommended screening timing.

Educational status and place of residence emerged as significant determinants of awareness, highlighting persistent disparities in access to healthcare information and screening services. In contrast, gender was not independently associated with awareness, suggesting that informational gaps are widespread across the population.

Overall, the study provides important public health insights by identifying critical gaps in awareness, knowledge, and uptake that must be addressed to improve the effectiveness of newborn screening programs. Integrating newborn screening education into routine maternal and child health services, strengthening counseling by healthcare professionals, and ensuring accessible, publicly supported screening programs may enhance equitable uptake and support the effective expansion of newborn screening within public health systems.

Declaration

Consent to Participate Statement

Informed consent was obtained from all participants prior to participation in the study. Participation was voluntary, and respondents were informed of their right to decline or withdraw at any stage without consequences.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

Funding Sources

This study was not supported by any sponsor or funder.

Data Availability Statement

The datasets generated and analyzed during the current study are available from the corresponding author upon reasonable request, subject to ethical and confidentiality considerations.

Acknowledgement

The authors sincerely thank all participants for providing their consent and for their voluntary participation in the survey. The authors also acknowledge the support of community members who facilitated data collection.

Author Contributions

A.S. (Anuradha Sharma) conceptualized the study, contributed to questionnaire design, and was primarily responsible for data collection. **A.K.D. (Amit Kumar Dutta)** supervised the study, performed the statistical analysis, interpreted the results, and served as the corresponding author. **M.V. (Minakshi Vashist)** contributed to study design, assisted in data interpretation, and critically reviewed the manuscript. **K.S. (Kiran Siwach)** contributed to data collection, literature review, manuscript preparation, and assisted in data interpretation. **A.K. (Amit Kaushik)** contributed to methodology development, data analysis support, and critical revision of the manuscript. **A.R. (Anisha Rani)** assisted in data collection, literature review. All authors contributed to manuscript drafting or revision, approved the final version of the manuscript, and agree to be accountable for all aspects of the work.

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