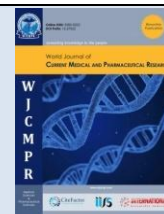




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BRIDGING THE GAP: AWARENESS AND UTILISATION OF NEWBORN SCREENING IN A SOUTH INDIAN TERTIARY CARE SETTING

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ARTICLE HISTORY	ABSTRACT
Received on: 14-07-2026 Revised on: 03-08-2026 Accepted on: 17-09-2026	Introduction: Newborn screening (NBS) enables early identification of conditions such as inborn errors of metabolism, congenital hypothyroidism, and G6PD deficiency. Timely diagnosis and intervention can prevent irreversible disability and death, improve long-term health outcomes, and reduce healthcare burden. However, limited parental awareness remains a challenge to effective NBS.
Keywords: Newborn screening, Antenatal mother, Postnatal mother, awareness, sociodemographic factors.	Aims: To assess awareness and utilisation of newborn screening services among antenatal and postnatal mothers.
Corresponding Author Dr. Manasi Garg Professor, Department of Paediatrics, Mahatma Gandhi Medical College and Research Institute, Sri Balaji Vidyapeeth (Deemed to be University), Puducherry, India.	Settings and Design: A hospital-based cross-sectional study was conducted over 18 months among antenatal and postnatal mothers attending a tertiary care centre in South India.
	Methods and Materials: Antenatal mothers attending the Obstetrics outpatient department for routine antenatal care and postnatal mothers attending the Paediatric outpatient department for follow-up until three months of age were included after written informed consent. Data were collected using a pre-tested and pre-validated questionnaire administered by the principal investigator.
	Statistical Analysis: Frequencies and percentages were used for descriptive analysis. Chi-square or Fisher's exact test was applied to assess associations between categorical variables.
	Results: Awareness regarding newborn screening was reported by 27.5% of antenatal mothers and 71.1% of postnatal mothers. Awareness was significantly associated with parity, infant gender, family type, monthly income, and type of house. Despite limited awareness, utilisation of NBS services was 100%. This high utilisation was attributed to the institutional policy of screening all neonates born at the centre, irrespective of prior parental awareness or counselling.
	Conclusion: Awareness of NBS was particularly low among primiparous mothers, highlighting the need for targeted counselling. Early counselling can facilitate informed decision-making and reduce parental anxiety. Hospital-based educational sessions, antenatal counselling, and social-media initiatives may improve parental awareness of NBS.

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INTRODUCTION

Newborn screening (NBS) helps identify disorders like inborn errors of metabolism, hypothyroidism, G6PD deficiency, and others. Early diagnosis of these conditions and prompt intervention can prevent irreversible damage, disability, or death, thereby improving long-term health outcomes and reducing the burden on healthcare systems. It is a preventive public health program designed to identify infants with specific congenital, metabolic, hormonal, and

genetic disorders at an early stage. By using a simple heel-prick blood sample, usually collected within the first 48–72 hours of life, NBS helps detect conditions such as congenital hypothyroidism, congenital adrenal hyperplasia, glucose-6-phosphate dehydrogenase (G6PD) deficiency, phenylketonuria, cystic fibrosis, sickle cell disease, and other inborn errors of metabolism. These disorders are often asymptomatic at birth but, if not detected, can lead to irreversible morbidity, long-term disability, or even death.

The cornerstone of NBS is that early diagnosis followed by timely intervention can significantly improve health outcomes, reduce disease burden, and save healthcare costs in the long term [1,2].

The rationale behind NBS lies in its potential to alter the natural course of serious but treatable conditions. For example, congenital hypothyroidism is one of the leading preventable causes of intellectual disability [3], while timely detection and treatment of congenital adrenal hyperplasia can prevent life-threatening adrenal crises(4). In the absence of screening, these disorders are often diagnosed only after significant clinical manifestations, by which time the damage is frequently irreversible. In many developed nations, NBS has become a standard practice and is integrated into national health policies, thereby contributing significantly to reduced infant morbidity and mortality. Beyond clinical benefits, NBS also plays a key role in reducing the psychosocial and economic burden on families and health systems by preventing lifelong disabilities that would otherwise require extensive care [5].

Due to its impact on healthcare, NBS was accepted as an essential preventive public health program and is now implemented worldwide. However, in India, NBS has not been included in government policy, nonetheless realising its importance, many private hospitals and laboratories have initiated NBS as a screening policy. Despite its proven benefits, newborn screening in India remains limited and unevenly implemented, with significant gaps between the public and private healthcare sectors.

METHODOLOGY

To assess the awareness and utilization of the Newborn Screening (NBS) program among expectant and postnatal mothers respectively, we conducted a hospital-based cross-sectional cohort study at a tertiary care centre from July 2023 to January 2025. The study population included antenatal mothers with a gestational age over 28 weeks and postnatal mothers up to three months postpartum, attending the outpatient and inpatient services of Obstetrics and Paediatrics.

A total of 830 participants, 415 in each group were recruited based on sample size calculation using prior prevalence data of NBS awareness. Consecutive sampling was used until the required sample size was reached.

Data collection was carried out using a pre-tested, semi-structured questionnaire administered by the principal investigator. The questionnaire comprised three sections: (i) socio-demographic details, (ii) awareness of NBS (purpose, timing of sample collection, conditions screened, possibility of false positives), and (iii) utilisation of NBS services and influencing factors. After administering the questionnaire, antenatal mothers received counselling about NBS, while postnatal mothers were asked about prior utilisation of NBS for their infants.

The primary outcome measured was the level of awareness in antenatal mothers and utilisation of NBS in postnatal mothers, while secondary outcomes included factors associated with awareness and utilization such as education, socioeconomic status, parity, and prior counselling. Data were entered in Microsoft Excel and analysed using SPSS v21. Descriptive statistics were expressed as frequencies,

percentages, means, and standard deviations. Associations between categorical variables were tested using Chi-square or Fisher's exact test, while multivariate regression was applied to identify independent predictors of awareness and utilization. A p-value <0.05 was considered statistically significant.

RESULTS

Table 01 shows the distribution of socio-demographic characteristics among the study participants, giving a total sample size of 830. The variables assessed include educational qualification, occupational category, type of family, type of house, socio-economic status, and monthly household income. The data are expressed as absolute numbers and corresponding percentages. This profile helps to understand the background characteristics of the mothers and provides context for interpreting their responses regarding awareness and utilisation of newborn screening services. Among the mothers, 223 (27.4%) were graduates, 121 (14.6%) had completed diploma courses, 252 (30.4%) had studied up to high school, and 234 (28.2%) had studied up to middle school. A total of 124 (14.9%) were skilled workers, 199 (23.9%) were engaged in agriculture and fishery, 192 (23.1%) in craft-related works, 142 (17.1%) were plant and machine operators, and 106 (12.8%) were in elementary occupations. Among the mothers, 222 (6.7%) belonged to nuclear families, 476 (57.3%) to joint families, and 132 (15.9%) to extended families. Among the mothers, 484 (58.3%) lived in pucca houses while 346 (41.7%) lived in semi-pucca houses. 452 (54.5%) belonged to the upper middle class and 378 (45.5%) to the lower middle class. Among the mothers, 136 (16.4%) reported a monthly income between ₹1,26,300–63,182, 666 (80.2%) between ₹63,182–47,266, 26 (3.1%) between ₹47,266–31,591, and 2 (0.2%) between ₹18,953–31,590.

Table 01: Socio-demographic profile of the mothers (n=830)

Variables	Number (n=830)	Percentage (%)
Education		
Graduate	223	26.9
Diploma	121	14.6
High school	252	30.4
Middle school	234	28.2
Occupation		
Skilled workers	124	14.9
Agriculture and fishery	199	24.0
Craft related works	192	23.1
Plant and machine operators	142	17.1
Elementary occupation	106	12.8
Type of family		
Nuclear	222	26.7
Joint	476	57.3
Extended	132	15.9

Type of house		
Pucca house	484	58.3
Semi pucca house	346	41.7
Socio-economic status		
Upper middle class	452	54.5
Lower middle class	378	45.5
Monthly income		
₹1,26,300–63,182	136	16.4
₹63,182–47,266	666	80.2
₹47,266–31,591	26	3.1
₹18,953–31,590	2	0.2

The table 02 illustrates the awareness, perceptions, and attitudes of antenatal mothers regarding the Newborn Screening (NBS) Programme. Most of the participants 346 (83.4%) had not received prior counselling on NBS, suggesting a significant gap in education and awareness. Furthermore, 301 (72.5%) respondents were unaware of the NBS programme before their newborn underwent testing, indicating that many parents are introduced to the concept only after childbirth. Less than half of the mothers, 175 (42.2%) understood the rationale for newborn screening, suggesting that around 240 (57.8%) mothers were unaware of the critical role of early screening in identifying congenital disorders. A significant proportion of participants 324 (78.1%) were unaware that NBS could yield false-positive results, which could lead to unnecessary anxiety among parents if not properly communicated. This highlights the need for better pre- and post-test counselling to manage parental expectations and improve understanding of test outcomes. Around 288 (69.4%) participants believed that genetic counselling should be provided both before and after NBS, reinforcing the need for structured counselling services to address parental concerns and enhance understanding. Around 173 (41.7%) considered NBS to be expensive, indicating that affordability may still be a concern for some people. Ensuring accessibility and affordability of NBS testing could help improve its acceptance and coverage. Despite gaps in knowledge and communication, 261 (62.9%) participants expressed willingness to recommend NBS to other parents, suggesting a generally positive perception about the screening programme. However, the 154 (37.1%) expressed their unwillingness to recommend the newborn screening process.

Table 02: Awareness and perceptions regarding Newborn screening among antenatal mothers

Variables	Number (n = 415)	Percentage (%)
Have you received NBS counselling previously?		
No	346	83.4
Yes	69	16.6
Were you aware of the Newborn screening programme before the test?		
No	301	72.5
Yes	114	27.5

Did you know why the NBS is performed?		
No	240	57.8
Yes	175	42.2
Did you know that the blood sample for NBS must be taken as soon as the 2 nd or 3 rd day after the delivery?		
No	113	27.2
Yes	302	72.8
Did you know that NBS could have false-positive results?		
No	324	78.1
Yes	91	21.9
Do you think that it is important to have genetic counselling before and after the newborn screening?		
No	127	30.6
Yes	288	69.4
Do you think the Newborn screening programme test is too expensive?		
No	242	58.3
Yes	173	41.7
Would you recommend Newborn screening programme to other parents?		
No	154	37.1
Yes	261	62.9

Table 03: Awareness and perceptions regarding Newborn screening among postnatal mothers

Variables	Number (n = 415)	Percentage (%)
Were you aware of the Newborn screening programme before the test?		
Yes	295	71.1
No	120	28.9
Did you know why the NBS is performed?		
No	206	49.6
Yes	209	50.4
Did you know that NBS could have false-positive results?		
No	120	28.9
Yes	295	71.1
Do you think NBS is essential and is it important to have genetic counselling before and after the newborn screening?		
No	120	28.9
Yes	295	71.1
Do you think the Newborn screening programme test is too expensive?		
No	130	31.3
Yes	285	68.7
Would you recommend Newborn screening programme to other parents?		
No	180	43.4
Yes	235	56.6

The table 03 presents the awareness and perceptions regarding the NBS program among postnatal mothers. Out of 415 postnatal mothers, 295 (71.1%) were aware of the NBS program before the test with information primarily provided by neonatologists. In contrast, 120 (28.9%) were

unaware of the program before their child underwent screening. When asked if they knew which conditions were included in NBS testing, 236 (56.9%) responded negatively, whereas 179 (43.1%) had knowledge of the conditions screened. Awareness regarding the purpose of NBS was almost evenly split, with 209 (50.4%) postnatal women knowing why the test was performed and 206 (49.6%) being unaware. Regarding the timing of sample collection, 259 (62.4%) postnatal mothers knew when the sample should be taken, while 156 (37.6%) was unaware. Around 295 (71.1%) women were aware that NBS could yield false-positive results, while 120 (28.9%) were not. When asked about the necessity of NBS and the importance of genetic counselling before and after the test, 295 (71.1%) respondents considered it essential, while 120 (28.9%) did not. Cost was a concern for many participants, with 285 (68.7%) perceiving the test as too expensive and 130 (31.3%) not considering it so. Among those 285 women who found the test costly, 295 (71.1%) felt that the government should provide it free of charge for all newborns. More than half, 235 (56.6%) mothers expressed willingness to recommend the NBS program to other parents, while 180 (43.4%) were not willing. These findings highlight the need for improved awareness and education regarding the significance, benefits, and affordability of the NBS program.

DISCUSSION

This cross-sectional study, conducted in Puducherry, assessed the awareness and use of the Newborn Screening (NBS) program among antenatal (ANC) and postnatal (PNC) mothers. The study's findings provided crucial insights into the knowledge and practices of the NBS program, as well as the gaps in maternal education, healthcare delivery, and awareness dissemination strategies. In our study, the awareness of NBS among antenatal mothers was low (27.5 %). This is similar to a survey conducted by Vohr et al [6], who found that only 21% of antenatal mothers were aware of the NBS program [6]. Similarly, Jatto et.al [7] from Africa and Fujii et al [8] from Japan, reported that 27% and 26.6% of mothers were aware of the NBS program during their antenatal visits, respectively. This was in strike contrast to studies conducted by Yin et al [9] from China and Wilaiwongsathien et al(10) from Thailand, who reported higher levels of awareness and knowledge at 72.9% and 59.6%, respectively.

Out of all the factors influencing awareness in antenatal mothers; parity, birth order of children and household income were found to be significantly associated. We found that 46.9% of multiparous women were aware, compared to 0.6% of primiparous women; the difference was statistically significant. This difference can be attributed to prior interactions with healthcare services during previous pregnancies, increased familiarity with maternal and child health programs, or greater exposure to healthcare professionals.

A similar pattern was observed with higher birth order, where 9.5% mothers with no children were aware of NBS, compared to 46.7% mothers with one child, 40.5% with two children, and both mothers with three children (100%)

($p = 0.01$). This reinforces the idea that every consecutive childbirth increases exposure to newborn healthcare services, including NBS awareness. We also found that the higher the monthly income, the higher the awareness about NBS. This indicates that higher-income mothers are more likely to have prior knowledge of NBS, potentially due to greater access to healthcare services, educational resources, and antenatal counselling. A study by Yin et al [9] from China found that parents with lower income and education levels, as well as those with no children or at least two children, had poorer awareness and knowledge of NBS. In our study, the education of the mother was not associated with significant awareness, which was the study by Yin et al [9], where parents with senior high school/technical secondary school education have higher awareness ($p < .01$).

Only 16.6% of antenatal women received counselling about the NBS test, and all of them were multiparous. This complete absence of NBS counselling among primiparous mothers underscores a critical gap in antenatal education, emphasising the need for targeted efforts to introduce NBS awareness early in the maternal care continuum. In the study conducted by Detmar et al [11], women were not well informed about NBS and would prefer to receive information during pregnancy.

Among the post-natal mothers, 71.1% were aware of the NBS program before the test and the information was primarily provided by neonatologists (71%). Stuart et al. highlighted that parents received information about newborn screening from various sources, including midwives, official NHS literature, non-NHS literature, and the internet. Midwife was cited as the most important stratifies source of information by 76.2% parents followed by official leaflets in 14% at various stages, from preconception through the infant's first few months of life [12]. In the study by Louise Moody et al, 61.2% remembered being given information about screening before the blood spot was taken [13]. They suggested that the NBS counselling should be done at 8-week antenatal appointment, after birth and just before the test. Franková et al. [14] found that multiparous mothers had significantly better awareness compared to primiparous mothers, and 77% of the mothers received the information from a nurse or physician.

A statistically significant association ($p < 0.05$) was observed between NBS awareness and various socio-demographic factors. In our study, higher awareness was observed in postnatal mothers from joint families (34.0%), extended families (40.9%), higher-income groups (47.1%), urban residence (74.1%), living in pucca house (32.6%), multiparity and those having female babies (50.0%) and all these factors were statistically significant ($p < 0.05$). In the study from China [9] which included expectant parents and parents of children aged 0-3 years found that older and younger participants, males, those living in the village, with lower income and education levels, and those with no children or at least two children had poorer awareness and knowledge of NBS. These findings highlight the role of socio-economic and demographic factors in shaping awareness of NBS and suggest a need for targeted educational interventions, particularly for lower-income, rural, and first-time mothers.

Studies from developed countries, such as Australia and the United States, have demonstrated significantly higher awareness levels ($p < 0.05$) among postnatal mothers, mainly due to standardized education modules, an informed consent process, and government mandates [1,15]. In the study conducted by Al-Sulaiman et al., 91.1% of women considered NBS to be very important and helpful, but only 36.4% were aware that NBS was done for detecting genetic disorders [16].

The difference in awareness rates among antenatal and postnatal mothers can be attributed mainly to multiparity, indicating a lack of integration of NBS education in routine antenatal care and the absence of a mass media campaign to promote such services [7].

In the present study, the utilisation of newborn screening (NBS) services was 100%. This high utilisation rate is attributable to the institutional policy of performing NBS for all neonates delivered in the hospital, irrespective of prior parental awareness or counselling.

Around 42.2% of the antenatal mothers were aware of NBS, primarily due to the higher educational status and occupation of the mothers. Similarly, a study conducted by Fitzgerald et al [17] states that lower educational status (OR 1.79; 95% CI 1.02, 3.15) was independently associated with poor NBS knowledge for all women in the antenatal period.

A study conducted by Audrey Tluczekau et al. found that only a small percentage of parents recalled receiving information about NBS from health professionals before birth (13%), with even fewer recalling receiving written literature (4%). Additionally, about (6%) of parents were either not informed or did not remember receiving any information about NBS. This suggests that prenatal care providers may not be consistently discussing NBS as part of routine education for expectant parents [18].

A study conducted by Bathra Tariq et al. shows that the intervention was highly effective in educating participants about NBS, resulting in a significant increase in awareness from 20% to 98%. The rise in willingness from 57.7% to 78.9% after awareness indicates a direct link between knowledge and behavioral intent. The p-value (<0.0001) confirms this change is statistically significant.

Among the antenatal mothers and postnatal mothers, 21.9% and 71.1%, respectively, were aware of false-positive results. In the study conducted by Tarini et al [19] only 1.6% of mothers knew that they had a false-positive newborn screening (NBS) result. These findings highlight the need for targeted educational interventions, particularly for first-time mothers and those from nuclear families, to improve understanding of the nuances of NBS, including the possibility of false-positive results [19].

Among antenatal women, 69.4% and 71.1% of postnatal mothers believed that genetic counselling should be provided before and after the NBS, reinforcing the need for structured counselling services to address parental concerns and enhance understanding.

NBS was considered expensive by 41.7% and 68.7% of antenatal and postnatal mothers respectively, indicating that affordability is still be a concern for our developing economy. A similar study conducted in India by Eiango et al

found that 56% of parents believe that the NBS is expensive [20].

CONCLUSION

Parental awareness and utilization of NBS remain low in Puducherry. Implementing structured antenatal education and promoting NBS at the institutional level are vital to enhance acceptance and coverage of NBS services. Policy initiatives should incorporate NBS into India's national health programs to guarantee equitable and universal access.

RECOMMENDATIONS

1. Incorporate mandatory NBS education into prenatal care (ANC) visits to ensure that all expecting moms receive accurate information.
2. Utilize digital tools and community health professionals to raise awareness and provide accurate information about NBS.
3. Provide subsidies or financial assistance to make NBS affordable and accessible to low-income families. Enhance follow-up and referral processes to facilitate the timely management of newborns with positive screening results.

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CONFLICT OF INTREST

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