RESEARCH ARTICLE

Factors associated with awareness of literate mothers about newborn screening: A cross-sectional study from a low-middle-income country

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Abstract

Objective: To explore the awareness level of literate mothers regarding newborn screening programmes, and to evaluate the associated factors.

Method: The descriptive, cross-sectional study was conducted at the Section of Chemical Pathology, Department of Pathology and Laboratory Medicine, Aga Khan University, Karachi, from January to September 2021, and comprised mothers aged 18 years or more. Data was collected using a structured questionnaire about newborn screening, and the subjects were compared in terms of age, residential background, education and parity. Data was analysed using SPSS 23.

Results: Of the 1016 responses, 896(88.2%) were analysed. The mean age of the sample was 37.7±10.87 years. There were 470(52.4%) mothers aged 31-45 years, 859(95.87%) were from urban areas, 751(84%) had a graduate degree, 652(72.7%) were multiparous and had 824(91.9%) had healthy children. Overall, 386(43%) mothers had awareness of newborn screening programmes. The main factors associated with awareness were age, education, primiparity, having healthy children, and province of residence being Sindh and Punjab ($p<0.05$), while the urban-rural divided was not a significant factor ($p=0.737$). Cost of healthcare 417(46.5%) and lack of awareness among physicians 356(39.7%) were identified as the main challenges in establishing newborn screening services in the country.

Conclusion: The awareness among mothers about newborn screening programmes was generally low among the subjects studied.

Keywords: Newborn screening, Inborn errors of metabolism, Public health, Knowledge attitude and practice survey, Metabolomics. (JPMA 73: 1805; 2023) **DOI: https://doi.org/10.47391/JPMA.7004**

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Introduction

Newborn screening (NBS) is a population-based programme to identify salvageable disorders in asymptomatic newborns soon after birth. Examples include congenital disorders, like congenital hypothyroidism, congenital adrenal hyperplasia, and biotinidase deficiency, and inborn errors of metabolism, like phenylketonuria, Maple Sugar Urine Disease (MSUD) and galactosaemia, etc.[1] These disorders warrant prompt treatment by avoidance of fasting, special diets, cofactor and vitamin supplementation, which can lead to improved quality of life and expectancy.[1-3] However, if these disorders are not included in NBS and diagnosis is delayed, the risk of morbidity and mortality increases,[2] which not only burdens the primary family affected, but also contributes to the overall healthcare burden of a country. High consanguineous marriage rates in Pakistan (46-62%) put populations at severe risk of inheriting genetic diseases and raising infant mortality, and strict cultural and social taboos make discontinuation of the practice a big challenge.[4]

With resource limitations of a low- and middle-income country (LMIC) and a medical infrastructure crippled by the burden of infectious diseases, public health genomics, especially NBS, do not enjoy the spotlight in community health in Pakistan.[4] As such, local or national guidelines need to be established for the early identification by NBS and management of the patients so diagnosed.[5] The method of NBS involves a simple prick from the newborn's heel to collect dried blood spot samples which undergo biochemical analysis by tandem mass spectrometry.[3,6,7]

Implementing an NBS programme with all its essential components, like screening, follow-up, confirmatory testing, management, education and evaluation of the programme, is a colossal task. As with all other screening programmes, limited awareness and understanding of physicians, allied health professionals, parents, and the public is a known barrier to the initiation of the NBS programme, especially when it is not state-run or a national programme. Awareness and education about NBS increased awareness of the child's health status and reduced the harm of psychosocial impact, promoting prompt follow-ups and parental autonomy through informed consent.[8]

At present, Pakistan lacks a national NBS programme, but since 2018, two bills about NBS have been passed, namely, the Sindh Institute of Child Health and Neonatology Act,

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2018, applicable to public hospitals in Sindh, and the Federal Newborn Screening Act of 2019, for the Islamabad Capital Territory (ICT).[9,10] The former bill makes no mention of which diseases will be screened for in Sindh, but the latter aims at screening technology for cystic fibrosis, congenital adrenal hyperplasia, biotinidase deficiency and galactosemia.[10] As of 2021, the National Institute for Child Health (NICH), in Sindh, has initiated an NBS programme for congenital hypothyroidism, and a few non-profit organisations and private hospitals independently screen for the diseases as well.[11,12] However, before developing a national NBS programme for Pakistan, it is important to know the current awareness level of the mothers regarding the subject, and to design and implement educational strategies to raise their awareness level. Without having basic knowledge of mothers' awareness about NBS, it would be difficult to get good NBS coverage, and even more difficult to strategise who, what, when and where to target for education.

The current study was planned to assess the awareness level of literate mothers regarding NBS programmes, and to evaluate the associated factors.

Subjects and Methods

The descriptive, cross-sectional study was conducted at the Section of Chemical Pathology, Department of Pathology and Laboratory Medicine, Aga Khan University, Karachi, from January to September 2021. After approval from the institutional ethics review committee, the sample size was calculated using OpenEpi[13] with 97% confidence interval (CI), 4% margin of error and a population size of 54,923,952 women aged >18 years.[14] The sample size was inflated by over 20% to account for dropouts and exclusions.

The participants were enrolled from the community. Those included were Pakistani mothers aged 18 year or older. Those not currently residing in Pakistan were excluded. Data was collected without identifiers and anonymity was guaranteed at the time of taking informed consent. Incorrect or missing information and duplicate entries were excluded from analysis.

Data was collected using a structured survey form for which literature review was conducted with two primary purposes; first to clearly define the construct, and, second, to determine if measures of the construct or related constructs already existed.[15] As most surveys were conducted in countries where the NBS programme existed for decades, prior survey forms were not used. The survey form (Annexure) was developed on Google Forms for ease of access to a diverse group of mothers. The form was designed in English, but for mothers who could not understand the language, forms in Urdu were also made available. All team members reviewed and piloted the form to ensure that items were clear and in an easily

Annexure:
Questionnaire

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Age: .....

City of Residence: .....

Educational Level

Graduate or above: .....

Inter/A levels: .....

Matric/O levels or below: .....

How many children do you have?

☐ 1 ☐ 2 ☐ 3 ☐ 4 ☐ 5 or above

What is the status of your children's health?

☐ Healthy ☐ Carrier of congenital or hereditary disease

☐ Have a congenital or hereditary disease

☐ Dead because of congenital or hereditary disease

I knew that NS (newborn screening) is a simple test for discovery of genetic disorders

☐ Yes ☐ No ☐ Uncertain

Babies with serious illnesses may look healthy when they are born

☐ Agree ☐ Disagree ☐ Uncertain

Have you ever received information about newborn screening from your health care provider?

☐ Yes ☐ No ☐ Uncertain

Would you like to know more about newborn screening?

☐ Yes ☐ No ☐ Uncertain

What do you think is the biggest challenge for newborn baby screening in Pakistan?

☐ Cost of diagnostics and treatment ☐ Lack of availability of therapeutics

☐ Lack of knowledge among physicians

Did you know that newborn screening can be done using few drops of baby's blood from heel prick?

☐ Yes ☐ No ☐ Uncertain

Questionnaire (Urdu):

سوال نامہ

عمر .....

رہائش کا شہر .....

تعلیمی درجہ .....

گریجویٹ یا اس سے اوپر .....

انٹر/اے لیولز .....

میٹرک/او لیول کی سطح یا اس سے نیچے .....

آپ کے کتنے بچے ہیں؟

☐ 1 ☐ 2 ☐ 3 ☐ 4 ☐ 5 یا اس سے اوپر

آپ کے بچوں کی صحت کا کیا حال ہے؟

☐ صحت مند ☐ پیدائشی یا موروثی بیماری کا کیریئر

☐ پیدائشی یا موروثی بیماری ہو۔ ☐ پیدائشی یا موروثی بیماری کی وجہ سے مردہ

میں جانتا تھا کہ NS (نوزائیدہ اسکریننگ) جینیاتی عوارض کی

کی دریافت کے لیے ایک سادہ ٹیسٹ ہے۔

☐ جی ہاں ☐ نہیں ☐ غیر یقینی

سنگین بیماریوں میں مبتلا بچے پیدا ہونے پر صحت مند نظر آتے ہیں۔

☐ متفق ہوں۔ ☐ متفق نہیں ☐ غیر یقینی

کیا آپ نے کبھی اپنے میٹرنٹی کیئر فراہم کنندہ سے نوزائیدہ بچوں کی اسکریننگ کے بارے میں معلومات حاصل کی ہیں؟

☐ جی ہاں ☐ نہیں ☐ غیر یقینی

کیا آپ نوزائیدہ بچوں کی اسکریننگ کے بارے میں مزید جاننا چاہیں گے؟

☐ جی ہاں ☐ نہیں ☐ غیر یقینی

آپ کے خیال میں پاکستان میں نوزائیدہ بچوں کی اسکریننگ کے لیے سب سے بڑا چیلنج کیا ہے؟

☐ تشخیص اور علاج کی لاگت ☐ علاج کی دستیابی کی کمی

☐ ڈاکٹروں کے درمیان علم کی کمی

کیا آپ جانتے ہیں کہ ایڑی کی چبھن سے بچے کے خون کے چند قطروں کے ذریعے نوزائیدہ کی اسکریننگ کی جا سکتی ہے؟

☐ جی ہاں ☐ نہیں

understandable language.

The survey consisted of two sections, comprising a total of 11 items. The first section collected the demographic details of the respondents, while the second section explored the level of NBS awareness. Once the survey design was complete, the form was shared on multiple social media platforms, like Facebook, Twitter and WhatsApp groups targeting Pakistani mothers.

The age of mothers was categorised into three groups (18-30 years, 30-45 years and >45 years). The residential background of the survey participants was broken down into Sindh, Punjab, Balochistan, Khyber Pakhtunkhwa (KP) and ICT groups, as well as urban and rural subgroups. Urban and rural areas were defined, according to population density and structured development.[16] Educational qualification was categorised into three groups (graduate or above, intermediate/A-level, and matriculation or below), while parity was categorised into four groups (1 child, 2 children, 3 children, ≥4 children). The health status of the child(ren) was categorised into four healthy, diseased, carrier and expired groups.

Data were analysed using SPSS 23. Data was expressed as mean and standard deviation (SD) or as frequencies and percentages, as appropriate. Chi-square test was used to determine the association of NBS awareness with education level, province of residence, parity of mothers, and current health status of the children. The level of statistical significance was set at $p<0.05$.

Results

Of the 1016 responses, 896(88.2%) were analysed (Figure 1). The mean age of the sample was 37.7±10.87 years. There were 470(52.4%) mothers aged 31-45 years, 859(95.87%) were from urban areas, 751(84%) had a

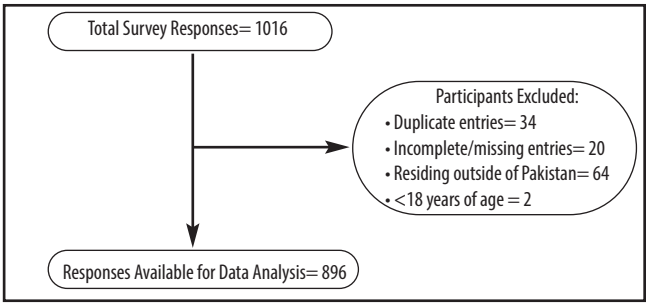

Figure-1: Study flowchart.

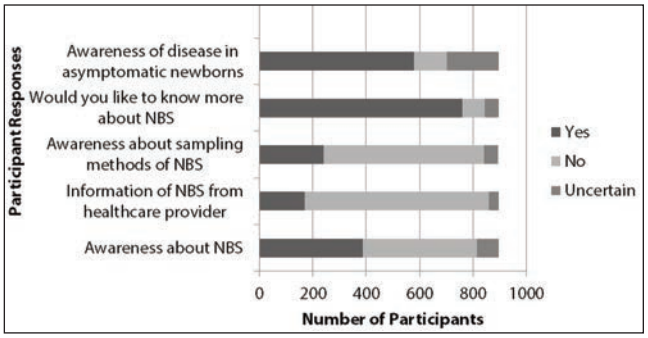

Figure-2: Responses about awareness regarding newborn screening (NBS) (n = 896).

Table: Demographic parameters associated with awareness of newborn screening (NBS).

Demographic parameter	Categories	% (n)	Awareness of NBS [% (n)]			*p*-value
			Yes	No	Uncertain	
			43 (386)	48 (428)	9 (82)	-
Age (years)	18-30 years	28.56 (256)	50.4 (129)	41.0 (105)	8.6 (22)	0.017
	31-45 years	52.45 (470)	43.6 (205)	47.6 (224)	8.7 (41)	
	>45 years	18.97 (170)	30.6 (52)	58.2 (99)	11.1 (19)	
Education of mothers	Graduate degree or more	83.81 (751)	94.5 (365)	35.6 (312)	12.4 (74)	<0.001
	Intermediate	8.9 (80)	18.7 (15)	77.5 (62)	3.7 (3)	
	Secondary School certification or below	7.2 (65)	9.2 (6)	83.0 (54)	7.6 (5)	
Type of Area	Urban	95.87 (859)	43.3 (372)	47.5 (408)	9.2 (79)	0.737
	Rural	4.12 (37)	37.8 (14)	54 (20)	8.1 (3)	
Province	Sindh	66.6 (597)	46.7 (279)	43.0 (257)	10.2 (61)	0.001
	Punjab	15.0 (134)	44 (59)	48.5 (65)	7.4 (10)	
	Balochistan	9.9 (89)	32.6 (29)	63 (56)	4.4 (4)	
	Khyber Pakhtunkhwa (KP)	3.7 (33)	27.3 (9)	63.6 (21)	9.1 (3)	
	Capital Territory	4.8 (43)	23.2 (10)	67.4 (29)	9.3 (4)	
Parity of Mother	1 child	27.2 (244)	55.7 (136)	32.8 (80)	11.5 (28)	<0.001
	2 Children	37 (332)	43.8 (144)	48.5 (161)	8.1 (27)	
	3 Children	22.9 (205)	40.5 (83)	52.2 (107)	7.3 (15)	
	≥ 4 children	12.83 (115)	20 (23)	69.5 (80)	10.4 (12)	
Current status of Child's Health	Healthy	91.96 (824)	45.1 (372)	45.6 (376)	9.2 (76)	0.001
	Diseased	3.9 (35)	22.8 (8)	68.6 (24)	8.6 (3)	
	Carrier	1.67 (15)	26.7 (4)	60 (9)	13.3 (2)	
	Expired	2.45 (22)	9.1 (2)	86.7 (19)	4.5 (1)	

graduate degree, 652(72.7%) were multiparous and had 824(91.9%) had healthy children.

Overall, 386(43%) mothers had NBS awareness (Figure 2). The main factors associated with awareness were age, education, primiparity, having healthy children, and province of residence being Sindh and Punjab ($p<0.05$), while the urban-rural divide was not a significant factor ($p=0.737$) (Table).

Cost of healthcare 417(46.5%), lack of awareness among physicians 356(39.7%) and limited availability of therapeutics 123(13.7%) were identified as the main challenges in establishing BS services in the country.

Discussion

Since the initiation of the NBS programme in the 1960s, more than 150 million infants have been screened for congenital diseases in the United States alone. Data from worldwide NBS programmes shows that awareness of parents, advocacy of NBS programme by parents, and their active engagement in the NBS is key to its success.[17] Considering the absence of a nationwide NBS programme in Pakistan, knowledge of the mothers' current awareness regarding NBS would facilitate leaders and healthcare professionals to strategize the design of future NBS programmes in the country, simultaneously maturing the programmes currently in their infancy.

As hypothesised, the current study observed low awareness of NBS in a significant majority of the diverse population of mothers surveyed. The findings match the results of a regional study conducted on the awareness of pregnant mothers about congenital hypothyroidism (CH). At baseline, the mothers had low awareness of CH and its implications (20%) which significantly increased after intervention (98%).[18] Other studies have stressed that when parents are unaware of the implications of congenital diseases, pre-symptomatic identification becomes an arduous process and definitive diagnosis and intervention can be dangerously delayed.[1,16]

The major factors associated with the low NBS awareness in the current study were mothers' age, education, parity, and health status of the child. In the study population, younger mothers showed higher awareness of NBS ($p=0.002$), which was consistent with the hypothesis that mothers of young ages may be more receptive to novel concepts in healthcare and make more educated decisions about their child(ren)'s health. This is supported by a study in which young population of mothers, despite having low awareness of NBS, willingly supported the NBS programme and were eager to have better awareness.[19]

Education takes a secondary role as even though most of the current sample were well educated, they exhibited split NBS awareness. As expected, participants with an intermediate and secondary level of education displayed a serious lack of awareness of NBS ($p=0.0001$). This finding is consistent with a study comprising Filipino mothers with diverse educational background and found higher knowledge of NBS amongst mothers with a university degree.[20] In the present study, a significant association between the place of residence and awareness of NBS was observed ($p=0.001$). Awareness about NBS was lower in mothers residing in KP and Balochistan provinces, and in ICT. Moreover, on categorising the population into urban and rural groups, data recorded from the rural areas in Balochistan and KP showed lower literacy rates[4] and lower levels of awareness, whereas urban areas in the Sindh and Punjab were relatively more aware of NBS, suggesting that a nationwide approach to NBS awareness is necessary. In ICT, despite 95% of mothers holding a graduate level of education, there was a generalised low awareness about NBS, and there is a need to explore the reasons for such lack of awareness.

Primiparous mothers displayed higher awareness of NBS than multiparous mothers ($p=0.0001$). This points to the possibility that with a single child, primiparous mothers may experience more free time to devote to their child's health, while their multiparous counterparts experience higher levels of exhaustion and compromised sleep, which is a finding corroborated by a previous study[21] A European study found that older parents, specifically those with 4 or more children, suffered worse physical, psychological and cognitive health, which could impede their child-rearing practices.[22] In LMICs with higher average birth rates, this finding may be a significant factor affecting the awareness of mothers. Additionally, the experiences of rearing multiple children may promote a higher sense of self-assurance in multiparous mothers, leading them to dismiss the utility of emerging health practices and services. This observation is supported in the literature, with multiple studies reporting that primiparous mothers are more willing to access health services and keep updated with their neonatal screening and vaccinations.[23,24]

On relating mothers' awareness of NBS with their child(ren)'s health status, the current findings were significant ($p=0.001$). Contrary to initial assumptions, mothers with healthy child(ren) displayed a higher awareness of NBS than those who had a disease. The results of a study conducted on parents in the Czech Republic supported the finding, stating that the difference appeared due to concerns over the child's health and the medical examinations performed after childbirth.[8]

Overall, the results of the current study indicated that NBS

was a novel concept in the Pakistani public, with many challenges facing the programme's initiation. While most of the survey population considered the cost of diagnostics and healthcare to be the biggest barrier to the NBS programme's success, nearly 40% of the mothers opined that the physicians' lack of awareness was a barrier. The next rational step would be to conduct more studies regarding the awareness and attitudes of the relevant physicians about NBS. Results of a 2017 study on the attitudes of Pakistani obstetricians towards non-invasive prenatal testing (NIPT) showed an overwhelming majority (97%) in favour of prenatal genetic screening in public hospitals. However, there was also concern about the added responsibilities and social burdens facing mothers who opt for testing and termination of pregnancy.[25]

The previous finding could also be attributed to the fact that 77.2% of mothers who responded were not informed by their healthcare providers about the existence of NBS tests.[25] This discovery can help improve the provision of future awareness programmes, such as deciding what groups of physicians should be targeted to educate the mothers. The timing of information delivery should also be considered before designing any awareness strategy. A study found the prenatal period to be effective as it offers a bridge to have concerns clarified between the mother and the physician.[20]

The current study has its limitations, like the data about socioeconomic status and source of NBS awareness was not collected, which could lead to incomplete inferences while designing appropriate educational strategy targeting Pakistani mothers.

Further studies are needed, especially comprising mothers who availed any NBS facility in Pakistan. In-depth structured interviews of such mothers may help in identifying the challenges in realistic terms to address the knowledge gap on the subject.

Conclusion

NBS awareness was found to be poor amongst the Pakistani women studied. However, their acceptance for NBS may be high, and doctors attending to mothers during the prenatal period can best bridge this gap and raise NBS awareness.

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