Original Articles

Knowledge and perception on expanded newborn screening in Sri Lanka among pregnant women attending antenatal clinics in Teaching Hospital Mahamodara

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Abstract

Introduction: Unlike in developed countries, there is no established expanded newborn screening (eNBS) in resource-limited settings. In Sri Lanka, the Ministry of Health has adopted screening only for congenital hypothyroidism, critical congenital heart diseases and congenital deafness as a policy in all newborns. Hence, there is a requirement to assess the need for eNBS in Sri Lanka. This requires epidemiological studies of the inherited metabolic disorders (IMDs) before investment to ensure cost-effectiveness of the programme and reflections on acceptance by beneficiaries.

Objectives: To evaluate the pregnant women's knowledge, attitudes and willingness towards eNBS.

Method: A descriptive cross-sectional study was conducted at the Mahamodara Maternity Hospital antenatal clinics, Galle, Southern Province, Sri Lanka. A convenient sample was obtained from the antenatal clinics. A pre-tested interviewer-administered questionnaire was used to assess basic socio-demographic details, knowledge and perception of eNBS

Results: A total of 602 pregnant women took part in the study. The mean age was 29±5.7 years; 67% were in the 25-40-year age group; 37.2% were primigravida and 60% were in the third trimester. More than 50% of the study participants knew about the heel prick test done at birth. However, only 34.8% and 28% were aware of the screening for congenital hypothyroidism and congenital heart disease, respectively; 93.6% of mothers had not heard of the term IMD, and only seven were aware of the term eNBS. Nevertheless, more than 80% of participants were willing to screen their babies at birth for underlying IMDs, even if it is available on payment, as the current programme was free of charge to the family. Similarly, 96.2% were willing to screen their babies for incurable diseases.

Conclusions: Though most women were aware of the heel prick test done at birth, only a suboptimal proportion of them was aware of the disease/s checked using the test. In addition, there was poor knowledge about the IMDs and eNBS. However, there were positive attitudes towards eNBS.

(Key words: Knowledge, Attitudes, Expanded newborn screening, Women, Sri Lanka)

Introduction

Inherited Metabolic Disorders (IMDs) are rare inherited disorders with diverse clinical manifestations. Although some individuals become symptomatic soon after birth, a significant proportion might develop symptoms later in life[1]. However, a delay in diagnosis can lead to irreversible neurological damage and even fatality[2]. Early identification of some of these disorders is possible and helps improve overall survival. Up to date, about 1450 IMDs have been classified, and the incidence of each disorder is rare. However, collectively, IMDs constitute a significant cause of mortality and morbidity in childhood[3]. The estimated case fatality rate of IMDs is 33% or higher in low or middle-income countries accounting for 0.4% of child deaths globally[4]. The exact prevalence of IMDs in Sri Lanka is not known. However, according to the 2018 Annual Health Bulletin report, out of 2200 neonatal deaths, there were 14 deaths due to IMDs, and 100 remained undiagnosed[5]. Newborn screening helps identify inherited disorders at their asymptomatic stage and intervene before babies enter the symptomatic phase. In 1960, Dr Robert Guthrie developed a test to diagnose

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phenylketonuria using a dry blood spot and this laid the foundation for newborn screening (NBS)[6]. Subsequently, a number of disorders were added to the newborn screening programme which is named expanded newborn screening programme (eNBS). However, in developing countries, the programme is not well established, and in most countries, congenital hypothyroidism (CH) is the widely screened disease[7].

NBS for CH was started in Sri Lanka in 2008 regionally and since 2016 has covered the entire country[8]. In January 2017, Sri Lanka introduced pulse oximeter screening for all newborns and subsequently hearing assessment[9,10]. In addition, screening for congenital adrenal hyperplasia, glucose 6-phosphatase dehydrogenase deficiency and congenital deafness are conducted in babies born in certain private hospitals in Sri Lanka. With advancement of perinatal care, neonatal mortality has gradually declined in most developing countries. As a result, NBS has become a public health priority to improve neonatal mortality further[7]. Therefore, Sri Lanka should try to expand newborn screening to incorporate IMDs. This requires epidemiological studies of the IMDs before investment to ensure the cost-effectiveness of the programme and reflections on the acceptance by beneficiaries.

Objectives

To evaluate knowledge of the existing NBS and to assess the attitudes and willingness towards eNBS.

Method

A descriptive cross-sectional study was conducted at the antenatal clinics of the Mahamodara Maternity Hospital, Galle, Southern Province, Sri Lanka. A convenient sample was obtained from the clinics until the sample size was achieved. A pre-tested interviewer-administered questionnaire consisting of three components was used. In the first part of the questionnaire, basic socio-demographic details of mothers, including their educational level, were obtained. Knowledge of the existing NBS was obtained in the second part of the questionnaire, and the third section focused on the knowledge and perception of the eNBS.

Ethical issues: Approval for the study was obtained from the Ethical Review Committee of the Sri Lanka College of Paediatricians (SLCP/ERC/2022/21). Informed written consent was obtained from all the participants.

Statistical analysis: The data were analysed using the Statistical Package of Social Science (SPSS) version 20.0.

Results

A total of 602 pregnant women took part in the study. The mean age was 29±5.7 years; 403 (67%) were in the 25-40-year age group. Table 1 gives the basic socio-demographic characteristics.

Table1: ***Basic socio demographic characteristics***

Basic demographic detail		Number (%)
Age category	<25 years	176 (29.2)
	25-40 years	403 (66.9)
	>40 years	23 (03.8)
Parity	P1	225 (37.4)
	P2	202 (33.6)
	P3	124 (20.6)
	P4	36 (06.0)
	P5 or more	15 (02.5)
Number of living children	0	246 (40.9)
	1	214 (35.5)
	2	112 (18.6)
	3	23 (03.8)
	4 or more	07 (01.2)
Gestation	1st trimester	52 (08.6)
	2nd trimester	186 (30.9)
	3rd trimester	364 (60.5)
Ethnicity	Sinhalese	531 (88.2)
	Muslim	50 (08.3)
	Tamil	20 (03.3)
	Other	01 (0.2)
Woman's educational level	Grade 5	31 (05.1)
	Ordinary/Level (O/L)	324 (53.8)
	Advance Level (A/L)	190 (31.6)
	Graduate	56 (09.3)
	Post Graduate	01 (0.2)
Husband's educational level	Grade 5	32 (05.3)
	Ordinary/Level (O/L)	365 (60.6)
	Advance Level (A/L)	172 (28.6)
	Graduate	31 (05.1)
	Post Graduate	02 (0.3)

Just above 50% of the study participants were aware of the heel prick test done at birth. However, only 34.8% and 28.0% were knowledgeable about the screening for CH and congenital heart disease, respectively (Figure 1). More than half of the participants were aware of the usefulness of the test and the type of disorders identified through that. However, the majority were unaware of the optimum timing of the test, the time taken to obtain results and confirmatory testing. Table 2 shows the awareness of the existing NBS in Sri Lanka.

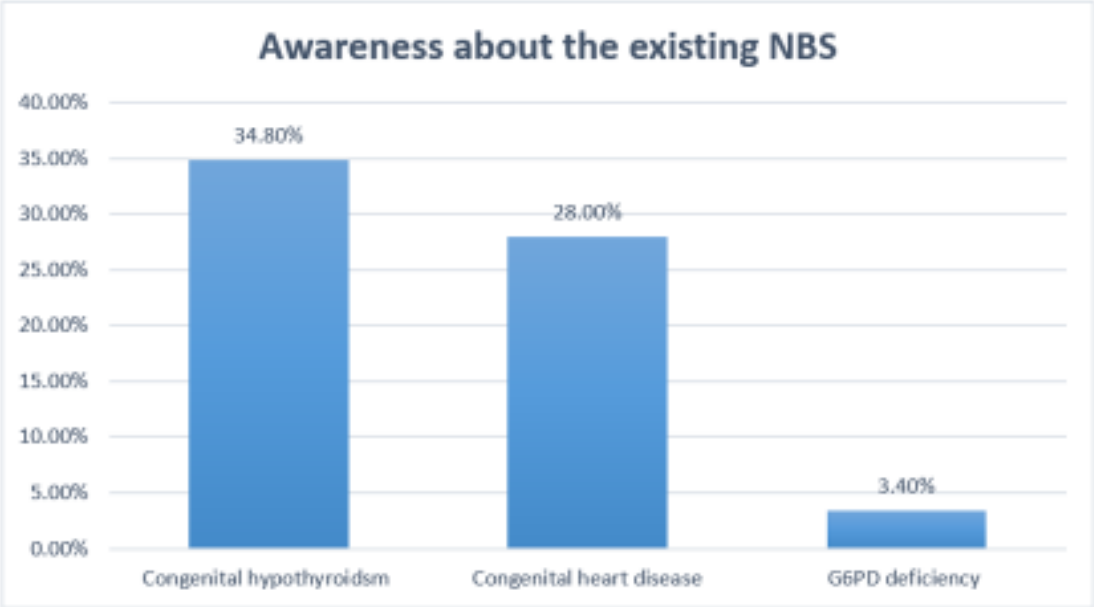

Figure 1: ***Awareness about the disease conditions screened at birth in Sri Lanka***

Table 2: ***Knowledge about the existing newborn heel prick testing in Sri Lanka***

Question	Response		
	Yes n (%)	No n (%)	Not aware n (%)
1. In Sri Lanka, heel prick blood test is done in all new-born babies	346 (57.4)	24 (03.9)	232 (38.5)
2. It is a simple procedure for the discovery of genetic and congenital diseases	325 (53.9)	13 (02.1)	264 (43.8)
3. The test helps an infant to avoid further deterioration of the child's health status	348 (57.8)	01 (0.16)	253 (42.0)
4. When diagnosed early, there is a chance of an excellent prognosis, and may be spared from lifelong impairment so that he/she can enjoy a normal life	346 (57.4)	02 (0.3)	254 (42.1)
5. The programme is operated by the Ministry of Health	251 (41.7)	07 (01.2)	344 (57.1)
6. The test can identify genetic and congenital diseases	316 (52.4)	13 (02.2)	273 (45.3)
7. The blood spot would help future research related to public health problems	261 (43.3)	19 (03.1)	322 (53.4)
8. The best time to do the test for a new-born is between three to seven days	118 (19.6)	152 (25.2)	332 (55.1)
9. The blood samples will be sent to the New-born Screening Laboratory and the result will be released in 7–14 working days	80 (13.2)	135 (22.4)	387 (64.2)
10. If the results of the heel-prick test are abnormal, it means that the new-born need to undergo another test to identify genetic or congenital disorder	288 (47.8)	15 (02.5)	289 (48.0)

Table 3 shows the knowledge and attitudes towards eNBS programme. Most (93.6%) of mothers have not heard of the term IMDs, and only seven were aware of the term eNBS. More than 80% of participants were willing to screen their babies at birth for underlying IMDs, even if it is available in

the private sector. Similarly, 579 (96.2%) were also willing to screen their babies for incurable diseases. Preventing delays in diagnosis and early preparation for a better lifestyle were the most commonly given reasons behind screening for fatal diseases. In contrast, unnecessary interventions and fear of anxiety were pointed out as leading factors against screening for incurable conditions at birth. However, most respondents had no moral obligations or religious restrictions against eNBS.

Figure 2 gives the reasons for screening incurable conditions at birth.

Figure 3 gives the reasons against screening for incurable conditions at birth.

Table 3: ***Knowledge and attitudes towards expanded newborn screening programme to identify inherited metabolic disorders***

Question	Response		
	Yes n (%)	No n (%)	Not sure n (%)
1. Have you heard about Inherited Metabolic Diseases (IMDs)?	37 (06.1)	565 (93.9)	-
2. Have you heard about expanded new-born screening to look for IMDs?	07 (01.2)	595 (98.8)	-
3. Are you willing to pay for expanded new-born screening if it is available in the private sector?	488 (81.1)	92 (15.2)	22 (03.7)
4. Babies with positive results are required for further investigations. Do you think it is still worthwhile screening?	587 (97.5)	15 (02.5)	-
5. Do you support screening of IEM, despite some of them being incurable?	579 (96.2)	23 (03.8)	-
6. The test results may not be 100% correct with possibilities of false positives and false negatives. Babies with positive results are required for further investigations. Do you think it is still worthwhile screening?	598 (99.3)	04 (0.7)	-
7. Screening babies at birth is morally justified	594 (98.6)	08 (01.4)	-
8. Screening babies at birth is against my religious belief	03 (0.5)	599 (99.5	-

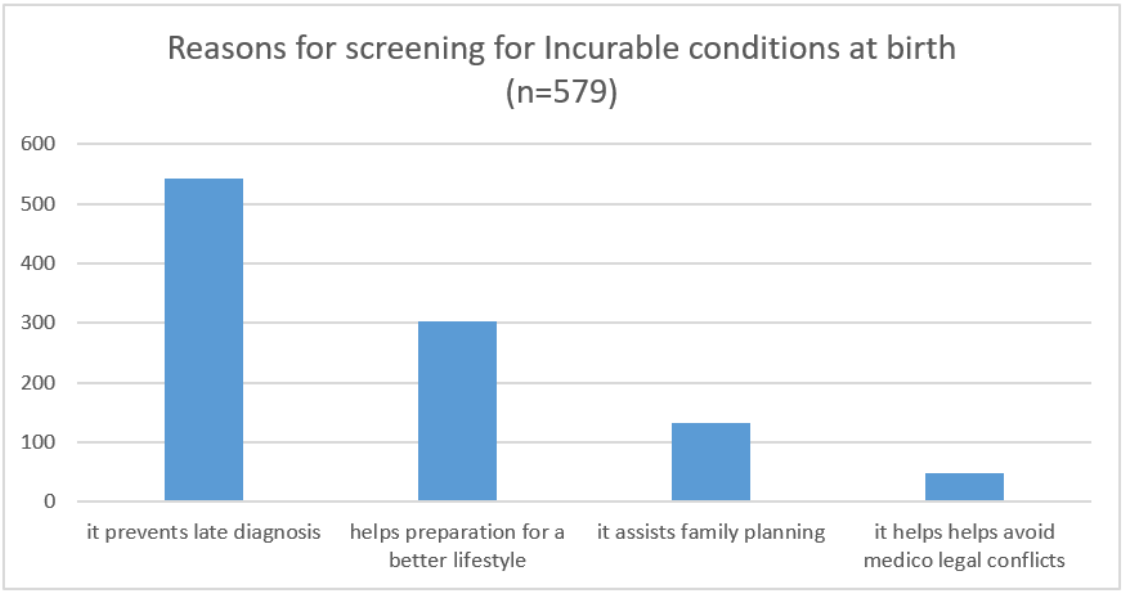

Figure 2: ***Reasons for screening incurable conditions at birth***

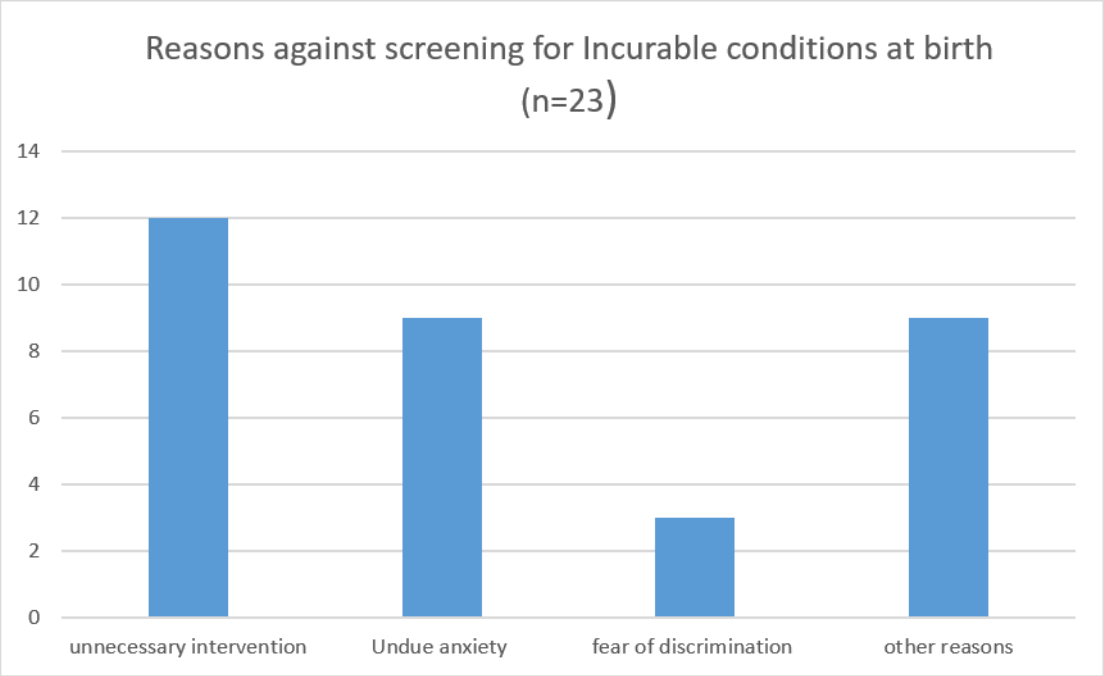

Figure 3: ***Reasons against screening incurable conditions at birth***

Discussion

This is the first study conducted in Sri Lanka to assess the knowledge of the existing NBS and parents' willingness towards eNBS. However, similar studies have been conducted in other countries for example, by Mak CM, *et al*[11] in Hong Kong in 2010 who showed that 83% of mothers were unaware of any NBS. Similarly, a study in Saudi Arabia in 2015 showed that only 22% of mothers were aware of the existing NBS[12]. In contrast, in the current study, more than half of the mothers knew about the heel prick test, but only 34% knew of the congenital hypothyroidism screening.

In our study, more than 93% of participants had not heard about the term IMD, and about 98% had not heard the term eNBS. Similarly, in the study done in Hong Kong, 87% of mothers were unaware of eNBS, and 80% of them ranked their awareness of IMDs as zero. Though there is a significant lack of awareness of IMDs and eNBS, most respondents were willing to screen their babies for many disorders at birth through eNBS. This finding was compatible with most of the studies done elsewhere. Al-Sulaiman A, *et al*[12] showed that 91% of women felt NBS was an essential and beneficial public health intervention but only 34% knew that NBS was done to check for a genetic disorder. Furthermore, a study by Kasem A, *et al*[13] in Jordan in 2020 revealed that most mothers had positive attitudes toward NBS but only moderate knowledge regarding NBS. Paquin RS, *et al*[14] in 2016 in USA have shown that there is a high parental intention to enrol children into eNBS programmes. In our study, 96% of mothers supported screening, even for incurable conditions at birth. Of them, 93% claimed that it prevents delay in the diagnosis, and more than half felt that it helps to prepare for a better lifestyle. Similar findings were described by Mak CM, *et al*[11] where more than 97% of participants supported screening for incurable conditions. According to our study, more than 99% of women felt that it was worth screening babies at birth despite some false positive results; a similar finding was observed in the study done in Hong Kong[11]. Only 3 participants felt that eNBS was against their religious beliefs, and eight thought it was not morally acceptable. These findings are on par with Kasem's study in Jordan[13].

Conclusions

Though most women were aware of the heel prick test done at birth, only a substandard proportion of them was aware of the disease/s checked using the test. In addition, there was poor knowledge about the IMDs and eNBS. However, there were positive attitudes towards eNBS.

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