

# Perception of Expanded Newborn Screening Programs in Saudi Arabia

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## ABSTRACT

The newborn screening procedures are known as public health programs as they help in diagnosing disorders that may not be identified before being manifested, clinically. The study aims to understand the significance of expanded newborn screening programs in Saudi Arabia. The study has conducted a cross-sectional survey by selecting 324 medical professionals involved in the neonatal screening program. The data was analyzed in the form of frequencies and percentages using SPSS version 20.0. The results have shown that 52.5% post-graduates agreed that NBS was an effective tool for better diagnosis; whereas 50% surprisingly mentioned that NBS is not an effective tool for diagnosis. Moreover, 85.8% post-graduate professionals agree that they can diagnose infancy impairments through NBS. Similarly, 30.4% nurses agree that they can diagnose infancy impairments through NBS. The findings have indicated that 45.7% post-graduate professionals agree that they can identify diagnostic problems related to surgeries. The study has concluded medical healthcare professionals are responsible for interacting with several newborn screening stakeholders, which include practices and healthcare providers, newborn screening treatment centers, families of infants screened, and prenatal educators assuring smooth execution of the newborn screening system.

## Introduction

The neonatal screening programs in Saudi Arabia are responsible for the screening and diagnosis of certain limited disorders to ensure long-term management [1]. A single test is allowed within the expanded newborn screening for early diagnosis and treatment of the concerned disorder, which may help in preventing serious health consequences during childhood. The newborn screening programs in Saudi Arabia are important as they provide coordinated and comprehensive system comprising of screening, diagnosis, treatment, management, and follow-ups, especially when considering the tribal culture and consanguinity rate of the Saudi population. The main aim of this strategy is to bring about better outcomes for the newborns suffering from various disorders [2]. Efforts are needed by the professionals to overcome significant problems, despite of the widespread acceptance of newborn screenings. There is a lack of comprehensive screening

programs to diagnose congenital diseases of newborn infants at national as well as regional level in Saudi Arabia. It is important to modify the comprehensive newborn screening programs based on the availability of the needs and resources. The screening programs are important to diagnose diseases such as the occurrence of abnormal hormones, congenital hypothyroidism, dehydrogenate deficiency, abnormal hemoglobin, incompatible blood groups, or the deficiency of glucose-6-phosphate dehydrogenase (G6PD) [3]. Various clinical symptoms are exhibited by inborn errors of metabolism, which comprise of heterogeneous and complex group of mono-genetic disorders. This takes place in the form of genetic code, which results in decreased activity of an enzyme functioning in a single pathway of intermediary metabolism [3].

The major causes leading to morbidity and mortality within the clinical practice are important due to the association of several

consequences with the inborn errors of metabolism, especially in pediatrics. The adverse disease outcomes result due to delay in diagnosis and treatment of certain disorders, which may include mental retardation, neuropsychological dysfunction, and death. Screening programs among the newborns are important as they help in identifying the infants suffering from serious inherited disorders that may be metabolic in origin. Some of the disorders diagnosed through screening can be treated through drug and dietary interventions before they lead to significant morbidity and mortality [4]. Therefore, the newborn screening programs are presented as a major advancement in child health care [5]. The stratification of the screened disorders and the implementation of the National Newborn Screening Program (NBS) in Saudi Arabia was addressed by Alfadhel, et al. [6]. A retrospective study was conducted from August 2005 to December 2012. The screening of 775 000 newborns was conducted in 139 hospitals of all regions of Saudi Arabia. However, a total 743 cases were evaluated in which information was provided regarding the incidents happened. The frequent disorders identified were: Congenital hypothyroidism, congenital adrenal hyperplasia whereas propionic academia was identified as the highest incidence among the Inborn Errors of Metabolism (IEM).

Al Hosani et al. [7] evaluated the data collected from January 1995 to December 2011 regarding the incidences of screened disorders and the molecular basis of positive screened cases. The study revealed the incidences of screened disorders which were: congenital hypothyroidism, amino acid, organic acid and fatty acid disorders, biotinidase deficiency, phenylketonuria, classical congenital adrenal hyperplasia, biotinidase deficiency, and sickle-cell traits and sickle-cell disease. The coverage of neonatal screening reached 95% in the year 2010. Al-Sulaiman et al. [5] assessed the attitude and knowledge toward newborn screening (NBS) program among the Saudi mothers. The study showed that majority of the women (91.1%) favored the NBS program and said that it is an essential tool for screening purpose. However, only 34.6% knew about the test and its nature. One of the primitive steps taken by Health authorities was to offer screening through communication counselling to the NBS patients. Guo et al. [8] examined the incidences of inborn errors of metabolism (IEMs) in different regions of the world. Tandem mass spectrometry (MS/MS) was used to expand the newborn screening for IEMs to diagnose early and to give the treatment to prevent the death ratio and permanent sequel. The study revealed 30 mutations in the nine genes associated with IEMs in the 28 authorized cases. Likewise, Hassan et al. [9] examined metabolic disorders by using the tandem mass spectrometry in Egypt. This test was conducted using newborn screening program at the Cairo University Children's Hospital in 2008. A total 13 disorders were found out such as phenylketonuria, methylmalonic academia, isovaleric academia, propionic academia,  $\beta$ -ketothiolase deficiency and primary carnitine deficiency. Results

revealed 235 cases diagnosed with the large number of diseases in Egypt. An early diagnosis, adequate management strategies and NBS program by using mass spectrometry were suggested to the nation.

Similarly, Bashir [10] studied the newborn screening to examine the newborn's medical condition that could lead to the early diagnosis to the treatment. Tandem mass spectrometry (MS/MS, DNA techniques, HPLC labelled bead technology, Guthrie test for Hyperphenylalaninemia (PKU), and IEF were used for the diagnosis. This test was started in Saudi Arabia to identify Congenital Hypothyroidism from 1991 and expanded to 2007 to add 23 more disorders that should be tested. Furthermore, in the period from 2005 to 2007, the Ministry of Health, King Faisal Specialist and Prince Salman Disability Research Centre examined 25% newborns while 50% newborns from 2007 to 2009 were tested in the country. Therefore, there was a need to examine the gaps, with the context of expanded Newborn Screening Programs in Saudi Arabia. Moreover, the knowledge level among the Saudi women towards NS program should be enhanced. Saudi Arabia has the most potential to increase its screening program for 16 diseases in 2005, but not in all of the regions of Saudi Arabia. There are three stakeholders guiding the development of Newborn Screening Programs in Saudi Arabia.

A recent study of Alfayez et al. [11] has demonstrated that increasing the awareness of screening programs require informing the parents about the screening benefits as well as its process based on the believe that adequate knowledge might improve the response for additional testing. Tu et al. [12] have supplemented that providing false positive screening results increase the parent's anxiety and stress for infants who are required to undergo more follow-up test, despite confirmation of the good health. Although, the recognition of pre-screening education of parents is considered as a crucial factor, little information about NBS shared with parents may still be oblivious to the screening of their baby. For instance, Abed [13] has revealed low awareness of mothers about NBS; however, their attitude was observed to be positive for NBS test. In Saudi Arabia, Al-Sulaiman et al. [5,14] have demonstrated the concerns of Saudi mothers concerning the improvement in the availability of the educational materials, and its awareness. Bakr et al. [15] have indicated that despite the execution of the mandatory program for Saudi national premarital screening (PMS), there exists a substantial need to escalate the awareness of the parents, particularly mother along with its screening time. The availability of the adequate medication, communication, genetic counselling, and awareness about the test and education materials is hindered among the Saudi women [11].

However, the newborn screening was done in the three major cities of Saudi Arabia but by only three institutes among which were from the same city. Thus, the country should generalize the

panel for newborn screening in the country. Therefore, the present study aims to understand the significance of expanded newborn screening programs in Saudi Arabia. It is well established that the newborn screening programs are helpful in reducing the overall costs and improve the outcomes. To the best of our knowledge, this study is the first to assess the significance of the expanded newborn screening program in Saudi Arabia and its impact.

## Material and Methods

### Research Design and Participants

A cross-sectional survey was conducted among 424 medical professionals from different private and government hospitals across different regions in Saudi Arabia. The respondents were asked to complete a structured questionnaire that sought their views upon the offered services. The study was approved by research advisory of Princess Nourah University in Riyadh, Saudi Arabia. The study confirmed to maintain confidentiality of all the information obtained from the respondents.

### Pilot Testing

Pilot testing was performed to understand the significance of expanded newborn screening programs in Saudi Arabia by recruiting 15 medical professionals from different private and government hospitals across different regions in Saudi Arabia. The reliability of the questionnaire considering 15 participants came out to be 0.78 (Table 1).

**Table 1:** Reliability Statistics.

Cronbach's Alpha	No. of Items
0.78	8

### Study Instrument

The questionnaire comprised appropriate questions that identifies the concerns regarding newborn screening programs for better diagnosis, infancy impairments, and potential diagnostic problems. The structured questionnaire comprised items to measure the attitude, beliefs, and knowledge about newborn screening programs. The questionnaire was administered to the corresponding coordinators associated with newborn screening programs around Saudi Arabia through email.

### Data Collection and Analysis

After obtaining information from the respondents, the questionnaires were coded and entered into Statistical Package of Social Sciences (SPSS) version 20.0. The data were analyzed in the form of frequencies and percentages. Moreover, inferential tests were also attempted to analyze the data for significance testing.

## Results

Both male and female healthcare professionals have responded purposively in this study, acknowledging that NBS is an important tool for the diagnosis at early phase. Table 2 has listed the demographics profile for the healthcare professionals selected for the study. Among these, 42.6% healthcare professionals were post-graduates while 24.7% were nurses. A total of 30.6% healthcare professionals were having an experience of 6-10 years while 48.1% healthcare professionals were having an experience of 0-5 years. Table 3 has presented the perceptions of healthcare professionals toward diagnosis conducted through NBS. From the findings, it is clear that 52.5% post-graduates agree that NBS is an effective tool for better diagnosis; whereas 50% surprisingly mentioned that NBS is not an effective tool for diagnosis. On the contrary, the views of medical professionals towards the diagnosis is not found significant at 5% level of significance. The perception of medical professionals toward infancy impairments diagnosed through NBS is presented in Table 4. The findings have shown that 85.8% post-graduate professionals agree that they can diagnose infancy impairments through NBS. Similarly, 30.4% nurses agree that they can diagnose infancy impairments through NBS that they can identify diagnostic problems related to surgeries. Moreover, 37% nurses agree that they can identify diagnostic problems by using NBS Table 5.

**Table 2:** Demographics.

		N	%
Gender	Male	162	50
	Female	162	50
Age	25-30	75	23.1
	31-35	153	47.2
	36-40	13	4
	41-45	69	21.3
	> 46	14	4.3
Designation	House officers	39	12
	Postgraduates	138	42.6
	Residents	23	7.1
	Nursing professionals	80	24.7
	Allied healthcare professionals	44	13.6
Years of experience	0-5 years	156	48.1
	6-10 years	99	30.6
	11-15 years	69	21.3
Marital status	Single	200	61.7
	Married	114	35.2
	Divorced	10	3.1

**Table 3:** Healthcare Professionals Perceptions towards Diagnosis.

Better Diagnosis							
		Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree	P-value
Designation	House Officers	4 9.80%	8 8.10%	17 14.70%	10 16.10%	0 0%	0.296
	Post Graduates	19 46.30%	52 52.50%	42 36.20%	23 37.10%	2 33.30%	
	Residents	1 2.40%	6 6.10%	8 6.90%	8 12.90%	0 0%	
	Nursing Professionals	10 24.40%	24 24.20%	28 24.10%	15 24.20%	3 50%	
	Allied Healthcare Professionals	7 17.10%	9 9.10%	21 18.10%	6 9.70%	1 16.70%	
Total		41 100%	99 100%	116 100%	62 100%	6 100%	

**Table 4:** Healthcare Professionals Perceptions towards Infancy Impairments.

Diagnose Infancy Impairments						
		Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
Designation	House Officers	6 13%	16 16.5%	10 8%	7 14%	0 0%
	Post Graduates	20 43.50%	41 42.3%	55 44%	18 36%	4 66.70%
	Residents	1 2.20%	7 7.20%	12 9.6%	3 6%	0 0%
	Nursing Professionals	14 30.40%	22 22.7%	31 24.8%	12 24%	1 16.70%
	Allied Healthcare Professionals	5 10.9%	11 11.3%	17 13.6%	10 20%	1 16.70%
Total		46 100%	97 100%	125 100%	50 100%	6 100%

**Table 5:** Healthcare Professionals Perceptions towards Diagnostic Problems.

Potential Diagnostic problems						
		Strongly Agree	Agree	Neutral	Disagree	Strongly Disagree
Designation	House Officers	8 19.5%	13 12.4%	13 10.7%	4 9.5%	1 7.1%
	Post Graduates	15 36.6%	48 45.7%	50 41%	20 47.6%	5 35.7%
	Residents	1 2.4%	5 4.8%	12 9.8%	2 4.8%	3 21.4%
	Nursing Professionals	15 36.6%	24 22.9%	30 24.6%	8 19%	3 21.4%
	Allied Healthcare Professionals	2 4.9%	15 14.3%	17 13.9%	8 19%	2 14.3%
Total		41 100%	105 100%	122 100%	42 100%	14 100%

## Discussion

The study has provided an in-depth information about the healthcare professionals' perceptions toward diagnosis using newborn screening programs. Post-graduates have found that NBS is an effective tool for diagnosis. Moreover, nursing professionals and allied healthcare professionals also found NBS as an effective tool for diagnosis. In this regard, the findings of Zuckerman are in-line with the findings of our study that NBS program is assessed as

an independent and stand-alone program for diagnostic problems. The use of NBS program has been well-established in improving the well-being of the baby and medical care of the newborn. In a similar context, Christoffel et al. [16] have supported the present study findings and stated that allied health professionals, nursing staff, and post-graduates assist and care each other during neonatal pain using NBS tool. Furthermore, Oerlemans et al. [17] have also found significant association of healthcare professionals with the newborn screening program during newborn screening. These



results have supported the findings of our study indicating that healthcare professionals positively perceive newborn screening issues using NBS. In addition, Osara et al. [18] have indicated that NBS program is used by healthcare professionals in improving patients care with inherited metabolic disorders.

Our study has also reported the positive perceptions of medical professionals toward infancy impairments. The findings have indicated that post-graduates, nursing professionals, and allied healthcare professionals were satisfied with the NBS program in identifying infancy impairments. These findings have been supported by Ojodu et al. [19] who found effectiveness of NBS program toward infancy impairments. Healthcare professionals have diagnosed infancy impairments during newborn care and well-being. Hayeems, et al. [20] have shown positive perceptions of healthcare professionals towards infancy impairments. Moreover, healthcare professionals also use NBS program for identifying and confirming infancy impairments. Sontag et al. [21] have found that healthcare professionals use NBS program for identifying and referring screen-positive newborns for follow-up and confirmatory diagnosis. These results also supported our findings which stated that healthcare professional's identify infancy impairments. Our study has found a significant and positive association for the perceptions of healthcare professionals towards better diagnosis of newborns. This finding has been supported by the findings of previous studies. For instance, Miller et al. [22] have indicated that positive perceptions of healthcare professionals toward better diagnosis of newborns using NBS program shows significant clinical benefit. Al-Zaidy et al. [23] have stated that the use of NBS program for screening of newborn impairments provide significant clinical benefits to healthcare professionals in identifying and halting the impairments.

The results of the present study would be beneficial for the medical practitioners and also great in term of benefit to the society. Screening for inherited metabolic disorders in Saudi Arabia is important to establish prevalence of these disorders and plan therapeutic interventions for the betterment of society. The assessment of overall success of these screening programs provide ongoing long-term disease management services for affected children and their families in an appropriate manner. Though our results have shown important and in-depth evidence related to newborn screening programs in Saudi Arabia by undertaking the perceptions of healthcare professionals, several limitations are also reported throughout the study. Firstly, the study has merely included a survey questionnaire, which shows the perceptions of healthcare professionals in the form of frequencies and percentages. However, qualitatively assessing such perceptions can give more benefit to this study in expanding and revising the program in Saudi Arabia. Secondly, the study has undertaken mostly medical professionals from King Faisal Specialist Hospital and Research Centre in Riyadh, Saudi Arabia, which shows that the validation of the results can be limited to the specific region. However, the study has included a

minimal extent of medical professionals from other centers too, but it was not legitimately focused.

Thirdly, due to lack of time and resources, the study has only collected data from Riyadh. Fourthly, the study has assessed the effectiveness of newborn screening programs toward the concerns experienced by medical professionals. However, specifying diseases and concerns can give additional opportunity to assess the effectiveness of newborn screening program. Lastly, parent's verdict was not involved, which can be beneficial for future studies to assess the effectiveness of the program.

## Implications

The findings of the study have suggested to heighten the public awareness about the NBS programs, which are found to be limited in Saudi Arabia. For expanding the NBS programs, it is essential to attain the public view which can provide new insights for the NBS expansion. Consequently, the study contributes as a platform for instigating the awareness campaigns for the professional societies to pose a strong and lasting influence on the professionals as well as the public. Also, the study suggests that these campaigns can lead to the incorporation of the newborn screening in the educational curriculum of the professionals and can also escalate the public acceptance toward the newborn screening. The educational campaigns are required because the population of the developing countries is generally reachable through various public media such as online platforms, radio, television, and newspapers. Therefore, it suggests the NBS advocates to avail the opportunity of participation on the radio, and television shows and also execute an online campaign within the country. Previous research conducted in Philippine has also endorsed the significance of extensive media campaign for influencing the NBS initial activities [24]. Moreover, the progression in the smart devices has also aided in improving the educational endeavors pertaining to the population while also assisting to identify the children who have been screened through newborn screening program. Since the screening of the newborn will be a national program; therefore, the initiated educational campaigns and programs must be consistent with the existing governmental system.

The support of the health ministry can play a vital role in initiating these NBS programs within the country government hospitals are supervised by the department. Thus, by using a similar supervisory infrastructure, a mechanism can be created which spreads newborn screening opportunities in the remote area as well. Also, the practitioners should discuss the issues of parental education, communicative outcomes, and the process of informed concerns at various forums.

## Conclusion

The study has aimed to understand the significance of expanded newborn screening programs in Saudi Arabia. The results depicted positive response from majority of the healthcare professionals

regarding the effectiveness of NBS for better diagnosis. There was a positive association between the expansion of newborn screening programs and its impact on newborn in Saudi Arabia. Therefore, it has been concluded that medical healthcare professionals in King Faisal Specialist Hospital and Research Centre are responsible for interacting with several newborn screening stakeholders, which include practices and healthcare providers, newborn screening treatment centers, families of infants screened, and prenatal educators that assure smooth execution of the newborn screening system. The study has suggested the policy makers to include an internal hospital NBS protocol for maintaining an inventory of NBS supplies, which include forms, educational materials, and cards for increasing parents' awareness. Secondly, medical professionals should record and enter the NBS card number on the electronic birth certificate to maintain tracking logs and screening results. Qualitative assessment of expanding newborn screening programs should be conducted for academicians and future studies to provide a more comprehensive understanding of the significance of the program. The future studies need to conduct comparative analysis in different cities of Saudi Arabia to assess the perceptions of both parents and medical professionals toward the NBS program.

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