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AWARENESS SURVEY ON NEWBORN, THALASSEMIA AND PREGNANCY SCREENING TESTING

Medical Science

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ABSTRACT

In developing countries, awareness about genetic conditions like Down syndrome, congenital defects, thalassemia, and inborn errors of metabolism is limited. We conducted a survey using a 19-question questionnaire to assess awareness of newborn, thalassemia, and pregnancy screening and whether these tests should be recommended or not among junior doctors, senior clinicians, MBBS students, and the general population. Results showed that only 60% of junior doctors knew about thalassemia screening. 50-70% of doctors and 60% of students supported invasive pregnancy testing. Approximately 35% of students were unaware of thalassemia carrier screening but 80% of them were open to routine testing. 90% of students recognized the importance of genetic screening awareness in current health scanario. About 40-60% of the general public lacked awareness about screening tests and the distinction between screening and diagnostic tests. Nearly 50% - 60% of people understood the importance of thalassemia screening. Although many clinicians and students acknowledged the benefits of screening tests, yet, increased awareness and attitude change, along with improved genetic education in medical curricula is needed.

KEYWORDS

Awareness, Newborn Screening, Thalassemia Screening, Pregnancy Screening.

INTRODUCTION

In India, there is a significant challenge posed by chromosomal disorders like Down syndrome, congenital birth defects, thalassemia, and inborn errors of metabolism (IEM), constituting a major burden of genetic disorders (I. C. Verma 2000). Down syndrome has an incidence of 1:1000 to 1:1100 according to the World Health Organization. Globally, 6% of children are born with serious birth defects annually (Global report on birth defect 2006), with 7% of under-five deaths in India attributed to congenital birth defects. Thalassemia affect 56,000 pregnancies worldwide (Antonio Cao,Y.W. Kan 2013; N.Madan et al. 2010), with an estimated prevalence of β-thalassemia carriers at 3-4%, and higher in certain ethnic groups. (Roshan B.Colah and A.C. Gorakshakar 2014; Roshan Colah, M.Mukherjee, K.Ghosh 2014.) Around 100,000 patients with β-thalassemia and about 150,000 cases of sickle cell disease in India are prevalent (A.A Durmaz et al 2014; I. C. Verma, R.Saxena, S.Kohli. 2011). Although the exact incidence of individual IEMs is not reported, collectively, it may approach 1 in 800 to 1 in 2500 births (A.A Durmaz et al 2014; I. C. Verma et al 2011) .The multidisciplinary treatment for these disorders imposes a significant financial burden on families, making prevention of affected births an idealistic goal.

Screening tests can identify genetic disease risk in asymptomatic individuals, and confirmatory tests confirm diseases in symptomatic cases, or assess the risk of passing on genetic conditions to the next generation. Early detection through screening reduces mortality and morbidity. Previous research highlights low awareness of these disorders and screening tests among people, the sub optimal level of knowledge and positive attitude of students, emphasizing the need for improved medical school curricula, additional physician education, and public awareness programs. (Azam Salehi et al 2019; C.L Clow and C.R Scriver reported 1977; Jia-Jia Chin and Hong-Wai Tham 2020; Rowe R, Puddicombe D, Hockley C, Redshaw M 2008; Susanne B. Haga, Esther Kim, Rachel A. Myers and Geoffrey S. Ginsburg 2019; Yau Shi Lin et al 2022.) . A survey was conducted to assess awareness of newborn, thalassemia, and pregnancy screening among junior doctors, senior clinicians, MBBS students, and the general population, aiming to understand their training, beliefs, readiness for adoption, and concerns, facilitating successful implementation of these tests in India.

Method

The survey took place at Lok Nayak Hospital (LNH) from May 1st to May 31st, 2023, with a total of 755 participants, of which 632 completed questionnaires. Participants were divided into three groups: Group 1 consisted of 200 (31.6%) junior doctors and senior clinicians, Group 2 included 253 (40%) MBBS students from Maulana Azad Medical College (MAMC), and Group 3 comprised 179 (28.3%) members of the general population, with 128 being highly educated and 51 having basic education.

A 19-question questionnaire was designed to assess awareness of newborn, thalassemia, and pregnancy screening, as well as basic knowledge about the reasons , importance of these tests . Participants were presented with four response options: yes, no, maybe, or not sure. The questionnaire was explained in Hindi or English to the general public for better understanding. Ethical committee approval was obtained, and written informed consent was obtained from all participants.

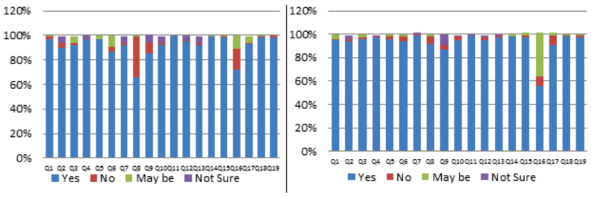
RESULTS

We analyzed 632 completed survey questionnaires, categorizing participants into three groups. Group 1 consisted of 200 junior doctors and senior clinicians (31.6%, M/F: 62/38), Group 2 included 253 MBBS students from Maulana Azad Medical College (40%, M/F: 52/48), and Group 3 comprised 179 individuals from the general population patients from LNH (28.3%, M/F: 26/74). Among the general population, 128 (71.5%) were highly educated, while 51 (28.5%) had basic education. The age distribution showed that 57% were below 25 years old, 28% were aged 25-50, and 15% were over 50 years old.

Group1: Comparing Junior Doctors (66) and Senior Clinicians (134) (Figure 1).

- In all 19 questions, over 90% of both junior doctors and senior clinicians answered "Yes."
- 60% of Junior doctors had awareness of thalassemia carrier screening.
- Over 70% of junior doctors and 50% of senior clinicians supported

invasive pregnancy testing.



Junior Doctors (66) Senior Clinicians (134)

Figure1:- Assessment Of Answers Between Junior Doctors And Senior Clinicians (200)

Group2: MBBS students' response (253). (Figure: 2)
Approximately 80-90% of MBBS students believed that newborn screening, thalassemia screening, and prenatal screening testing could effectively prevent these disorders early. They expressed willingness to routinely perform these screening tests (Questions 3, 6, 9, 13, 14, and 15) and considered them important in the current health scenario (Question 18).

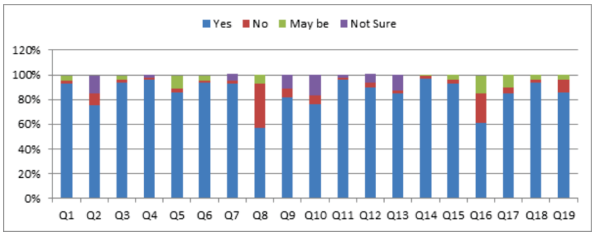
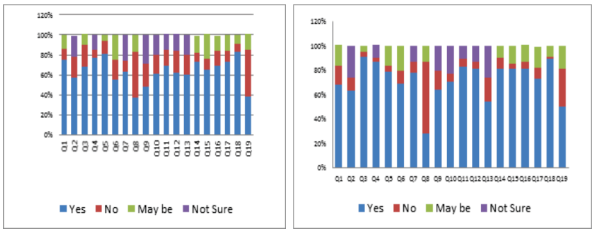


Figure 2: Assessment Of Answers Of MBBS Students. (253).

Group3: Group 3: General Population Responses (179). (Figure: 3)

- 60% of highly educated and 50% of less educated individuals were aware of newborn screening (Questions 2 and 6).
- Over 90% of highly educated individuals were willing to undergo newborn screening for their baby (Question 3).
- Approximately 60% of highly educated and 46% of less educated individuals were unaware of thalassemia carrier screening.
- About 50% of highly educated and 60% of less educated individuals recognized the importance of thalassemia screening during premarital, preconceptional, and prenatal periods (Question 13).
- 80% of highly educated individuals were open to invasive testing during pregnancy (Question 16).
- 50% of highly educated and 46% of less educated individuals did not distinguish between screening and diagnostic tests (Question 19).
- Both highly educated and less educated individuals believed that increased awareness about pregnancy screening is needed in the current health scenario (Question 18).



Uneducated And Basic Educated (51) Highly Educated (128)

Figure3: Assessment Of Answers Of General Population

Supplementary Material:
Questionnaire.
Assessment of common answers.
Assessment of uncommon answers
Assessment of answers between Students, Doctors and General population.

DISCUSSION

In the United States, newborn, thalassemia, and prenatal screening are integrated into healthcare services. However, in India, various factors, such as low education levels, sociocultural influences, limited access to reliable labs, cost concerns, maternal anxieties, and low awareness, have prevented these tests from becoming mandatory in primary healthcare. This survey assessed awareness levels about these screening tests among clinicians (Group 1), medical students (Group 2), and the general population (Group 3) in India.

Group 1

- Some practicing physicians (10%) had limited knowledge of screening tests, including their genetic basis and the role of consanguinity in these disorders. They were unaware that early screening and counseling could prevent these conditions.
- Some of the doctors even were not willing to have screening tests for their baby.
- Surprisingly, 2% of doctors were unsure, and 6% did not know about thalassemia screening.
- Regarding newborn screening, 2% lacked confidence, and 4% did not believe it could prevent or reduce disease severity.
- In terms of invasive pregnancy testing, 36% were uncertain, and 56% were in favor, indicating a lack of awareness about screening benefits and diagnostic test risks.
- Physicians would benefit from additional education to improve patient care and provide accurate information (Azam Salehi et al. 2019; Susanne B. Haga et al. 2019).

Group 2

- 14% of medical students were uncertain about newborn screening's ability to detect high-risk babies within 72 hours of birth.
- 1.7% didn't understand the role of genetics in metabolic disorders.
- 35% were unaware of thalassemia carrier screening.
- 17% were unsure about the connection between consanguinity and thalassemia, and 6.6% didn't know that thalassemia can be inherited from healthy carriers. 6.6% disagreed to do thalassemia screening for every pregnant lady.
- 60% agreed to perform invasive pregnancy testing, while 6.6% disagreed.
- 1% didn't realize that pregnancy screening can detect birth defects in early pregnancy, and 3% disagreed with noninvasive screening tests during pregnancy.
- 94% of students acknowledged the need to raise awareness about pregnancy screening in the current healthcare context.
- The data highlighted the students' lack of knowledge about screening tests, emphasizing the importance of comprehensive education covering all aspects of Wilson Jungner criteria (Anne E. Greb et al. 2009; Duaa Olwi, Leena Merdad, Eman Ramadan 2016).

Group 3

- Around 70% of individuals with basic education were willing to have newborn screening for their baby.
- Approximately 50% did not believe that newborn screening helps prevent or reduce the severity of diseases.
- 46% were unaware of thalassemia carrier screening.
- 46% had limited knowledge about screening and diagnostic tests during pregnancy, with roughly 70% willing to undergo invasive testing.
- Over 60% were open to noninvasive pregnancy testing, and 73% agreed that it should be mandatory for every pregnant woman.
- These findings suggest that with proper awareness and accurate information, more individuals would be willing to undergo these tests. Achieving awareness could be through one-on-one interactions, awareness campaigns, workshops, seminars, and mass media (Thomas Karagkiouzis et al. 2015).

CONCLUSION

- We need targeted educational programs to raise awareness and change attitudes about these tests among clinicians and medical students.
- Disseminating accurate and detailed information can combat unawareness and unfavorable attitudes, reducing guilt among people.
- This survey enriches medical education on preventive genetic diseases and screening tests, benefiting future healthcare.
- The impact extends beyond individual health, ultimately influencing the well-being of society and the nation.

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Conflict of Interest: None

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