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Original Research Article

Knowledge, attitude and acceptance of Down syndrome screening among pregnant women

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ABSTRACT

Background: Down syndrome (DS) is the most common chromosomal disorder, characterized by intellectual disability and various congenital anomalies. With increasing access to prenatal screening and diagnostic tools, understanding pregnant women's knowledge, attitudes, and acceptance (KAA) of DS is crucial for informed decision-making and supportive care.

Methods: A cross-sectional study was conducted among 260 pregnant women. Data were collected using a structured questionnaire assessing demographic information, knowledge of DS, attitudes toward children with DS, and acceptance of prenatal screening and possible outcomes. Statistical analysis was performed to determine correlations between sociodemographic factors and participants' responses.

Results: In a cross-sectional study of 260 pregnant women, 54.2% demonstrated adequate knowledge about DS and its screening, awareness varied, with only 44.2% understanding the difference between screening and diagnostic tests. A positive attitude toward screening was observed in 53.5%, and 63.8% showed acceptance, though fewer (56.9%) were willing to undergo invasive testing. Significant associations were found between higher knowledge and factors like primigravida status, higher education, and socioeconomic class. Similarly, satisfactory overall KAA (51.5%) was linked to these variables.

Conclusions: The study revealed that over half of pregnant women had satisfactory KAA regarding DS screening, particularly among primigravidae, graduates, and those from higher socioeconomic groups. While awareness of basic screening was good, gaps remained in diagnostic understanding and invasive testing acceptance, highlighting the need for targeted antenatal counseling to strengthen informed decision-making-fulfilling the study's objectives effectively.

Keywords: Down syndrome, Antenatal screening, Antenatal counselling

INTRODUCTION

Down syndrome (DS), or trisomy 21, is the most common chromosomal aneuploidy, with an estimated incidence of approximately 1 in 700 live births. The risk increases with advancing maternal age; however, it can occur across all populations irrespective of ethnicity or geographic location. It is associated with a wide spectrum of clinical manifestations, including congenital anomalies, intellectual disability, developmental delay, and characteristic facial features. These lifelong conditions impose considerable emotional, social, and financial burdens on affected families and healthcare systems. In the absence of definitive curative therapies, reduction in the burden of chromosomal abnormalities relies primarily on secondary prevention through prenatal screening and diagnosis.[1]

Prenatal screening for DS has evolved significantly over the past few decades, with improvements in detection rates and risk stratification. Current screening strategies combine ultrasonographic markers with maternal serum biochemical parameters to identify pregnancies at increased risk. Women categorized as high-risk are offered confirmatory diagnostic procedures such as chorionic villus sampling, amniocentesis, or fetal blood sampling. However, only a small proportion of these pregnancies are ultimately confirmed to have DS, leading to increased utilization of invasive procedures and an associated healthcare burden.[1]

The understanding of DS has advanced considerably with developments in cytogenetics. It is now well established that the condition results from the presence of an additional copy of chromosome 21 (human chromosome 21, HSA21). The term “DS” was formally adopted by the World Health Organization in 1965, replacing earlier inappropriate terminology.[2]

Evaluation of cell free DNA in the maternal blood is now the most common method of screening for DS. But in third world countries, it is expensive test. Hence other tests like biochemical markers and ultrasound estimation of nuchal translucency (NT) is an alternative procedure routinely offered to all pregnant women. The main idea of screening is to decreased the burden of the disease on the affected individual and family members. When prenatally diagnosed there is an opportunity for the parents whether to continue the pregnancy with affected baby or to terminate the pregnancy. Though lot of information can be obtained regarding screening, the mother may have lot of anxiety and fear to undergo the test and if screening test is positive, added fear to undergo the diagnostic test. There is also the risk of fetal loss during the procedure. There is also the possibility of false positive and false negative screening result. Counselling should be done to provide information which can be easily understood, explaining the risks and benefits.

At our centre, first-trimester combined screening is routinely offered between 11 and 13^{+6} weeks of gestation. This approach incorporates dual maternal serum biochemical markers-free β-human chorionic gonadotropin (β-hCG) and pregnancy-associated plasma protein-A (PAPP-A)-along with ultrasonographic measurement of NT to generate an individualized risk estimate.

This combined screening test demonstrates a detection rate of approximately 90% with a specificity of around 95% for DS.[3] It is offered universally to all pregnant women irrespective of maternal age and is performed for those who opt for screening after appropriate counselling.[4]

METHODS

A cross-sectional study was conducted in the Department of Obstetrics and Gynaecology, M. S. Ramaiah Medical College and Hospitals, Bangalore, from May 2023 to December 2024.

A total of 260 pregnant women with singleton pregnancies upto 13+6 weeks of gestation, irrespective of parity or age, were included after informed consent. Women with twin gestations or previous pregnancies with DS or other congenital anomalies were excluded. Data were collected using a pre-validated questionnaire assessing KAA regarding DS and its screening. Scores above the median indicated adequacy in KAA domains. No interventions were performed. Statistical analysis involved descriptive statistics and chi-square tests to examine associations between KAA scores and demographic variables.

Inclusion criteria

Early pregnancy (6 to 13+6 weeks), Singleton pregnancy, all primigravida and multigravida, All age group pregnant patients.

Exclusion criteria

Twin gestation, previous pregnancy resulting in DS previous pregnancy with any other congenital anomalies.

Sample size determination

The sample size was calculated using the standard formula: $n=Z^2\times p\times q/d^2$

Where, p is the prevalence of good knowledge regarding DS (21.1%), q=1- p, Z is the standard normal deviate at 95% confidence level (1.96), and d is the absolute precision (5%). Based on these parameters, the minimum required sample size was calculated to be 256. This was rounded off to 260 participants for the purpose of the study.

Statistical analysis

Data were entered into Microsoft excel and analyzed using SPSS version 26 (IBM SPSS Statistics, Somers, NY, USA). Descriptive statistics were expressed as mean±standard deviation for continuous variables and as frequencies and percentages for categorical variables.

Categorical data was analyzed using the Chi-square test. Fisher’s exact test and Yates correction were applied where appropriate, particularly for 2×2 tables when chi-square assumptions were not met.

Normality of continuous variables was assessed using the Kolmogorov-Smirnov and Shapiro-Wilk tests. For normally distributed data, the independent t-test was used to compare means between groups. For non-normally distributed data, the Mann-Whitney U test was applied to compare median values.

Graphical representations such as bar charts, pie charts, line diagrams, and scatter plots were generated using Microsoft excel. A $p<0.05$ was considered statistically significant.

RESULTS

Among the 260 women assessed, 48.8% were primigravidae and 51.2% were multigravidae, indicating a near-equal distribution and adequate representation of both first-time and experienced mothers, whose prior pregnancy experience may influence screening-related perspectives. The participants ranged in age from 18 to 43 years, with a mean of 27.22±4.53 years and a median of 27 years; the majority (43.8%) were aged 26-30 years, followed by 28.8% in the 21-25 years group, reflecting a predominance of women in their prime reproductive years. Nearly half (47.7%) were graduates, while 35.8% had pre-university education and 16.5% had only school-level education, indicating a relatively well-educated cohort with a good baseline capacity to understand health information. In terms of socioeconomic status (SES), most participants belonged to the upper middle (48.1%) and lower middle (41.9%) classes, with only 8.8% from the upper class and 1.2% from the lower class, suggesting that the majority had reasonable access to healthcare services, including antenatal care and screening programs.

Out of 260 respondents, 66.9% were aware of genetic diseases affecting babies and 65.8% knew that ultrasound can detect fetal malformations, while 60.4% recognized that combining blood tests with ultrasound improves DS detection. However, knowledge of specific aspects was lower, with only 40.4% aware of DS tests and 44.2% understanding the difference between screening and diagnostic tests, while about half had awareness of DS, its consequences, and timing of tests. Overall, 54.2% of participants demonstrated adequate knowledge, whereas 45.8% had inadequate knowledge, with a mean score of 18.82±6.69 and a median of 20. These findings indicate moderate overall awareness with good general knowledge but persistent gaps in DS-specific understanding, highlighting the need for improved targeted education.

A majority of participants (85%) were comfortable undergoing screening tests, and 74.2% supported prenatal testing for DS. However, in the event of a positive diagnostic result, only 26.5% chose to continue the pregnancy, while 43.8% opted for termination. Additionally, 29.6% remained uncertain. These findings indicate high acceptance of prenatal testing but varied decision-making regarding pregnancy outcomes, likely influenced by personal, cultural, or religious factor.

Among the participants, 53.5% demonstrated a positive attitude toward DS screening, while 46.5% had a negative attitude. The mean attitude score was 7.95±1.37, with a median of 9 and a narrow score range (3-9), indicating clustering toward higher values. Overall, this reflects a generally favorable attitude, though a substantial proportion with negative views may affect universal screening uptake.

Among the participants, 63.8% demonstrated acceptance of screening, while 36.2% did not. The mean acceptance score was 4.75±1.43, with a median of 5 out of 6, indicating generally favorable acceptance. However, substantial proportion of non-acceptance highlights persistent barriers such as limited access, fear/inadequate counseling.

Among participants, 81.9% were willing to undergo screening tests, indicating good overall acceptance. Additionally, 56.9% were willing to proceed with invasive testing following a positive screening result, while 16.9% declined and notable proportion remained unsure/unaware. This suggests that although initial screening is widely accepted, hesitancy increases with invasive procedures, likely due to safety concerns/limited awareness.

Table 1: Demographic details of the study subjects, (n=260).

Subjects	N	Percentage
Parity		
Primi	127	48.8
Multi	133	51.2
Age (in years)		
18 to 20	16	6.2
21 to 25	75	28.8
26 to 30	114	43.8
31 to 35	44	16.9
>35	11	4.2
Education		
School	43	16.5
PU	93	35.8
Graduation	124	47.7
Socio economic status		
Upper class	23	8.8
Upper middle	125	48.1
Lower middle	109	41.9
Lower class	3	1.2

Table 2: Responses to the questions on assessment of knowledge of the study subjects, (n=260).

Subjects	Yes		No		To some extent	
	N	%	N	%	N	%
Do you know about genetic diseases affecting babies?	174	66.9	72	27.6	14	5.4
Are you aware of consequences of screening tests for genetic diseases?	120	46.2	107	41.2	33	12.7
Have you heard about DS in babies?	128	49.2	105	40.4	27	10.4
Do you know DS can present with physical and mental abnormalities?	128	49.2	119	45.8	13	5.0
Are you aware of implications and complications of having child with DS?	145	55.8	95	36.5	20	7.7
Have you heard of any test which can detect DS?	105	40.4	123	47.3	32	12.3
Do you know that a simple ultrasound can detect the possibility of malformation in the fetus?	171	65.8	54	20.8	35	13.5
Are you aware that any blood test with ultrasound will help in increasing the detection rate of DS?	157	60.4	82	31.5	21	8.1
Are you aware of time necessary to undergo screening test?	147	56.5	72	27.7	41	15.8
Do you know difference between screening and diagnostic tests for DS?	115	44.2	112	43.1	33	12.7

Table 3: Distribution of the study subjects based on their knowledge, (n=260).

Subjects	N	Percentage
Adequate	141	54.2
Inadequate	119	45.8

Table 4: Responses to the questions on assessment of attitude of the study subjects.

Subjects	Yes		No		Doubtful	
	N	%	N	%	N	%
Are you comfortable undergoing both the tests?	221	85.0	10	3.8	29	11.2
Do you support prenatal genetic testing for DS?	193	74.2	26	10.0	41	15.8

Table 5: Responses to the questions on assessment of attitude of the study subjects.

Subjects (N=260)	Continuation of pregnancy		Confused		Termination of pregnancy	
	N	Row%	N	Row%	N	Row%
If diagnostic test is positive which option will you choose?	69	26.5	77	29.6	114	43.8

Table 6: Distribution of the study subjects based on their attitude, (n=260).

Subjects	N	Percentage
Positive	139	53.5
Negative	121	46.5

Table 7: Distribution of the study subjects based on their acceptance, (n=260).

Subjects	N	Percentage
Yes	166	63.8
No	94	36.2

Table 8: Responses to the questions on assessment of acceptance among the study subjects, (n=260).

Subjects	Yes	No			Doubtful	
	N	%	N	%	N	%
Are you willing to undergo test if there is an opportunity?	213	81.9	25	9.6	22	8.5
Are you willing to undergo invasive testing if the blood test and ultrasound results are positive?	148	56.9	44	16.9	68	26.2

DISCUSSION

This cross-sectional study was conducted over a period of 18 months in the outpatient department of Obstetrics and Gynaecology at Ramaiah Medical College and Hospitals, Bengaluru. The study aimed to assess the KAA of DS screening among pregnant women. The study population comprised pregnant women 6 to 13+6 weeks of gestation with singleton pregnancies, including both primigravida and multigravida women of all age groups. Women with twin gestation, previous pregnancies affected by DS, or other congenital anomalies were excluded.

Eligible participants were enrolled consecutively upon providing informed consent. Data were collected using a pre-validated structured questionnaire that assessed KAA related to DS and prenatal screening. Responses were scored, with scores above the median indicating adequate knowledge, positive attitude, good practice. while scores below the median reflected inadequate levels. The data collection involved no clinical intervention. Statistical analysis, including chi-square tests, was used to identify associations between KAA scores and participant characteristics such as age, parity, education, and socio-economic status. This design allowed the researchers to evaluate prevailing awareness and behavioral trends regarding prenatal screening in an urban antenatal population during early pregnancy.

Age distribution

In the present study, the majority of participants (43.8%) were between 26-30 years, with a mean age of 27.22 years. This age pattern is consistent with the reproductive age group targeted in antenatal screening programs. Among previous studies, Rabiee et al reported a mean age of 27.7±5.1 years with most women in the 25-29 age group, closely matching the present findings.[5]

Parity

The present study revealed a nearly even distribution between primigravida (48.8%) and multigravida (51.2%) women, indicating a balanced representation of first-time and experienced mothers. Among other studies, Rabiee et al. reported a higher proportion of primigravida women (66.7%), suggesting a greater inclination among first-time mothers to engage in screening or attend prenatal education. Mulvey et al, Pop-Tudose et al and Smith et al did not report specific parity data, limiting direct comparison.[6]

Gestational age

The present study recorded a mean gestational age of 9.41 weeks. This timing is appropriate for assessing awareness and acceptance of early DS screening. Among previous studies, Rabiee et al explicitly mentioned recruiting women during the first trimester, aligning well with the present study. Salvi et al also targeted antenatal women during early pregnancy, although exact gestational age was not provided.[7]

Education level

The present study reported that 47.7% of the women were graduates and 35.8% had completed pre-university education, indicating a well-educated antenatal population. Among previous studies, Rabiee et al showed that 87.1% had a diploma or higher, with 33.8% possessing university-level education, indicating a similarly educated population. Salvi et al found 70% of their sample had education beyond high school, aligning well with the present study.[7]

Knowledge

The present study found that 54.2% of women had adequate knowledge about DS and its screening, with 66.9% aware of genetic diseases and 40.4% aware of available screening tests. However, only 44.2% understood the distinction between screening and diagnostic tests. Among previous studies, Rabiee et al reported 15.2% with good knowledge and 54.3% with moderate knowledge, while only 33.3% were aware that screening tests were not diagnostic. Mulvey et al noted that although 91% supported screening, only 1% were aware of miscarriage risk and only 34% knew DS was chromosomal.[8]

Acceptance

The present study revealed high screening acceptance, with 81.9% of women willing to undergo DS screening and 56.9% prepared to opt for invasive testing following a positive result. Rabiee et al reported a 64.8% acceptance rate, with the remaining expressing uncertainty or refusal, mainly due to fear and lack of information. Pop-Tudose et al found only 24% initially accepted screening, but 95% expressed willingness post-diagnosis. Dahl et al and Mulvey et al noted high acceptance rates of 91%, with decisions often made after being informed of miscarriage risks and benefits.[8] The present study reflects one of the highest active acceptance rates, suggesting that Indian women, when informed early, are more willing to engage with antenatal screening programs than their counterparts in lower-awareness or culturally hesitant settings.

Overall KAA

In the present study, overall KAA was satisfactory in 51.5% of participants, with mean total scores of 31.52/45 across the three domains. The highest performance was observed in the attitude domain, while knowledge and acceptance scores were slightly lower but still substantial. Rabiee et al presented a fragmented KAA profile, with 15.2% good knowledge, 51% positive attitude, and 64.8% acceptance. Smith et al identified passive acceptance and incomplete understanding despite routine participation.[9]

Socio-economic status

In the present study, 48.1% of the participants belonged to the upper-middle class, while 41.9% were in the lower-middle class, indicating a predominantly moderate to high SES cohort. Among previous studies, SES was not consistently quantified. Rabiee et al recruited participants from urban prenatal centers, suggesting a middle-class demographic similar to the present study. Pop-Tudose et al reported that 82.7% of respondents lived in urban areas, indicating relatively high access to healthcare, although direct SES classification was absent and Smith et al. recruited women through public hospitals in Australia, implying equal access to care regardless of SES due to universal health coverage. Belahcen et al however, reported that 86% of participants were unemployed and 42% lacked health insurance, suggesting a lower SES profile.[10]

Attitude

In the present study, 53.5% of participants had a positive attitude towards DS screening, with 85% expressing comfort undergoing both screening and diagnostic tests and 74.2% supporting prenatal testing. Rabiee et al reported a similar attitude profile, with 51% having a positive outlook toward screening. Belahcen et al observed high acceptance of screening, with 84% and 90% favoring first and second-trimester screening respectively, despite poor knowledge.[10]

Nambiar et al found that 64.7% of the woman had right attitude towards screening tests and 46.1% patients had undergone the test.[11] In a study done by Pruksanusak 77.6% had the positive attitude towards the DS screening.[12]

It is now possible to screen with high degree of accuracy for cell free DNA fragments.[13] Cell free DNA can be a non-invasive screening test. Non-invasive pre-natal testing of cell free DNA can be an intermediate step between screening and diagnostic test.[14]

Though woman were able to understand the concept of screening but failed to explain the intricacies of screening.[15]

The present study has certain limitations that should be considered while interpreting the findings. The sample size was relatively small, which may limit the statistical power and the ability to generalize the results to a broader population.

Additionally, the study was conducted over a short duration, which may not adequately capture variations in KAA over time or across different stages of pregnancy. These factors may restrict the external validity of the study and highlight the need for larger, multicentric studies with longer follow-up periods to obtain more comprehensive and generalizable results.

CONCLUSION

This study aimed to assess the KAA of DS screening among pregnant women and found moderately favorable outcomes. Over half of the participants demonstrated adequate knowledge (54.2%), with higher scores among primigravidae, graduates, and those from upper socioeconomic classes. While 85% were comfortable undergoing screening and 74.2% supported prenatal testing, decisions following a positive diagnosis varied, reflecting personal and cultural influences. Acceptance of screening was high (81.9%), though willingness declined for invasive testing (56.9%). Overall, 51.5% showed satisfactory KAA scores, significantly associated with education, parity, and SES. These findings justify the study's objective by highlighting that although general awareness and acceptance are appreciable, knowledge gaps persist, particularly regarding diagnostic testing and its implications. The results underscore the need for targeted educational initiatives, especially for multigravidae and less educated groups, to ensure informed decision-making and improve utilization of prenatal screening services for DS.

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