

Down syndrome and its screening: knowledge, attitude and practice among pregnant women in Patan Hospital

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ABSTRACT

Introduction: Down syndrome (DS) is the most common cause of intellectual disability among live born children which can be detected prenatally. This study aims to assess the knowledge attitude and practice of pregnant women about Down syndrome and its available prenatal diagnosis.

Method: A study was conducted in Department of Obstetrics and Gynecology of Patan Academy of Health Sciences in 2021. All pregnant ladies who attended the antenatal clinic received a self-administered validated questionnaire to determine their knowledge of Down syndrome, its screening test and willingness to do prenatal testing.

Result: Majority of the participants (88%) belonged to age group of 18 to 35 years. 59% were primigravidas. Almost 90% were Hindus and 55% had graduate level education. More than half of the participants had poor knowledge of DS and 60 % had no idea regarding the screening tests. However, more than half (56%) of them were willing to undergo screening for DS. One fifth of the women did not want to continue the pregnancy if baby had DS. Higher income is significantly associated with good knowledge in both domain (knowledge about DS, and knowledge about its screening). Illiterates are found to have better knowledge than educated group which seems a bit contradictory, but this might have been due to our smaller sample size.

Conclusion: There is a significant gap between women's knowledge and their attitudes and practice, which has to be addressed with local and national policies and protocols. Informed decision making should be the norm after empowering pregnant women with knowledge.

Keywords: Down syndrome, knowledge and attitude, maternal knowledge, prenatal diagnosis

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INTRODUCTION

Down syndrome (DS) is the most common cause of intellectual disability among live born children with an incidence of 1:700-800 livebirths.¹ It is a genetic condition caused by the presence of all or a part of an extra copy of chromosome 21. The 2011 national census² reported 1.9% and the national living standard survey³ 2010-2011 estimated 3.6% of the population living with this disability. However, both the studies do not clearly state the percentage of people with Down syndrome. This is observed even in countries where elective termination of pregnancy is legalized and is being increasingly practiced since the widespread use of antenatal diagnostic methods.⁴

DS is amenable to prenatal detection by combined ultrasound and serologic screening and confirmed by invasive genetic techniques. Since 2007, ACOG recommends screening for DS for all pregnant women regardless of age⁵ but due to resource limitation, prenatal diagnosis of DS in Nepal is offered only in selective private institutions and that too in selective high-risk pregnant women only.

Studies concerning knowledge and attitudes of DS screening tests in pregnant women are important because they directly influence the woman's decision to either accept or decline the tests. However, most of these studies have been carried out extensively in Western countries⁵⁻⁷ with only a few in Asia.⁸ Unfortunately there has been no study regarding knowledge and attitude of DS screening tests in the Nepalese medical literature. Therefore, we aimed to assess the knowledge and attitudes of pregnant Nepalese women towards Down syndrome screening for preliminary implementation and to encourage the use of screening tests in our hospital as well as to determine factors affecting both knowledge and attitudes. The study provides an evidence-based appraisal of the notions and misconceptions about DS in our community. Such data will help healthcare professionals provide important information to the parents as well as communicate targeted messages in public awareness campaigns on DS.

The aim of the present study is to assess the knowledge, attitude and practices of pregnant women attending our antenatal clinic regarding Down syndrome and its prenatal screening test.

METHOD

This cross-sectional study was conducted in Department of Obstetrics and Gynecology of Patan

Academy of Health Sciences in April 2022-June 2022. Before conducting the study, the research proposal was submitted to the Ethical Review Board of Patan Academy of Health Sciences and a formal approval was obtained. All pregnant ladies who attended the antenatal clinic and those who gave consent for the study were included. The participants received a self-administered validated questionnaire in Nepali/English⁹. The questionnaire included demographic details, evaluation of the knowledge of DS and its screening tests and assessment of attitude towards DS screening test and the acceptance of having a Down syndrome child. The questions are detailed in the result section. The basic demographic data was collected and the responses were kept anonymous. The sample size was calculated to be 191 using the appropriate formula for prevalence study.¹

Sample Size: 191

$$n = z^2 \times p \times q / d^2$$

$$z = 1.96$$

$$p = 0.58^1$$

$$q = 1 - p$$

$$d = 0.05$$

Statistical analysis

All the statistical analyses were performed using Statistical Package for Social Sciences (SPSS) version 22. Descriptive statistics were used to determine the frequencies of each answer. To assess the level of knowledge regarding screening of DS, knowledge of DS, we calculated a score based on the number of correct responses from each section. Adequate knowledge was assumed based on scoring 50% or higher correct responses for each category respectively. Thus, we calculated the frequency and percentage of people who have adequate knowledge and who have favorable attitude. Correlations among variables were analyzed using Chi-square and Fisher's exact test and the significance level was set at $p < 0.05$.

RESULT

Total of 244 pregnant women were administered the questionnaire after taking their informed consent. Any incompletely filled questionnaires were excluded at the initial stage of the study. Table 1 shows the descriptive characteristics of the participants. Most of the participants i.e., 88 % belonged to age group of 18 to 35 years. 58.6% were pregnant for the first time. Almost 90% were Hindus and 55% had graduate level education. Almost 60% of women belonged to family with income >Rs.15, 000 and 5.7% had family history of DS.

As per the analysis of knowledge of DS questionnaire shown in table 2, around 41.4% thought that DS is a genetic abnormality. More than half of pregnant women knew that DS will cause mental impairment and physical abnormalities. 47.1% of women had no idea of relationship between increasing incidence of DS with increasing age of mother. One fourth of the women had the wrong idea that having on baby with DS increased the risk of having another baby with DS and 22.5% of participants thought that DS could be treated.

On analyzing knowledge regarding screening of DS, shown in table 3, 46.3% women knew that it could be detected during prenatal period and 38.9% knew that screening tests could be done during early four months of pregnancy. However, only 9.8% of women knew that screening tests are not confirmatory and may need further tests to confirm the syndrome. Nearly one third of participants thought that it should be done in every pregnant woman and not only those with risk of having baby with DS. 27.9% of pregnant mother knew that screening tests only talk about chances of having the baby with DS and almost 60 % participants had no idea regarding the screening tests. 29.5 % of mother thought that biochemical test of blood was done while 44.3% thought that

methods like ultrasound imaging was used for screening of DS in antenatal period. Almost half of participants thought that blood tests of mother along with ultrasound imaging increased the chances of predicting those babies with DS. 17% of the participants knew about the false positives and 11% knew about the false negative of the screening test.

Table 4 shows the attitude of participants towards Down syndrome and their practices. Among the participant mother, 55.7% showed interest in doing screening for DS while one fifth of the women did not want to do the test and remaining participants were undecided regarding the screening tests for DS. Approximately half of the mothers wanted further invasive testing to confirm the diagnosis while 34% were undecided and remaining participants did not want invasive tests in the form of amniocentesis/CVS. 41.8% participant mothers would want to continue their pregnancy despite having baby with DS, 17.6% did not want to continue the pregnancy if baby had DS while 40.6 % were undecided about further decision to continue or terminate the pregnancy.

Regarding practice of DS screening tests 68% women in the study had never undergone any screening tests till now.

Table 1. Descriptive characteristics of the participants (n = 244)

Characteristics	Categories	Frequency (%)	Characteristics	Categories	Frequency (%)
Age group	18-35 years	214(87.7)	Education	Primary	28(11.5)
	> 35 years	30(12.3)		Secondary	73(29.9)
Parity	Primigravida	143(58.6)		Graduate and above	134(54.9)
	Multigravida	101(41.4)		Illiterate	9(3.7)
Religion	Hindu	220(90.2)	Family income (monthly)	≤ 15,000 rupees	97(39.8)
	Buddhist	11(4.5)		> 15,000 rupees	147(60.2)
	Christian	7(2.9)	History of Down syndrome	No	230(94.3)
	Islam	(2.5)		Yes	14(5.7)
		Total			244(100.0)

Table 2. Item wise scores on knowledge regarding Down syndrome

SN	Questions	True	False	Don't know	Total
1*	Down syndrome is a genetic abnormality	101 (41.4%)	30 (12.3%)	113 (46.3%)	244 (100%)
2*	Children with down syndrome have mental impairment	165 (67.6%)	18 (7.4%)	61 (25%)	244 (100%)
3*	Down syndrome babies have congenital physical abnormalities.	139 (57%)	14 (5.7%)	91 (37.3%)	244 (100%)
4*	The risk of having a Down syndrome baby increases with advancing age.	102 (41.8%)	27 (11.1%)	115 (47.1%)	244 (100%)
5*	Young women less than 35 can also have babies with Down syndrome.	87 (35.7%)	29 (11.9%)	128 (52.4%)	244 (100%)
6*	A previous baby with Down syndrome increases the chance of Down syndrome in this pregnancy	73 (29.9%)	63 (25.8%)	108 (44.3%)	244 (100%)
7*	Down syndrome is not treatable.	73 (29.9%)	55 (22.5%)	116 (47.5%)	244 (100%)

*True is the correct response.

Table 3. Item wise responses for Knowledge regarding screening of Down syndrome

SN	Questions	True.	False.	Don't know	Total
1*	Down syndrome can be screened during the prenatal period.	113 (46.3%)	21 (8.6%)	110 (45.1%)	244 (100%)
2*	It is done ideally in the first four months of pregnancy.	95 (38.9%)	16 (6.6%)	133 (54.5%)	244 (100%)
3#	Down syndrome screening tests confirm definitely that your fetus has Down syndrome or not.	100 (41%)	24 (9.8%)	120 (49.2%)	244 (100%)
4#	The screening tests should be done only in high-risk group.	51 (20.9%)	79 (32.4%)	114 (46.7%)	244 (100%)
5*	The Down Syndrome screening tests only tell you that your fetus has more or less chance of having Down Syndrome.	68 (27.9%)	30 (12.3%)	146 (59.8%)	244 (100%)
6*	The Down Syndrome screening is carried out by taking maternal blood for biochemical tests.	72 (29.5%)	16 (6.6%)	156 (63.9%)	244 (100%)
7*	Down syndrome can be screened for by a detailed ultrasonography.	108 (44.3%)	25 (10.2%)	111 (45.5%)	244 (100%)
8*	Maternal blood tests and detailed ultrasonography together can increase the detection rate of down syndrome screening.	119 (48.8%)	19 (7.8%)	106 (43.4%)	244 (100%)
9#	If Down syndrome screening tests are positive, it means the fetus has Down syndrome.	54 (22.1%)	43 (17.6%)	147 (60.2%)	244 (100%)
10#	If the Down syndrome screening tests are negative, it means the fetus doesn't have Down syndrome.	68 (27.9%)	27 (11.1%)	149 (61.1%)	244 (100%)

*True is correct response #False is the correct response

Table 4. Attitude of participants towards Down syndrome and their practices

	Questions	No	Yes	Don't know	Total
Attitude	Will you undergo the screening test to detect Down syndrome?	50 (20.5%)	136 (55.7%)	58 (23.8%)	244 (100%)
	If preliminary maternal test is positive for Down syndrome, will you undergo the diagnostic invasive tests - amniocentesis/CVS?	41 (16.8%)	120 (49.2%)	83 (34%)	244 (100%)
	If the result of the test comes positive, will you continue with your pregnancy?	102 (41.8%)	43 (17.6%)	99 (40.6%)	244 (100%)
Practice	Have you ever undergone any screening tests to detect Down syndrome?	166 (68%)	35 (14.3%)	43 (17.6%)	244 (100%)

Table 5. Categorization of Knowledge regarding Down syndrome and its screening

Characteristics		Frequency	Percentage (%)
Knowledge regarding Down syndrome	Good Knowledge	115	47.1
	Poor Knowledge	129	52.9
	Mean $\pm$ SD (min, max)	3.03 $\pm$ 2.18 (0,7)	
Knowledge regarding screening for Down syndrome	Good Knowledge	83	34.0
	Poor Knowledge	161	66.0
	Mean $\pm$ SD (min, max)	3.07 $\pm$ 2.53 (0,9)	
Total		244	100.0

Note: Good knowledge have been categorized based on scoring of 50% or higher, i.e. 3.5 or higher for knowledge regarding Down syndrome, and 5 or higher for screening knowledge

Only 14.3% had undergone DS screening test at any point of time in this or previous pregnancy.

On further analyzing the data in table 5, 52.9% of participants had poor knowledge of DS. Similarly, 66% of participant mothers had poor knowledge regarding screening tests for DS in our study population. Family income was positively correlated with having her income with good knowledge of DS and its screening tests and was statistically significant in this study. Illiterate group

are found to have better knowledge than educated group which might be due to small sample size (only 9 illiterate participants)

DISCUSSION

The cross-sectional study investigated pregnant women's knowledge about DS, its screening tests and their attitude and their practice regarding DS in a study population in Kathmandu. The study supports the existence of gaps in knowledge about DS, its screening tests, attitude regarding DS and

the practice of screening tests of DS. This might be due to many social misconceptions about people with DS and lack of knowledge regarding DS.

There is no cure for DS. Antenatal diagnosis allows for preparation for the birth and subsequent care of a baby with DS or for the offer of a termination of pregnancy.¹⁰ The focus of antenatal screening of DS has been on the first trimester of pregnancy, during which period ultrasound imaging is used to measure nuchal translucency along with absence or presence of nasal bone and a double biomarker test is used to measure the serum level of free β -HCG and pregnancy associated plasma protein A (PAPP-A) in the pregnant women. The tests are noninvasive and have high sensitivity for detecting pregnancies with a high risk of DS.¹¹ However, the definitive prenatal diagnosis relies on genetic tests, which require the use of more invasive methods, such as amniocentesis or chorionic villus sampling, which is associated with 1-2% risk of procedure related fetal loss.¹¹ In Nepal, there is no established national prenatal screening protocol for DS, further invasive genetic tests and management thereafter if parents want to have a child with DS. The basic knowledge and understanding of DS can help parents to adopt strategies to plan their pregnancy. It can help them decide whether to terminate the pregnancy or if they decide to continue, what kind of support a baby with DS requires in the future and its implications on them and their needs.

We can conclude that knowledge of both Down syndrome and its screening tests are poor in our study population. We have found that the participants' educational status however did not guarantee good knowledge and awareness about Down syndrome and the screening tests available. This finding could be attributed to the small sample size in our study. This result needs further research and exploration. However, our finding was consistent with the study done in Kasturba Medical College in 2018.¹ When considering women with family history of Down syndrome we found that though these women had good knowledge of Down syndrome, their knowledge regarding the screening tests were limited. This could be due to personal experience with a Down syndrome baby which increased their awareness to know more about this condition which in turn made their knowledge of DS comparatively better. The lack of knowledge regarding screening tests available might be because of lack of proper counseling or financial and logistic barriers.

There is a gap between women's knowledge and their attitudes and practice considering Down syndrome and their screening tests. This could be due to our practice of imposed decision making as opposed to informed decision. This gap can be minimized by providing adequate information to the parents regarding Down syndrome and the available screening and confirmatory tests. The pregnant women should be educated regarding the syndrome and the available tests so that she can make an informed decision. This may also help them to prepare for the eventuality of going through invasive confirmatory tests if their screening test comes positive and provide them with an option of either terminating or continuing the pregnancy if the diagnosis is confirmed.

CONCLUSION

Informed decision making must be practiced along with dispersion of effective education regarding Down syndrome and its screening tests at the earliest antenatal visit. More significantly, this gap between women's knowledge, attitude and practice needs to be addressed with local and national policies and protocols for it to be worthwhile.

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Conflict of Interest

None

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