Original Article :

Knowledge and attitude of Down syndrome and prenatal screening among women in Hail, Saudi Arabia.

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Abstract

Background and Aims:
Down syndrome is the most common chromosomal abnormality occurring in live born infants. In Saudi Arabia termination of pregnancy is considered illegal and both religiously and culturally unacceptable. Our aim was to explore views, attitudes, and knowledge about Down syndrome, prenatal screening, integrating Down syndrome individuals in the community, willingness to terminate pregnancy.

Methods:
This cross-sectional study was conducted using a pre-validated questionnaire and carried out through face-to-face interviews, in Hail, Saudi Arabia from July-August 2019.

Results:
A total of 337 married women participated in this study, about Half were college graduates 157\337(46.6%). Participants expressed opposition as "extremely opposed" to Down syndrome individuals to be involved in the community in schools, have jobs and attend public places 108\337 (32.0%), 140\337(41.5%) and 139\337(41.2%) respectively. Illiterate participants were more willing to continue a pregnancy with Down syndrome fetus (AOR 9.7,95%CI: 2.2 – 42.8, p=0.003). In comparison, participants with college education were more willing to terminate (OR 1.6 95% CI: 1.0- 2.4, P=0.038). Moreover, awareness about prenatal tests were of no significant association with the willingness to terminate.

Conclusions:
This study has shown lack of knowledge about Down syndrome and most Saudi women in Hail are willing to undergo NIPT to prepare mentally for the child, but not go as far as TOP. This study also highlights the opinions of women towards the integration of DS individuals in the community. Further research on these questions would be useful to explore the reasons for the stigma surrounding the inclusion of DS individuals.

Keywords:
Down Syndrome, Trisomy 21, DS Fetus, Complicated Pregnancy, DS Child, Willingness to Terminate, Termination of Pregnancy.

Introduction

Down syndrome (DS) is the most common chromosomal abnormality occurring in live born infants[1]. In Saudi Arabia, the prevalence of DS has been reported to be 6.6 per 10,000 live births[2]. Most individuals with DS have extra copy chromosome 21 resulting in trisomy, which occurs due to meiotic nondisjunction. The other causes are Robertsonian translocation and mosaicism[3]. The prevalence of DS increases with advancing maternal age, recognized at 35 years and progressively increasing each year [4].

DS is associated with a number of conditions including craniofacial abnormalities, intellectual disability, congenital heart defects which are the most common cause of death among patients with DS[5], neurologic dysfunction, eye disease, hearing loss, obstructive sleep apnea, otitis media, gastrointestinal atresia, hip dislocation, and thyroid disease[6].

Concerning life expectancy, most people thought that a person with Down syndrome would die younger compared to the general population. However, today the survival rate of people with DS has dramatically improved in developed countries, estimates of the life expectancy of someone with DS has increased to nearly 60 years of age during the last two generations depending on the severity of their health problems[7].

The Prenatally and Postnatally Diagnosed Conditions Awareness Act (2008) emphasis providing accurate and emphasis information about prenatal genetic conditions to pregnant women as well as expanding the opinions of people towards disabilities and hopefully changing the negative perspective and stigma. Its goal is to enable pregnant women to make independent decisions about their pregnancy that fits their personal, cultural, religious, and social backgrounds[8].

Non-invasive prenatal tests(NIPT) is a screening tool for DS[9].

In recent years, there has been growing interest in understanding the knowledge and attitudes towards Down syndrome and prenatal screening, as it plays a crucial role in promoting informed decision-making and improving healthcare outcomes. The use of NIPT is increasing and with-it different attitudes and views about its social influence on the acceptance of DS people and the likelihood of pregnant women to undergo termination if the test is positive[10]. In the Netherlands, many individuals were concerned that a rise in the uptake of NIPT will cause a decline in the population with DS as more women will choose to terminate, resulting in a loss of acceptance as well as a loss of governmental and insti-

tutional support for the affected individuals[11]. However, in Saudi Arabia termination of pregnancy is considered illegal and both religiously and culturally unacceptable except in specific circumstances as an example, if the mother's life was at risk and based on the severity and treatability of the fetus's condition in which the pregnancy can legally be terminated[12]. Interestingly enough, in Pakistan, an Islamic country such as Saudi Arabia, the majority of the parents with a child with DS were willing to advise others to undergo termination of pregnancy (TOP), mainly driven by the negative backlash of their extended families with the lack of national health service and physical health centers for DS[10]. Only limited data are available on how aware women are with DS in Saudi Arabia. Attention toward DS can spread more into the population for the sake of DS individuals to be able to blend into our normal lives in schools, jobs, and society without any prejudges against them. For this hope to be achieved, we need the general population to fully understand the different aspects of DS its etiology, risk factors, manifestations, and the expected life morbidity. Hence, the need to evaluate married women's knowledge about DS, its risk factors, complications, and screening methods is crucial. DS specialized support centers are limited in Saudi Arabia and only available in larger cities, families in rural areas have extreme difficulties and they must be referred to larger centers for treatment[13]. Based on all of that, this cross-sectional study was needed to explore women's views on many aspects including measuring their knowledge about prenatal screening and diagnostic tests which is essential for making an informed decision, this, in turn, will help evaluate the effectiveness of educational resources and health professionals in terms of providing accurate information about screening to pregnant women and helping them understand the results

Methods

This cross-sectional study was conducted after Institutional Review Board (IRB) approval using a validated questionnaire and carried out through face-to-face interviews by the 1st, 2nd and 3rd authors (medical students trained and supervised by the principal medical consultant). A total of 337 married Saudi women aged between 17-70 years were included. Women who had a previous child with DS were excluded. Participants were recruited at a community setting (e.g., like malls and out-patients clinics waiting rooms) from July 2019 to August 2019 in Hail, Saudi Arabia. The agreement to participate in the study was considered as proof of consent.

Demographic data of the participants were recorded and analyzed.

Content of the Survey Instrument

The questionnaire was translated to Arabic and carried out through face-to-face interviews, 21 questions were developed and organized in four sections; the first section contained sociodemographic data. The second section; women knowledge about DS, their knowledge about screening tests and their availability noting that participants were given no prior information about prenatal screening. The third section assess participant's impression and how they perceive DS individuals. Answers were weighted on a scale of 1 to 5 (extremely supportive, supportive, neutral, oppose, extremely oppose). The fourth section contained questions about their decision and attitude toward prenatal tests and TOP.

Sample size

A total of 337 participants were needed to estimate attitudes, and knowledge of married women about DS. The sample size estimated for this cross-sectional study used following formula =Z 1-ɑ/2 2 SD2 / d2, where Z1-α/2= is standard normal variate (5% type 1 error) and a confidence level of 95%. SD standard deviation of variable. d = absolute precision

Statistical Analysis

All Categorical variables such as age group, parity, marriages status, income and education etc. presented as numbers and percentages were analyzed by Chi-square and Fisher's exact according to whether the cell expected frequency is smaller than 5. Univariable and multivariable logistic regression models were used to obtain the crude and the adjusted odd ratios (OR) and their associated 95% confidence interval (CI), to determine the most significant risk factors to either continue or terminate pregnancy with down syndrome fetus. A P-value of less than 0.05 considered as statistically significant. All data entered and analyzed through statistical package SPSS 28 (SPSS Inc., Chicago, IL, USA).

Consistency and Validation of Questionnaire

The questionnaire was prepared based on the available information from literature reviews. The questionnaire was reviewed by a panel of experts and revised based on their comments. After that, a pilot study was conducted to evaluate the internal consistency and validity of the Arabic version of the questionnaire by asking 50 participants to complete the translated questionnaire. Cronbach's alpha was calculated, and it was within the acceptable level (α = 0.80, 0.78 and 0.85). Most items

appeared to be worthy of retention.

Ethical approval

This research has been reviewed and approved by the institutional review board at king Fahad medical city, IRB approval 19-366.

Results

A total of 337 married women participated in this study. Table 1 presents sociodemographic of the participants. Majority of the participants 163/337 (48.4%) were over 35 years of age. Most of the participants had three children or more 205\337(60.8%) (Table 1). About half of participants were college graduates 157\337(46.6%), and only 57\337(16.9%) had an income of more than 10,000 riyals a month (Table 1). Majority of participants 254\337(75.4%) were housewife unemployed and 282\337(83.7%) lives in the city (Table 1). Participants married for more than 20 years occurred in 89\337(27.3%) (Table 1).

Table1. Sociodemographic for studied participants (n = 337)

Variables	Description	N (%)
Age group (years)	<26	23 (6.8)
	26 – 30	84 (24.9)
	31 – 35	67 (19.9)
	> 35	163 (48.4)
Parity	no children	35 (10.4)
	one child	48 (14.2)
	two children	49 (14.5)
	≥ 3 children	205 (60.8)
Married duration (years)	0 – 5	70 (21.5)
	6 – 10	78 (23.9)
	11 – 15	53 (16.3)
	16 – 20	36 (11.0)
	> 20	89 (27.3)
Income Saudi Riyals	< 3000	151 (44.8)
	3100-5000	65 (19.3)
	5100-10000	64 (19.0)
	> 10000	57 (16.9)
Educational level	Illiterate	35 (10.4)
	Basic school	58 (17.2)
	High school	74 (22.0)
	College	157 (46.6)
	Post-grade	13 (3.9)
Employment status	Not employed	254 (75.4)
	Employed	83 (24.6)

Variables	Description	N (%)
Residency	City	282 (83.7)
	Rural	55 (16.3)

Table-2 shows participants expressing their support as "extremely supportive" to DS individuals to go to schools together with normal individuals, have jobs and attend public places 69\337(20.5%), 55\337(16.3%) and 77\337(22.8%) respectively. On the other hand, participants expressed more opposition as "extremely opposed" to DS individuals 108\337 (32.0%), 140\337(41.5%) and 139\337(41.2%) respectively.

Table 2. Attitudes, and knowledge about DS, prenatal screening, integrating DS individuals in the community, willingness for termination

Question	Answer	N (%)
Do you know what genetic mutation causes DS?	Yes, I do	40 (11.9)
	No, I do not	297 (88.1)
Can a child with DS be cured?	Yes, they can	32 (9.5)
	No, they can not	198 (58.8)
	I do not know	107 (31.8)
What is the life expectancy for people with DS? years	1-10	34 (10.1)
	11-20	106 (31.5)
	21-30	82 (24.3)
	31-40	51 (15.1)
	41-50	17 (5.0)
	51-60	10 (3.0)
	older than 60	37 (11.0)
Life expectancy (Categorized)	Incorrect	300 (89.0)
	Correct	37 (11.0)
Are you aware of prenatal screening tests availability?	Yes, I am	227 (67.4)
	No, I am not	110 (32.6)
When do you think is the best time to screen for DS?	Before pregnancy	118 (35.0)
	1st trimester	129 (38.3)
	2nd trimester	48 (14.2)
	3rd trimester	14 (4.2)
	After pregnancy	28 (8.3)
The correct time to screen (Categorized)	Incorrect	208 (61.7)
	Correct	129 (38.3)
Are you aware of prenatal diagnostic tests availability?	Yes, I am	193 (57.3)
	No, I am not	144 (42.7)
Do you support DS children to be enrolled in school with normal children?	Extremely oppose	108 (32.0)
	Oppose	73 (21.7)
	Neutral	36 (10.7)

Question	Answer	N (%)
Do you support DS children to be enrolled in school with normal children?	Supportive	51 (15.1)
	Extremely Supportive	69 (20.5)
Do you support DS adults to have jobs?	Extremely oppose	140 (41.5)
	Oppose	85 (25.2)
	Neutral	31 (9.2)
	Supportive	26 (7.7)
	Extremely Supportive	55 (16.3)
Do you support DS individual's presence in public areas and events?	Extremely oppose	139 (41.2)
	Oppose	88 (26.1)
	Neutral	15 (4.5)
	Supportive	18 (5.3)
	Extremely Supportive	77 (22.8)
Are you familiar with DS support centers in your city?	Yes, I am	119 (35.3)
	No, I am not	218 (64.7)
If you are expecting a child, will you undergo a prenatal screening program?	Yes, I do	281 (83.4)
	No, I do not	36 (10.7)
	I do not know	20 (5.9)
If you are expecting a child with DS, do you think prenatal screening will be beneficial emotionally to you?	Yes, I do	209 (62.0)
	No, I do not	70 (20.8)
	I do not know	58 (17.2)
If you are expecting a child and your physician informed you that your pregnancy is affected by DS, are you willing to terminate or continue?	I will terminate	62 (18.4)
	I will continue	194 (57.6)
	I do not know	81 (24.0)

The results of the univariate and multivariate regression analysis are presented in Table-3. We found at univariate analysis that no significant association between most of the sociodemographic characteristics and the willingness to continue or terminate pregnancy with DS fetus. But there was greater willingness in continuing pregnancy with a DS fetus in illiterate participants (AOR 9.7, 95% CI: 2.2-42.8, P=0.003). On the other hand, (60%) of participants who had a college degree were more willing to terminate the pregnancy with DS fetus (OR 1.6 95% CI: 1.0-2.4, P=0.038). Moreover, participant awareness about prenatal tests and their attitudes were of no significant association with the willingness to terminate the pregnancy. Participants who were married for more than 20 years expressed their likelihood to continue a pregnancy after knowing that the fetus have DS (OR 2.5, 95% CI: 1.5-4.2, P=0.001). Participants who were married for 6-10 years (70%) expressed their willingness to terminate a pregnancy after knowing that the fetus have DS (OR 1.7 95% CI: 1.0- 2.8-, p=0.039).

Table 3. Univariate logistic regression the impact of participants characteristics Toward terminating of pregnancy

Variables	Description	Termination Choice		OR [95% C.I]	P - value
		No (n = 194)	Yes (n = 143)		
Age	<26	15 (7.7)	8 (5.6)		
	26 - 30	45 (23.2)	39 (27.3)	0.8 [0.5 - 1.3]	0.393
	31 - 35	33 (17.0)	34 (23.8)	0.7 [0.4 - 1.1]	0.124
	> 35	101 (52.1)	62 (43.4)	1.4 [0.9 - 2.2]	0.114
Parity	no children	20 (10.3)	15 (10.5)		
	one child	24 (12.4)	24 (16.8)	0.7 [0.4 - 1.3]	0.252
	two children	24 (12.4)	25 (17.5)	0.7 [0.4 - 1.2]	0.188
	>= 3 children	126 (64.9)	79 (55.2)	1.5 [1.0 - 2.3]	0.071
Married duration	0 - 5	37 (19.6)	33 (24.1)		
	6 - 10	37 (19.6)	41 (29.9)	0.6 [0.4 – 1.0]	*0.039
	11 - 15	29 (15.3)	24 (17.5)	0.9 [0.5 - 1.6]	0.647
	16 - 20	21 (11.1)	15 (10.9)	1.0 [0.5 - 2.0]	0.922
	> 20	65 (34.4)	24 (17.5)	2.5 [1.5 - 4.2]	*0.001
Income	> 10000	31 (16.0)	26 (18.2)		
	5100-10000	35 (18.0)	29 (20.3)	0.9 [0.5 - 1.5]	0.605
	3100-5000	40 (20.6)	25 (17.5)	1.2 [0.7 - 2.1]	0.471
	<3000	88 (45.4)	63 (44.1)	1.1 [0.7 - 1.6]	0.812
Educational level	Post-grade	4 (2.1)	9 (6.3)		
	College	81 (41.8)	76 (53.1)	0.6 [0.4 – 1.0]	*0.038
	High school	41 (21.1)	33 (23.1)	0.9 [0.5 - 1.5]	0.670
	Basic school	39 (20.1)	19 (13.3)	1.6 [0.9 – 3.0]	0.101
	Illiterate	29 (14.9)	6 (4.2)	4.0 [1.6 - 9.9]	*0.001
Employment status	Not employed	147 (75.8)	107 (74.8)	1.0 [0.6 - 1.7]	0.842
	employed	47 (24.2)	36 (25.2)		
Residency	City	163 (84.0)	119 (83.2)	1.0 [0.6 - 1.9]	0.844
	Rural	31 (16.0)	24 (16.8)		
Prenatal screening tests availability	Yes, I am	66 (34.0)	44 (30.8)	0.9 [0.5 - 1.4]	0.529
	No, I am not	128 (66.0)	99 (69.2)		
Screening time	Incorrect	124 (63.9)	84 (58.7)	1.2 [0.8 - 1.9]	0.334
	Correct	70 (36.1)	59 (41.3)		
Prenatal diagnostic tests availability	Yes, I am	77 (39.7)	67 (46.9)	1.3 [0.9 - 2.0]	0.189
	No, I am not	117 (60.3)	76 (53.1)		
willingness to undergo a prenatal screening program	Yes, I do	27 (13.9)	9 (6.3)		
	No, I do not	156 (80.4)	125 (87.4)	0.6 [0.3 - 1.0]	0.088
	I do not know	11 (5.7)	9 (6.3)	0.9 [0.4 - 2.2]	0.811
Will prenatal screening be emotionally beneficial	Yes, I do	38 (19.6)	32 (22.4)		
	No, I do not	123 (63.4)	86 (60.1)	1.1 [0.7 - 1.8]	0.542
	I do not know	33 (17.0)	25 (17.5)	1.0 [0.5 - 1.7]	0.910

Discussion

The current study set out to assess women's awareness and attitude about DS. Our findings showed that women beliefs and attitudes towards the social inclusion of individuals with DS are perceived negatively, and intentions of integrating individuals with DS in three aspects (employment, attending public areas and regular school systems) are unfavorable. Another important finding is that most women are willing to undergo a prenatal screening and believed it will offer emotional benefits in their pregnancy but didn't report the correct time for screening. Overall, more than half of women are disinclined to undergo TOP.

Regarding the level of knowledge about DS. The study found that for life expectancy, majority of women thought that individuals with DS have less life expectancy and most believed they only reach 20 years of age. When asked if DS is curable, more than half of women answered with no. These results are in accord with other previous studies.[14,15] Regarding the negative attitudes towards the inclusion of DS individuals, these findings are somewhat surprising given the fact that other similar research shows positive attitudes.[15,16] This intriguing finding may be due to the low levels of knowledge about DS and the minimum representation of individuals with DS in media. Another possible explanation is that, in Arab countries the responsibility of taking care of a child relays on the woman solely, while the man takes on the financial load. Women take on the burden and the stress of looking after children which is increased when taking care of a child with a genetic disease. Mothers reported many stressors for taking care of a child with disabilities.[17,18]

The present study demonstrates a deficiency in the knowledge regarding the best time to undergo prenatal screening. However, the overall awareness of the availability of prenatal screening, including NIPT, was higher than that of previously reported levels in Saudi Arabia[9,14,19] In addition, more than half of women reported emotional benefits of prenatal screening. This finding was also reported in a recent Saudi study[9]. The possible interference is that women want to be prepared mentally and financially before the birth of the child which emphases on the importance of screening and is contrary to other studies reporting more distress with screening results.[20,21]

In countries that follows the Islamic law, a pregnancy confirmed with prenatal diagnosis of DS or other chromosomal abnormalities, can be permitted to be terminated. In Saudi Arabia, it's only permissi-

ble before the 120th days of conception as it's believed that fetal viability occurs after 120th day of pregnancy, beyond that it's only permissible if the life of the mother is in danger. In our study, more than half of women are willing to continue a pregnancy affected by DS. This finding is expected and in line to previous studies conducted in Saudi Arabia where the majority did not accept TOP as an option.[19,22] Moreover, a study conducted in Lebanon showed that participants who were unwilling to undergo TOP for religious reasons were more likely to decline NIPT as well.[23] Up to date information about DS and its screening method must be provided to the public so the participants were handed a brochure containing information about DS risk factors and NIPT after concluding the interview.

Conclusion & limitations

This study has shown lack of knowledge about DS and most Saudi women are willing to undergo NIPT to prepare mentally for the child, but not go as far as TOP. This study also highlights the opinions of women towards the integration of DS individuals. The main strength of this study is the interview-based questionnaire that allows more accuracy than an online distributed questionnaire. However, we didn't investigate the possible effect between knowledge and negative attitudes regarding DS with any related factors. Further research on these questions would be useful to explore the reasons for the stigma surrounding the inclusion of DS individuals.

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