Original Article

Are Pregnant Women Adequately Equipped for Autonomy in Pregnancy Screening?

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Abstract

Introduction: Aneuploidy screening is widely practised in the field of obstetrics in current times. This study thus aims to gain an insight on pregnant women's knowledge and risk perception of Down syndrome and first trimester screening (FTS), as well as their views on various potential pregnancy outcomes and how these may affect their decision-making processes. Materials and Methods: A cross-sectional questionnaire-based qualitative study of consecutive 50 women choosing to undergo FTS at KK Women's and Children's Hospital (KKH), Singapore was conducted. The women completed a questionnaire after their FTS pretest counselling session. Basic knowledge of Down syndrome and FTS as well as participants' risk perception with regards to various cut-off values used in FTS were examined. Patients' views of various potential pregnancy outcomes were also studied. Results: Most patients had good retention and comprehension of what FTS entailed after a FTS counselling session at the KKH Antenatal Monitoring Clinic. However, knowledge of the risks of invasive diagnostic testing was poor. Patients also did not possess an adequate understanding of FTS risk values. With regards to risk perception, patients had very different views on acceptable pregnancy outcomes and what constituted a high-risk FTS value to them personally. A significant number of women were concerned even at medically low-risk values of 1:500 and 1:1000 in FTS. The majority of patients viewed highest detection rate followed by a lowest false positive rate as the more important factors impacting their choice of a Down syndrome screening test. Conclusion: This study demonstrates the diversity of pregnant women's risk perception, risk aversion and participation in decision processes when there are 2 different values in competition. The study also highlights our patients' gaps in knowledge and lack of understanding of risk values used in FTS.

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Key words: Choice, Decision-making, Pregnancy, Prenatal test, Risk assessment

Introduction

The concept of first trimester screening (FTS) to evaluate the risk of fetal aneuploidies was first introduced in 1997 by Orlandi et al.[1] The Fetal Medicine Foundation, London, has refined and populated this screening test that uses a combination of maternal age, nuchal translucency (NT) thickness and biochemical markers, namely beta-human chorionic gonadotropin (beta-hCG) and pregnancy-associated plasma protein A (PAPP-A), at 11 to 13^{+6} weeks gestation to determine one's risk of having a pregnancy with aneuploidies and a broader range of pregnancy disorders.[2] Since its introduction, many developed nations around the world have been offering this screening test to pregnant women as part of routine antenatal care, with many patients choosing to undergo it. However, there is relatively little published knowledge on whether these patients understand their own motivations for choosing to undergo the test, and the potential impact of the results of the screening test on their pregnancy, be it emotionally in terms of anxiety regarding the results, or physically if invasive diagnostic testing is done following a high-risk FTS. In addition, we also recognise that as individuals, one's perception of risk may differ from another, and as such, their decision-making processes and preferences may differ accordingly as well.

This study aims to assess our patients' knowledge on Down syndrome screening and examine their individual risk

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perception, risk aversion, preferences with regards to what they view as important in a Down syndrome screening test, as well as their views on selected pregnancy outcomes to see if these factors in combination affect their decision processes. This information may then allow us to better counsel patients before their decision to undertake aneuploidy screening, as well as to tailor post-screening recommendations based on patients' personal risk perception and preferences.

Materials and Methods

This study was a qualitative cross-sectional study conducted between March 2012 to July 2012. All women presenting for antenatal care in KK Women's and Children's Hospital (KKH) were offered assessment of risk for trisomy 21, 18 and 13 by a combination of maternal age, fetal NT thickness, and maternal serum free beta-hCG and PAPP-A at 11 to 13^{+6} weeks gestation as per local guidelines' recommendations. Patients who had chosen to undergo FTS for aneuploidies were invited to participate in the study. Patients were reassured that their participation or non-participation would not affect their antenatal care in any way. In addition, none of the researchers were involved in the direct care of the patients. Approval of the study was attained from Singhealth's Centralised Institutional Review Board (CIRB), and waiver of consent was obtained.

All eligible women were given a detailed information sheet explaining the background and aims of the study immediately after they had undergone their FTS pretest counselling session. During this pretest counselling session, patients were counselled by trained nurse counsellors with regards to the risk factors for Down syndrome, implications of a child with Down syndrome, high risk cut-off values for aneuploidy screening, as well as the available options for subsequent management of a pregnancy with a high-risk screening test result, including chorionic villus sampling (CVS) or amniocentesis. This being a qualitative study, it was decided that an arbitrary number of 50 patients would be enrolled into the study over the 2-week period.

A questionnaire was designed to assess the knowledge of patients with regards to aneuploidy screening and Down syndrome, and explore their preferences of what factors they regarded as important in a Down syndrome screening test. Risk perception of patients was also examined by presenting to them various FTS reported risks of Down syndrome in the form of numerical values and pictorial representations on a diagram depicting 1000 women with the affected women (pregnancies with Down syndrome) highlighted in red, with subsequent questions on whether they would be concerned and if they would do invasive testing at these reported Down syndrome screening risks. A question which aimed to examine patients' personal views regarding possible pregnancy outcomes was also included in the questionnaire, with patients being asked to rank in order of preference which pregnancy outcomes were most or least acceptable to them. The questionnaire was given out to participants of the study immediately after FTS pretest counselling was completed. Multilingual purpose-trained staff were on standby to answer any queries or clarifications patients may have regarding the questionnaire to allow for a more complete collation of responses. Data of the survey was collated and analysed using Microsoft Excel statistical analysis package.

Results

Study Population

Fifty patients were interviewed over a period of 2 weeks at the Antenatal Monitoring Centre at KKH. The characteristics of the study population are summarised in Table 1.

Table 1. Characteristics of Study Population and Singapore's Population

	Study Population n = 50 (%)	**Singapore's Population* (%)**
Race		
Chinese	35 (70)	74.1
Malay	5 (10)	13.4
Indian	8 (16)	9.2
Others	2 (4)	3.3
Age		
<35 years	42 (82)	-
>35 years	9 (18)	-
Type of housing		
HDB 1/2 room flat	4 (8)	4.6
HDB 3/4/5 room/ executive flat	43 (86)	77.5
Condominium	2 (4)	11.2
Terrace house/bungalow	0 (0)	5.7
Missing	1 (2)	-
Educational attainment		
High school graduate or less	23 (46)	77.2
Attained a college degree	27 (54)	22.8
Religion		
Christianity	8 (16)	18.3
Islam	7 (14)	14.7
Hinduism	6 (12)	5.1
Buddhism	16 (32)	33.3
Taoism	2 (4)	10.9
Free-thinker	11 (22)	17
Parity		
Primiparous	32 (64)	-
Multiparous	16 (32)	-

*Census of Population 2010 Statistical Release 1. Demographic Characteristics, Education, Language and Religion. Department of Statistics, Ministry of Trade and Industry, Singapore. Available at: http://www.singstat.gov.sg/publications/publications_and_papers/cop2010/census_2010_release1/cop2010sr1.pdf. Accessed 23 December 2013.

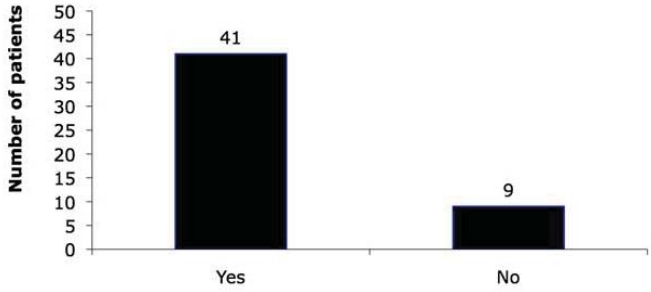

Fig. 1. Number of patients who responded to the question, "Is maternal age related to an increased risk of Down syndrome?".

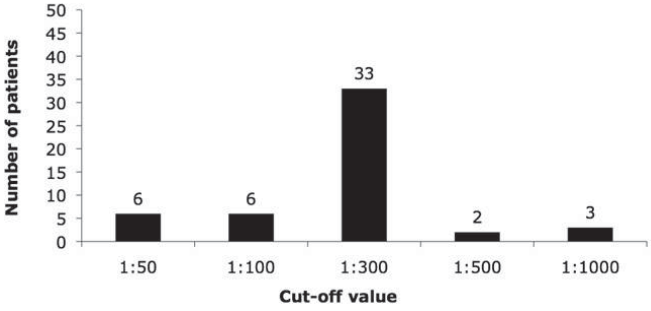

Fig. 2. Number of patients who responded to the question, "What current value is considered "high-risk" for Down syndrome in the FTS test in KKH?".

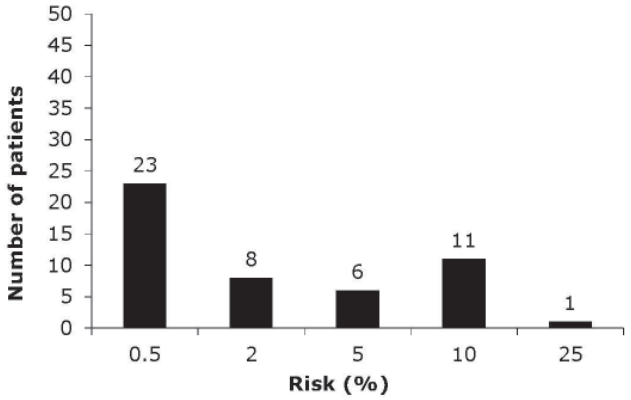

Fig. 3. Number of patients who responded to the question, "What is the risk of miscarriage associated with CVS/amniocentesis?".

Knowledge

In general, most patients had good retention and comprehension of what FTS entailed after a FTS counselling session at the Antenatal Monitoring Clinic. A total of 41 patients correctly identified that increased maternal age was a contributing factor in the risk for Down syndrome in pregnancy (Fig. 1). Also, 33 patients (66%) could recall KKH's cut-off value for high-risk as 1:300 (Fig. 2).

However, patients' knowledge of the risks of invasive diagnostic testing was poor. Eleven (22%) patients thought the risk of miscarriage was as high as 10% (Fig. 3). Patients also did not possess an adequate understanding of FTS risk values. When asked what a high-risk value of 1:50 in FTS meant to them, 34 patients had the incorrect impression that a high-risk value equated to the fetus either having Down syndrome most or all of the time (Fig. 4).

Preferences

Patients were asked to rank in order of preference which factors were important to them in a Down syndrome screening test (Fig. 5). The 4 options given were "lowest false positive rate", "highest accuracy", "fastest results" and "lowest cost". A total of 32 patients chose "highest accuracy" as the most important factor in a Down syndrome screening test. This was followed by 7 patients who ranked "lowest false positive rate" as the most important factor. Only 1 patient regarded cost of the screening test as the most important determining factor for a Down syndrome screening test.

Risk Perception

Patients were shown various FTS risks of Down syndrome in the form of numerical values and pictorial representations on a diagram depicting 1000 women with the affected women (pregnancies with Down syndrome) highlighted in red. They were asked if they would be concerned and if they would do invasive testing at these risks.

In our study, it was observed that the higher the reported risk of Down syndrome based on the FTS test, the larger would be the number of patients who would choose to do invasive testing for Down syndrome. However, 17 and 12 women expressed that they would still be concerned at a risk of 1:500 and 1:1000 respectively. Also, some patients opted for invasive testing even at low-risk values of 1:500 and 1:1000. It was also noted that the number of women who would undergo invasive diagnostic testing for each FTS risk quoted was smaller than the number of women who were concerned for each corresponding FTS risks (Fig. 6).

In a subgroup analysis of the 14 patients who had a correct understanding of FTS risk values as shown in Figure 4, 8 out of 14 were still concerned at risk of 1:500 and 5 out of 14 were still concerned at risk of 1:1000.

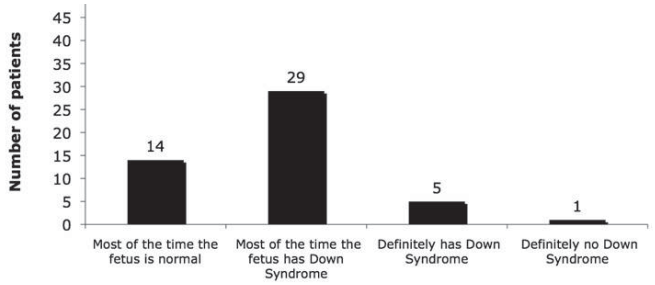

Fig. 4. Number of patients who responded to the question, "If FTS result is high risk e.g. 1:50, what does it mean for the pregnancy?".

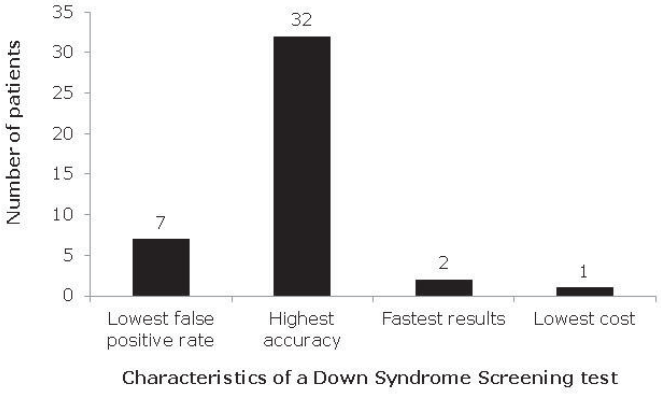

Fig. 5. Number of patients who responded to the question, "Which of the following is most important to you in a Down syndrome screening test?".

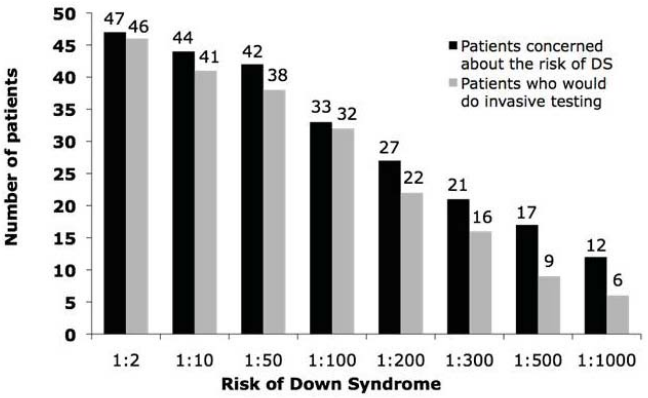

Fig. 6. Number of patients who responded to the question on the "Relationship between Down syndrome risk and threshold for concern and invasive testing".

Desired Pregnancy Outcomes

The question in Figure 7 aimed to assess patients' personal views of various possible pregnancy outcomes. Of the 50 patients involved in the survey, 39 patients ranked the outcomes in entirety, with 11 patients leaving blanks for certain options and were thereafter excluded from this aspect of the analysis. The most desired pregnancy outcome chosen by all patients was predictably "having a healthy child". Interestingly, the choice of least desired pregnancy outcomes varied greatly among the patients, as represented in the graph in Figure 8. The majority (26 out of 39 patients) felt that having a child with Down syndrome was their least desired pregnancy outcome of all the given options. However, 8 patients ranked "losing a fetus with Down syndrome through a chosen abortion" as their least desired pregnancy outcome. In addition, in a subanalysis of results with specific reference to the second and fourth options, 12 patients ranked having a child with Down syndrome as more acceptable than losing a healthy fetus through a miscarriage as a complication of chorionic villus sampling (CVS) or amniocentesis.

Discussion

This study on FTS is focused on patient autonomy, and analyses Down syndrome screening in particular. The College of Obstetricians and Gynaecologists, Singapore (COGS) recommends that all women, regardless of age, should be considered to be at risk for fetal aneuploidy and should be offered screening for Down syndrome, and they should be made aware of the availability of screening tests for Down syndrome and other chromosomal abnormalities.[3] This recommendation is similarly reflected in many other countries' guidelines, such as that of the National Institute for Health and Care Excellence (NICE),[4] American Congress of Obstetricians and Gynecologists (ACOG),[5] and joint Human Genetic Society of Australasia (HGSA) and Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG)[6] guidelines. However, adequacy on the exercise of patient autonomy has not been examined widely. As advocates for our patients, we need to understand our patients' motivations for undergoing complex screening, and assess whether they understand the potential psychological, physical and emotional impact of doing so. It is also important for us to make sure that our patients' knowledge of Down syndrome and other aneuploidies are accurate and up-to-date such that they may be able to make informed decisions regarding screening so as to achieve true patient autonomy, which is one of the 4 important principles in the field of biomedical ethics.[7]

Currently, patients in KKH undergo counselling prior to deciding whether or not to undergo the FTS test. They are presented with information regarding the various risk factors

Rank in order the below possible pregnancy outcomes from 1 to 7, 1 being the pregnancy outcome you wish **most** to happen, 7 being the pregnancy outcome you wish **least** to happen.

_____ Having a healthy child
_____ Having a child with Down syndrome
_____ Losing a healthy fetus through a spontaneous miscarriage
_____ Losing a healthy fetus through a miscarriage as a complication of chorionic villus sampling or amniocentesis
_____ Losing a fetus with Down syndrome through a spontaneous miscarriage
_____ Losing a fetus with Down syndrome as a complication of an amniocentesis or chorionic villus sampling
_____ Losing a fetus with Down syndrome through a chosen abortion

Fig. 7. Ranking question on pregnancy outcomes.

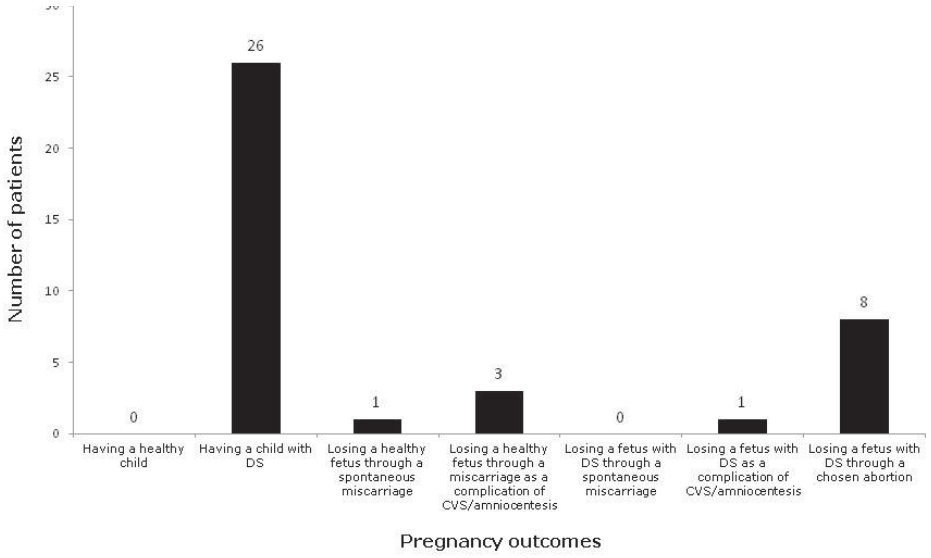

Fig. 8. Number of patients who responded to the question on the "Least desired pregnancy outcomes".

for Down syndrome, the cut-off value that would indicate a high-risk FTS, and the procedure of invasive diagnostic tests with its associated risks. Having undergone these sessions, the majority of our patients were able to identify maternal age as a risk factor for Down syndrome, and 66% of the patients surveyed were able to recall KKH's cut-off for high-risk in the FTS test. Nevertheless, certain gaps in knowledge of our patients regarding Down syndrome still exist. A total of 34 patients (68%) had the incorrect impression that a high-risk value equated to the fetus either having Down syndrome most or all of the time. These gaps in knowledge and understanding of the FTS test may affect patients' ability to make informed decisions regarding FTS and invasive testing, and these gaps need to be bridged in order to achieve true informed decision-making.

We also saw from the survey that patients' preferences with regards to the type of FTS test they would choose to undergo vary from individual to individual. Most of the patients felt that highest accuracy was of highest priority in a FTS screening test, followed by lowest false-positive rate, timing of the test and lastly cost of the test. In other simple preference studies, women report preferring an earlier over a later test, mainly because of the easier termination of pregnancy and/or the earlier reassurance provided.[8] In more recent times, the advent of non-invasive prenatal testing for trisomy 21, 18 and 13 using cell-free DNA (cfDNA), whilst understanding its inherent limitations, is possibly a step towards providing a test with a higher sensitivity and lower false-positive rate.[9] Nonetheless, this study and most other studies have shown that it is difficult to generalise each patient's preference with regards to choice of a screening test. Information for consideration of a choice (consent process) should thus include descriptions of different types of tests available and the benefits and disadvantages of each of them, such that patients can make decisions based on what they consider to be important to their circumstances and context. Otherwise, if the availability of the different choices of screening tests (that are supported by adequate

evidence) or important information regarding these tests is withheld from the patient, and the eventual decision of the patient conflicts with their personal values as a result of the omission, it could be considered unethical behaviour on the part of the medical practitioner.[10]

We see in our study that patients' perception of risk may not reflect the reported risk. This was observed in a study by Georgsson et al[11] in 2009 and is reflected in our study, where a number of women were concerned even at low-risk values of 1:500 and 1:1000. This may either be a reflection of the poor understanding of risk values or an inherent higher risk aversion in some patients as a result of them coming to screening with different backgrounds and life experiences, which impact on their decision-making processes. This highlights that doctors' perception of high- and low-risk does not automatically equate to patients' perception of risk, therefore when counselling patients, we need to take into consideration that even low-risk values may lead to patient anxiety and concerns.

Our study also showed how patients' perception of risk of Down syndrome does parallel with their willingness to undergo invasive testing procedures (Fig. 6). These results are consistent with a previous study published by Nicolaides et al[12] in 2005, which examined whether pregnant women were able to incorporate sophisticated screening information about risk assessment into their decisions about invasive testing in an appropriate way. In Nicolaides' study, it was seen that the rate of invasive testing increased exponentially with increasing estimated risk, and this observation was noted to be significant. However, in a retrospective study by O'Connell et al,[13] 15% of women who had a positive test did not agree with the cut-off of 1:250 and declined invasive testing, while 0.08% of women with a negative triple test result requested for amniocentesis. This is similarly observed in our study, where women indicating concern about a given risk figure did not automatically commensurate with a decision to undergo invasive testing. This again may be due to different degrees of risk aversion and differing acceptability of various pregnancy outcomes. When faced with a high-risk screening result, discussion with the patient should consider her risk aversion to invasive testing and the values she attaches to different pregnancy outcomes to help decide on the next course of action.

Patients also have very different views on various pregnancy outcomes. Eight patients ranked having a chosen abortion for a fetus with Down Syndrome as the least desired pregnancy outcome of the given options, and 12 patients ranked having a child with Down syndrome as more acceptable than losing a healthy fetus through a miscarriage as a complication of CVS or amniocentesis. This highlights to us that different women view various pregnancy outcomes differently. Thus, after receiving an increased screening result, obstetricians need to convey the message that invasive testing is not a self-evident step for them to take, but rather, an informed decision regarding invasive testing should be made based on the values an individual woman attaches to the possible pregnancy outcomes.

Strengths and Limitations

One of the strengths of our study was the providence of purpose-trained Malay and Chinese-speaking interviewers for patients who were not fluent in English, allowing the inclusion of as many women as possible to reduce selection bias. Patients were also allowed to clarify uncertainties regarding the questions with the interviewers, such that their answers would most reflect their true values and preferences. The inclusion of pictorial representations of risk rather than solely a numerical risk value helped to maximise their understanding of risk by focusing on the numerator (number of affected women) versus the denominator (number of unaffected women).

Currently, a validated questionnaire for risk perception in FTS does not exist. As such, one limitation of the study was that questions had to be created to assess risk perception with regards to FTS. In addition, this study was based on a relatively small sample size of 50 women, and thus, it cannot be generalised to depict all women choosing to undergo FTS. Also, the results should be viewed in a local context.

Conclusion

Aneuploidy screening is widely practiced in the field of obstetrics in current times. As such, examining the impact of this hitherto lesser considered aspect (autonomy) of screening on our patients is of substantial importance. This study demonstrates the diversity of pregnant women's risk perception, preferences, risk aversion and participation in decision processes when there are 2 different values in competition. The study also highlights our patients' gaps in knowledge and lack of understanding of risk values used in FTS. More needs to be done in the form of educating our population to address and bridge these gaps before a patient can truly exercise her autonomy to make informed decisions for her own pregnancy.

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