Why do pregnant women accept or decline prenatal diagnosis for Down syndrome?

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Abstract To investigate if actual knowledge of Down syndrome (DS), influences the decision to accept or decline prenatal diagnosis (PND). Secondary aims were to elucidate reasons for accepting or declining PND and investigate differences between the accepting and declining group in perceived information, knowing someone with DS and thoughts about decision-making. A questionnaire was completed by 76 pregnant women who underwent invasive testing and 65 women who declined tests for chromosomal aberrations in Uppsala, Sweden. Apart from one question no significant differences were found in knowledge of DS between women declining or accepting PND for DS. Both groups had varying and in several respects low levels of knowledge about DS and its consequences. Most common reasons to accept PND were ‘to ease my worries’ and ‘to do all possible tests to make sure the baby is healthy’. Corresponding statements declining PND were ‘termination of pregnancy is not an option’ and ‘because invasive tests increase the risk of miscarriage’. More women declining PND knew someone with DS. Knowledge of DS at these levels is not a major factor when women decide to accept or decline PND for DS. Their choice is mostly based on opinions and moral values.

Keywords Prenatal diagnosis · Down syndrome · Knowledge of condition screened for · Decision-making · Informed choice

Introduction

According to Swedish law (Swedish Law on Genetic Integrity. Swedish Code of Statutes 2006), all pregnant women should be offered information on prenatal screening and diagnosis. If accepted, this general information should be given at the antenatal clinic; thereafter, there should be ample time to reflect before the decision to accept or decline prenatal diagnosis (PND) is made (National Board of Health and Welfare in Sweden 2013). The information should be sufficient and well balanced to enable the pregnant woman to make an informed choice regarding PND and to make a choice in line with her own needs and values.

The prenatal screening systems employed in Sweden is similar to that of many other healthcare systems within Europe. In the Uppsala region, both invasive tests and combined ultrasound and biochemistry (CUB) test are available to pregnant women. Women over 35 who wish to have an invasive test (amniocentesis or chorionic villus sampling), without a previous CUB-test, are allowed such a test, whereas younger women are allowed the tests as far as capacity of the Fetal Medicine unit allows. In addition to a high CUB-probability, other reasons such as anxiety, advanced maternal age, ultrasound findings and a previous pregnancy with a chromosomal aberration can motivate invasive testing. The CUB-tests are offered at a cost of 30 € to women over 35 and to younger women at a higher cost of 160 €. The cost of an invasive test is 30 €.

In a qualitative study, Garcia et al. (2008) found that both women accepting and declining prenatal screening tests described their parental responsibilities as being context dependent, and all admitted that their choice could be different

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in different circumstances. A substantial difference between those accepting and those declining prenatal screening was the expected burden of the emotional and physical impact that a disabled child would have on them and their family (Garcia et al. 2008). A prerequisite to estimate such factors concerning DS is knowledge about medical, cognitive and social consequences of DS. Thus, one could argue that knowledge about the condition tested for could affect pregnant women's decision to accept or decline PND for DS.

The aim of this study was to investigate whether today's knowledge levels about DS could be a factor influencing the decision to accept or decline PND. Secondary aims were to elucidate reasons for accepting or declining PND and to examine differences between the accepting and declining group in perceived information, knowing someone with DS as well as thoughts about decision-making concerning invasive procedures and termination of pregnancy.

Method

A prospective cohort study with a descriptive and comparative design was conducted based on two five-page long questionnaires consisting of three parts, with a total of 38 questions. The first part of the questionnaire included socio-demographic characteristics such as age, education, marital status and previous children. The second part focused on reasons for the women to accept an invasive test or decline testing for chromosomal aberrations, the specific kind of information they had received and if they were satisfied with that information. For those women who accepted invasive testing, there were additional questions on decision-making in the event of a DS-diagnosis. The last part contained questions concerning medical, cognitive and social consequences of DS. Most questions were multiple-choice format, with space for free comments.

According to the procedures in Uppsala, when a pregnant woman books her first appointment at the antenatal care unit, she is asked if she wishes to receive information about PND. If she accepts, then the midwife provides information at the first visit. The woman can then choose to undergo CUB-testing or invasive tests at the Fetal Medicine Unit at Uppsala University Hospital. Even if the woman declines both CUB and invasive tests, almost every woman accepts the routine mid-trimester fetal ultrasound scan, which is an examination that is not primarily focused on chromosomal aberrations.

Pregnant women who came to the Fetal Medicine Unit for invasive tests or those who declined PND tests for DS, between November 2010 and April 2011, were invited to participate. The group who came for invasive testing was included at 13–17 weeks of gestation, and the group who declined PND was included at 16–18 weeks when the routine mid-trimester fetal ultrasound scan was performed. Only women over 35 were included in the declining group to assure

that everyone had received information and an offer to undergo PND for chromosomal diagnosis and thus made an active choice. Most women included in the accepting group came directly for invasive testing, without a previous CUB-test (83 %). Only persons with a good command of the Swedish language were included. The questionnaires were completed directly after the examinations. The intention was to include participants sequentially, e.g., all pregnant women who came during this time period and had good command of the Swedish language should be asked to participate. However, invitations were canceled during periods of high clinical workload. The questionnaires were collected anonymously, and no record was made of non-participants; however, the majority consisted of persons with difficulties in the Swedish language and those not invited due to a high clinical workload.

The questionnaires were anonymous with no possibility of identification. Participation was optional and did not affect the care given at the unit. The study was approved by the ethics committee at the Medical Faculty of Uppsala University (2010/403).

The statistical analyses were performed using the IBM Statistical Package of Social Sciences (SPSS) version 22, Armonk, New York. One of the predefined response alternatives from the questions regarding knowledge about DS was defined as correct. In the analyses the responses were dichotomized into correct and incorrect. For calculations of differences in knowledge (correct vs. incorrect) between women accepting invasive testing and women declining PND, a two-tailed Chi-square test was used. Among the women who answered incorrectly on the knowledge questions, the Chi-squared test was also used to calculate differences in overestimating and underestimating between the accepting and declining group. For comparison between groups regarding other questions, the independent sample *T* test was used. The significance levels were set to 0.05. A power calculation was done concerning the knowledge questions. In order to obtain a power of 80 % with a significance level of 0.05, a sample of 94 participants would be needed in each group to be able to find a difference of 20 per cent units, assuming that 50 % of the control group (accepting group) answered the knowledge questions correctly. However, due to difficulties in recruiting participants and in order to avoid a too long study period, only 76 participants in the accepting group and 65 in the declining group were included. With this number of participants, a difference of 23 per cent units could be detected.

Results

The mean age for women accepting invasive testing was 35 years and for women declining PND 37 years. More women in the declining group were multiparous. Detailed data on the participants appear in Table 1.

Table 1 Data concerning the 76 pregnant women accepting PND and the 65 pregnant women declining PND

	Accepting PND		Declining PND	
	<i>n</i>	%	<i>n</i>	%
Total	76	100.0	65	100.0
Age ≥ 35 years	45	59.2	63	96.9
Residence				
Suburban	21	27.6	24	36.9
Urban	54	71.1	40	61.5
Co-habiting	71	93.4	64	98.5
Education				
Elementary school or High school	26	34.2	18	27.7
University	50	65.8	47	72.3
Previous children	47	61.8	54	83.1

The questions regarding residence and previous children were not answered by all participants

The reasons to accept or to decline PND for DS are shown in Table 2. Among women in the accepting group the most commonly stated reasons were “to ease my worries” and “to do all possible tests to make sure the baby is healthy”. Other stated reasons given as comments were high age, DS or a sick child in the family or sick child in a previous pregnancy. Among women declining PND the most common reasons were “termination of pregnancy is not an option” and

“because invasive testing increases the risk of miscarriage”. Other stated reasons were “ethical reasons,” “that everyone has a right to be born” and “not knowing what to do with the information if the test indicates DS.”

Table 3 shows participants who had someone with DS in their vicinity. Significantly more women declining PND (28 %) knew someone with DS, compared to women choosing to undergo invasive testing (13 %) ($p = 0.03$).

When comparing the groups regarding information received prior to accepting or declining PND, significantly more women declining PND were satisfied with the information ($p = 0.01$) and how it was given ($p = 0.01$). Significantly more women declining PND (34 %) felt they had received sufficient and adequate information about what it means to live with a child with DS ($p = 0.02$), compared with women choosing PND (20 %).

Both groups had varying and in several respects low levels of knowledge about DS and its consequences (Table 4). Apart from one question, no significant differences were found when comparing the level of knowledge between participants accepting or declining PND. No significant differences were recorded when comparing the two groups in regard to over- or underestimating.

Of the women who underwent invasive testing, 21 % had not decided what to do with the pregnancy if DS was diagnosed. Seventy-four per cent responded that they

Table 2 The main reasons for accepting or declining PND for DS

	Accepting PND		Declining PND	
	<i>n</i>	%	<i>n</i>	%
Why accept invasive testing (PND)?				
Because you should do it if older than 35	18	23.7		
To perform all possible tests available to make sure the baby is healthy	34	44.7		
To ease my worries	48	63.2		
Because I/my partner have/has a previous child with an illness	5	6.6		
Screening indicated high risk	10	13.2		
Because my partner wanted to	6	7.9		
Because of the cost of CUB-testing	1	1.3		
Why decline PND?				
Termination of pregnancy is not an option			38	58.5
Can take care of a child with DS			28	43.1
I have a low risk			19	29.2
DS is not a serious condition			23	35.4
Because invasive testing increases the risk of miscarriage			37	56.9
Because of the cost			1	1.5

Several response alternatives could be given. The data concern responses from the 76 pregnant women accepting PND and the 65 pregnant women declining PND. Other reasons were given by 28 participants from the accepting group and 8 participants from the declining group.

PND prenatal diagnosis, DS Down syndrome

Table 3 Participants who had someone in their vicinity with DS

	Accepting PND		Declining PND	
	<i>n</i>	%	<i>n</i>	%
Do you have someone with DS in your vicinity?				
Yes	10	13.2	18	28.1
- In my family	2	2.6	5	7.8
- In my circle of friends	7	9.2	13	20.3
- Both in my family and circle of friends	1	1.3	0	0.0

The data concern responses from the 76 pregnant women accepting PND and the 65 pregnant women declining PND. One participant in the declining group did not answer the questions.

DS Down syndrome, PND prenatal diagnosis

might consider a termination of pregnancy if DS was diagnosed, while 5 % answered that they would prepare to take care of the child.

Discussion

Our primary aim was to investigate if knowledge of the condition searched for, here DS, could be a factor influencing the decision to accept or decline PND. Apart from one question, which may well be due to chance, no significant differences were found in knowledge of DS among women who declined or accepted PND for DS. Moreover, women accepting PND did not have a more negative view of DS. Both groups had varying and in several respects low levels of knowledge about DS and its consequences not different from our previous report on women and partners coming for a CUB-test (Ternby et al. 2015). Thus, knowledge at these levels does not substantially influence the decision to accept or decline PND. The most common reasons to accept PND were “to ease my worries” and “to do all possible tests to make

sure the baby is healthy.” The corresponding statements declining PND were “termination of pregnancy is not an option” and “because invasive tests increase the risk of miscarriage.” More women who declined PND knew someone with DS.

Our quantitative data show that variation in knowledge about DS is not a major factor when making a decision whether to accept or decline PND for DS. A minor qualitative study by Gottfredsdottir et al. (2009a) pointed in the same direction, that is, that expectant parents who underwent DS-screening did not base their decision to participate on knowledge. Michie et al. (2003) reported that knowledge about the test method did not seem to influence the decision. Knowledge might, however, affect the psychological and emotional response after receiving the final test result and help to reduce decisional conflict (Dahl et al. 2006). Based on our study, we are unable to answer the question of whether greater knowledge would affect the decision making or the decisions made, as that would require an interventional study.

A substantial number of women declining PND felt they “could cope with the task of caring for an affected child” and many of them “did not think DS was a severe enough

Table 4 Participants from the accepting and declining group who answered correct versus incorrect on the knowledge questions about DS and families with a child with DS

Knowledge questions	Correct (<i>n</i>)		Incorrect (<i>n</i>)		<i>p</i> value
	Accept	Decline	Accept	Decline	
1. Mean survival age	38	34	37	28	0.63
2. How many learn to walk?	65	45	10	18	0.03
3. How many learn to talk?	35	31	40	31	0.70
4. How many learn to read?	26	13	48	50	0.06
5. Frequency of heart anomalies?	33	33	41	28	0.27
6. Frequency of gastrointestinal anomalies?	50	40	23	20	0.82
7. Frequency of hearing problems?	9	8	65	53	0.87
8. How many live with biological parents?	49	37	26	27	0.36
9. Number of siblings	35	35	38	29	0.43
10. Divorce rate for parents with DS-children	49	36	26	28	0.27
11. Frequency of parents working	41	36	34	28	0.85

The data concern responses from 76 pregnant women accepting invasive testing and 65 pregnant women declining PND. Some questions were not answered by all participants. DS Down syndrome, PND Prenatal diagnosis

condition to justify termination of pregnancy.” Similar results have previously been reported by Garcia et al. (2008) and Gottfredsdottir et al. (2009b). Moreover, many women declining PND did so because they could not consider terminating the pregnancy, a finding in line with Garcia et al. (2008). In contrast, women accepting PND did not automatically link PND with termination of pregnancy if DS was diagnosed. Rather, they saw it as a way to gain information, to be reassured that the fetus was healthy and to ease their worries, results that resemble observations from previous reports (Santalahti et al. 1998; Garcia et al. 2008; Ternby et al. 2015).

Women declining PND more often had someone with DS in their vicinity (Table 3). Does having personal experience with DS infer a more positive attitude toward DS, leading to a decision to decline PND for DS (Santalahti et al. 1999; Gottfredsdottir et al. 2009b)? Or is there an element of guilt involved in the decision to undergo PND for DS? On the other hand, Bryant et al. (2005) found that a personal experience of a family member with DS could result in a negative view of DS if the perceived impact on the persons themselves and their family was negative. It might be a difference between having someone with DS in the immediate family or among friends in how the impact on the family is perceived. This could explain our findings that some women in the accepting group chose invasive tests “because of DS or a sick child in the family” while more women declining PND knew someone with DS.

Women declining PND were more satisfied with the received information, which could be explained by a finding by Dahl et al. (2006) indicating that satisfaction with information is unrelated to knowledge, but associated with having one’s expectations met. Women who declined PND because termination of pregnancy was not an option were probably not in need of extensive information. In contrast, women who underwent PND without being sure what to do if a DS-pregnancy was diagnosed probably wanted extensive information as a basis for their decision. The choice to decline PND might be more founded on a feeling while the choice to accept PND might be driven by a need for more information concerning the pregnancy.

In a previous study on expectant parents who underwent a CUB-test (Ternby et al. 2015), we found that 43 % of the participants responded that they might consider termination of pregnancy if DS was diagnosed, a figure far below the 80.5 % reported by Gekas et al. (1999) who asked women who underwent an invasive test. In the present study, however, 74 % of women who underwent an invasive test reported that they might consider a termination of pregnancy if DS was diagnosed, a result well in line with that of Gekas et al. (1999). It might well be that women who underwent an invasive test had gone through a more meticulous decision-making process, compared to women who underwent a screening procedure, as invasive tests are diagnostic and involve increased risks for the pregnancy. Screening tests might

be decided on more easily since no such risks are involved. Moreover, screening tests might also be viewed as routine procedures that do not require a decision (Press and Browner 1997; Ternby et al. 2015).

Strengths and limitations

One of the strengths is that data were collected in direct connection with the invasive testing or the routine ultrasound. Thus, our results reflect the knowledge and opinions held by the participants at the time when it was of immediate interest. The questionnaires were completely anonymous, in order to encourage participants to respond to questions openly, without concern for personal identification or repercussion. However, a weakness with anonymous questionnaires is the impossibility to follow up non-participants, but we have no reason to believe that the recruitment of participants was systematically skewed. Since a majority of women took part in a CUB-screening program we met recruitment difficulties resulting in fewer participants than originally planned. The questionnaire was developed specifically for the study and thus not validated. The knowledge questions were of multiple-choice character, which may fail to capture the full range of contextual factors involved. On the other hand, having open questions would have made it more difficult to perform analyses and comparisons. Since non-Swedish speaking women were not included, the generalizability might be somewhat limited.

Conclusion

Knowledge of DS, on these levels, is not a major factor when women decide to accept or decline PND for DS. Their choice is mostly based on opinions and moral values. We see these results as important when developing guidelines for prenatal counseling. There is a need for more research, where a qualitative approach could allow for stronger claims to be made around motivation for testing.

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Compliance with ethical standards The present study has been approved by the Regional Research Ethics board in Uppsala (2010/403) and was performed in accordance with the ethical standards as laid down in the 1975 Declaration of Helsinki (revised 2000). Informed consent was obtained from all individual participants included in the study. The study was financially supported by funds from Allmänna BB:s minnesfond and Uppsala University. The funders were not involved in study design, data collection, data analysis, manuscript preparation, or publication decisions. The authors declare no conflict of financial or non-financial interest.

Ethical approval The present study has been approved by the Regional Research Ethics board in Uppsala #2010/403. All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1975 Helsinki declaration (revised 2000). Informed consent was obtained from all individual participants included in this study.

Conflict of interest The authors declare that they have no conflicts of interest.

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