Knowledge, attitudes and decision-making in Czech women with atypical results of prenatal screening tests for the most common chromosomal and morphological congenital defects in the fetus: Selected questionnaire results

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Aims. The primary aim was to investigate variables affecting compliance in pregnant women recommended for genetic consultation for abnormal screening test results, family predisposition or medical history. Our main focus was on a women's knowledge of particular screening tests, their initial feelings and changes in these feeling with time, as well as variables relevant to further decision making.

Methods. We used an anonymous questionnaire based on previous qualitative research. The questions were formulated by a medical geneticist, and the questionnaires were distributed prior to prenatal screening tests performed by doctors or trained nurses. The research cohort consisted of 271 women aged 16-42 years. Six hypotheses were tested using the statistical programme STATISTICA; significance levels were set to $P < 0.05$.

Results. The questionnaire results showed insufficient knowledge. The women were confused about invasive, screening and ultrasound tests. Genetic test recommendation was largely associated with stress in these patients. Between recommendation and consultation, the women mostly looked for support from their partners. There was a surprisingly low percentage of women who looked for help from their medical specialists and a surprisingly high percentage of those who did not seek any help at all.

Conclusion. Women's distress can be reduced if the information about recommended genetic consultation is conveyed correctly and this can also help them make the right informed decision about their future course of action.

Key words: prenatal counselling, screening programmes, genetic tests, invasive tests, amniocentesis, karyotyping, stress, anxiety, fear, questionnaire

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INTRODUCTION

In the past decade, there has been a rise in publications of studies investigating the knowledge of women about screening tests for Down syndrome and the most common chromosomal and morphological congenital defects in the fetus, particularly as there is a yearly increase in such incidents. The majority of studies point to inadequate awareness of women who are offered individual methods of screening¹⁻⁵. These authors concentrated particularly on stress, coping strategies and provision of support for pregnant women in connection with invasive screening tests⁶⁻¹⁰. More studies are discussed below.

Among Czech authors who have studied the problem of social and psychological support in pregnancy are Doudová and Calda¹¹, Peňázová¹² or Vágnerová¹³ in the nineties. More recent Czech studies concentrate more on the perinatal period^{14,15}. Tyrlik et al.¹⁶ published a study on aspects of the positive enjoyment of pregnancy.

Our aim was to map the level of knowledge of screening tests that were recommended to pregnant women by their practitioners. Additionally, we concentrated on the decision making process in women with abnormal screen-

ing test results, of older age, with family or medical predisposition, with regard to their feelings and the effects of time on these feelings about invasive examinations.

MATERIALS AND METHODS

The study was carried out at the Department of Medical Genetics, University Hospital, Hradec Kralove and in the genetic surgery of Litomyšl Hospital. Our investigation took place between September 2011 and March 2012. The study was part of a larger research that mapped the psychosocial aspects and their impact on patient compliance during prenatal examinations. We focused on the combined screening of first trimester¹⁷⁻²², biochemical screening of second trimester^{21,22}, a genetic consultation associated with atypical results of any screening tests and ultrasound examination at 20th to 22nd week²².

This study followed previous qualitative investigation which enabled us to set areas of interest. A preliminary cohort of 50 women was used to validate individual questions prior to starting this study²³. Based on the findings of the qualitative survey, a questionnaire was devised for

quantitative research. This procedure was chosen because the initial use of a qualitative approach allowed us to identify the variables that could be neglected using only a quantitative approach. The questions were created by a medical geneticist.

The questionnaires contained a combination of different types of questions. Demographical data were extracted from multiple choice questions, and open questions mapped the knowledge of women with respect to the individual tests. The level of knowledge of individual screening tests was assessed by a Likert scale.

A total of 300 respondents were reached. The return rate was 281 respondents, 7 questionnaires were inadequately completed. The process therefore used 274 questionnaires, i.e. 91.3%. The questionnaire was distributed to women before genetic consultations, who have been recommended for consideration for amniocentesis with subsequent fetal karyotyping. Although current diagnostic possibilities shift to the earlier stages of pregnancy and the number of chorionic biopsy is growing, amniocentesis is so far the most common invasive prenatal examination.

Instructions on how to fill in the questionnaires were provided by doctors and trained nurses. The study was anonymous and required the consent of informed patients' as well as the consent of legal guardians for patients who were younger than 18 years.

Women aged 35-39 years formed the largest group (37.6%), followed by women aged 30-34 years (27.3%), then women aged 25-29 years (17%). Older women (aged 40 years plus; 4.8%) and very young women (15-19 years; 3%) were least represented.

The majority of women (39.1%) completed vocational qualifications at colleges of higher education, 32.8% of women obtained an apprenticeship, 23.6% of patients completed university education and 4.4% of women completed secondary school. Nearly 70% (67.9%) of women identified themselves as people of faith/believers, 57.6% of women spent less than an hour browsing the internet, 30.3% of women spent 1-2 h per day on the internet and 12.3% of women browsed the internet for more than 2 h a day. Pregnancies were spontaneous in the majority of cases (94%), 6% of women had conceived using methods of assisted reproduction. Additionally, the majority of pregnancies were planned (80.1%). More than one quarter of all patients (27.3%) were primigravidae. Of the remaining multigravidae 33.9% were pregnant for the second time, 18.5% were pregnant for the third time, others were represented in small numbers. 14.4% of women had a history of one spontaneous abortion, 5.5% had two miscarriages and 3.7% women had miscarried 3 times or more, 6.3% of patients had a previous diagnosis of congenital anomaly of the fetus/child.

Nearly half of all patients (48.3%) were referred for a genetic consultation on the basis of an abnormal biochemical screening test in the second trimester, 14.4% on the basis of an abnormal combined first trimester screening test, 4.1% on the basis of an abnormal result of an integrated screening test, 31.7% of the patients were referred for an amniocentesis based on their older age and 1.5% of women due to a family history of medical predisposition.

We set 5 hypotheses to work with in the future:

H1: Knowing more information concerning individual tests reassures pregnant patients.

H2: Women referred for genetic consultation following an abnormal first trimester screening test result are more anxious and show more negative feelings compared to women referred on the basis of an abnormal second trimester biochemical screening test, or due to older age.

H3: There is a correlation between previous spontaneous abortion(s) and negative feelings about the amniocentesis.

H4: Women who discussed abnormal screening test results with their gynaecologist were less anxious than those seeking information on the internet.

H5: There is a dependence between the decision regarding going ahead with the amniocentesis and screening test results.

Data analysis

Descriptive statistical methods were chosen to characterise the cohort (age, sex, education etc.). The STATISTICA programme was then used to test our hypotheses, using Chi-square methods with Yates correction, t-test and Mann-Whitney. Levels of significance were set at $P < 0.05$.

RESULTS

We investigated women's knowledge of amniocentesis using open questions to which women had to provide an answer.

The majority of women (82.3%) had attended a genetic consultation during a previous pregnancy, 17.7% of women were referred for a consultation for the first time. Nearly 90% (86.7%) of patients claimed to have sufficient information about amniocentesis before their genetic consultation. Therefore, we investigated what women associate with the term amniocentesis. Responses are recorded in Table 1. Sources of information are recorded in Table 2, in which the number of responses are given (as opposed to percentages) as the question offered multiple answers.

The study also investigated emotions and their dynamics in women referred for genetic consultation. This was also multiple choice. Predominant emotions were negative; 152 women reported fear, 76 anxiety, 55 sadness, 27 hopelessness and 14 women reported anger. In contrast, only 71 women were glad to have been offered the consultation, 45 women were not surprised and 36 patients did not have any unusual feelings.

Women are informed about abnormal screening test results by their gynaecologists and they were then advised to book an appointment for a genetic consultation. In the Department of Medical Genetics at the University Hospital in Hradec Kralové, the consultation usually takes

Table 1. Possibilities/purpose of amniocentesis and karyotyping.

Response	Percentage
I don't know	31.7
Down syndrome	19.9
Down syndrome and chromosomal anomalies	14.8
Genetic anomalies	12.2
Cleft anomalies	11.8
Everything	3
Other answers (represented in small numbers)	6.6

Table 2. Source of information.

Source of information	Answers
Internet	121
Gynaecologist	115
Family	72
Friends	63
Specialised literature	43
Specialist	35

place within a week of being informed of abnormal test results. However, some women take longer. During this interim period, no change in emotions was reported by 60.9% women, there was emotional improvement in 27.7% and heightened emotional state in 11.4% of patients.

The majority of patients looked for help from their partners (178), 84 women turned to their parents, 35 to their friends, 19 to wider family and only 15 women turned to their gynaecologists. Interestingly, 69 women did not seek any help or support.

Our study also mapped reasons, which influence the decision making processes as to whether to undergo further invasive tests. Reasons for undergoing invasive tests are summarised in Table 3 and those against in Table 4.

Table 3. Reasons for undergoing invasive tests.

Reason for undergoing amniocentesis	Percentage of answers
To make sure fetus/baby is healthy	74.8
Peace of mind	9.4
Woman does not want a disabled child	6.9
Medical recommendation	5.4
Older maternal age	2.5
Family or medical predisposition	1

Table 4. Reasons against undergoing invasive tests.

Reasons against undergoing invasive tests	Percentage of answers
Risk or complications associated with the test	46.4
Low reliability of the biochemical screening test which was abnormal	17.4
Don't know, don't want to, will wait for geneticist recommendation and then make up my mind	14.5
No reason given	11.6
I want a baby as it is	8.7
Ultrasound examination will replace amniocentesis	1.4

Based on our clinical experience, we proposed that the possibilities of screening and invasive prenatal tests are often confused in the layman's view. This proposal was tested by 2 questions in the questionnaire, i.e. whether any test can substitute amniocentesis with karyotyping and what they are. Despite the fact that the questionnaires were filled by women referred on the basis of abnormal screening tests, older age, or family/medical predisposition as stated above, 57.6% of patients did not know whether amniocentesis with karyotyping can be fully replaced by another test, and 11.4% of women thought it could be fully substituted by a different test. An ultrasound examination was identified as an alternative test by 17 patients in this last group, other screening tests by 9 women, chorionic villus sampling biopsy by one patient (medic herself), and one patient did not know.

Patients' compliance to undergo tests is boosted by their confidence in the test. Table 5 summarises levels of patient confidence for the combined first trimester screening test, biochemical second trimester screening test and karyotyping following amniocentesis.

Table 5. Confidence of patients in individual screening tests (% of answers).

	Combined first trimester screening test	Biochemical second trimester screening test	Amniocentesis with karyotyping
Confident	25.6	22.0	41.3
Mostly confident	63.1	62.0	42.4
Not sure	10.5	11.4	14.9
Mostly not confident	0.8	2.3	0.7
Not confident	0	2.3	0.7

As stated above, one of the aims of this research was to test several hypotheses. We set 6 hypotheses in total and succeeded in proving some of them but failed to get

conclusive results for others. Our results were generated by the STATISTICA programme:

H1: Knowing more information concerning individual tests reassures pregnant patients.

The correlation between these parameters was investigated using correlation analysis based on Pearson's correlation coefficient. The correlation coefficient was 0.235, which is indicative of a correlation between the two variables. Despite the relatively weak strength of this correlation it is still possible to conclude that more relevant information reassures pregnant women.

H2: Women referred for genetic consultation on the basis of abnormal first trimester screening test are more anxious and show more negative feelings than women referred on the basis of abnormal biochemical screening test of the second trimester, or due to older age.

A Chi-square test was not significant. The value was 0.386.

H3: There is a correlation between previous spontaneous abortion(s) and negative feelings about amniocentesis.

A Chi-square test was significant. The value was 4.347, using $P < 0.05$ and 1 degree of freedom. The critical value was 3.841 and we therefore accept the correlation.

H4: Women who discussed abnormal screening test results with their gynaecologist were less anxious than those seeking information on the internet.

This relationship was analysed using Pearson's correlation coefficient. This was 0.750, which is indicative of a significant and relatively strong correlation between these variables. This correlation confirms that women informed by their gynaecologist have less negative feelings than those who seek help over the internet.

H5: There is a dependence between the decision with regards to going ahead with the amniocentesis and results of the screening test.

A Chi-square test was insignificant. The value was 0.664, using $P < 0.05$. There was no dependence.

DISCUSSION

The decision whether to undergo an invasive prenatal test following an abnormal screening test, or family or medical predisposition can have a significant impact on a woman's pregnancy as well as her future life and that of the whole family. To make the right decision the woman and her partner require relevant information presented in a way they can understand. A patient's gynaecologist should play a key role in conveying such information²⁴.

Studies investigating the level of women's knowledge of prenatal tests started to appear in literature in the nineties¹. However, they mostly concentrate on screening programmes. Chilaka et al.² investigated what British women

knew about Down's syndrome using questionnaires, and found that only 33% of patients responded correctly. The authors also proved that the level of awareness was influenced by the presence of a disabled child in a woman's neighbourhood/social circle. It was also interesting that despite prior information about the test, only 48% of women knew that they had undergone a screening test for Down's syndrome. In the study of Stefansdottir et al.⁴, 57% of women reported to have sufficient information to make the right decision whether to undergo a screening test for Down's syndrome. Similar conclusions were reached by Mulley and Wallace³, Comerford et al.²⁵ and Dahl et al.^{5,26}. Compared to studies related to screening programmes, there are significantly fewer published studies on patient awareness, decision making, and the need for support before amniocentesis^{6,7,10,27-30}. These studies agree on the insufficient knowledge of women regarding amniocentesis; their expectations related to this test are often unrealistic³¹. Our conclusions are in agreement with the above findings. We found inadequate responses to even basic questions about women's knowledge of the test. Although 86.7% of women claimed to have sufficient awareness about the possibilities of amniocentesis, only 34.7% of women responded correctly to factual questions about amniocentesis, 31.7% of patients could not answer at all, and 15.1% of patients thought that amniocentesis could investigate all genetic tests. These findings are supported by the fact that only 42.4% of women were provided with information by their doctors with regards to the test whilst 44.7% of women sought information on the internet, which often provides incorrect or misleading facts.

Dahl et al.⁵ showed correlation between higher level of patients' knowledge and their well-being, in contrast they did not prove a correlation between higher level of knowledge and anxiety concerning the health of the baby. Provision of adequate information about an abnormal test result can lessen the stress associated with a referral for a genetic consultation and possible subsequent amniocentesis and karyotyping. Referral for a genetic consultation was associated with negative feelings (sadness, anxiety, sorrow, hopelessness) in 56.1% of respondents. These negative feelings improved in only 27.7% of these patients in the period between finding out an abnormal result and genetic consultation, whilst there was no change of feelings in 60.9% of patients, and increased distress and worsening of feelings in 11.4% of women. Women with a previous spontaneous miscarriage showed more negative feelings about amniocentesis testing. Other studies^{32,33} also confirmed increased levels of stress and anxiety during this period. Tercyak et al.⁶ showed the level of anxiety was highest just before the consultation. Previous experience with prenatal diagnostic testing, increased the perceived risk of a birth anomaly, and favourable attitudes towards abortion were independently associated with increased pre-counselling anxiety. Kowalczyk et al.⁷ described depressive mood of women before chorionic biopsy, amniocentesis or ultrasound tests. Brajenović-Milić et al.¹⁰ reported that women's stress was significantly different according to the indication for amniocentesis, and women

Questionnaire distributed before the genetic consultation

Dear patient,

Here we present you with a questionnaire related to the examination that will follow. Answering the questions will take you only a couple of minutes. The results will be used to improve the quality of the consultation and for processing a Dissertation Thesis. The questionnaire is anonymous and all the data obtained will be used only for the above stated purposes. At the choice-questions, please, tick off your answer. For the multi-choice questions, you are free to choose more answers. Thank you for filling it in!

1. **Your age.....**
 2. **Your highest education level achieved:**
 - Elementary
 - Apprenticed
 - High School with a Final School-Leaving Exam
 - University degree
 3. **Your current occupation** (if you are on maternity leave, state your previous occupation)
.....
 4. **Your family status:**
 - Single, living with a partner
 - Single, living alone or at parents' house
 - Married
 - Divorced
 5. **You live in a settlement of:**
 - up to 5 000 inhabitants
 - 5 000–10 000 inhabitants
 - 10 000–50 000 inhabitants
 - 50 000–100 000 inhabitants
 - 100 000 and more
 6. **Are you religious?** Yes No
 7. **How many hours a day do you spend on the internet?**
 - Less than an hour
 - 1–2 hrs
 - 2 hrs and more
 8. **Is this:**
 - A spontaneous pregnancy?
 - An *In vitro* fertilization?
 9. **And is this pregnancy**
 - Planned
 - Unintended
 10. **How many time have you already been pregnant?** Please, write down the number of spontaneous miscarriages, extrauterine pregnancies, abortions and also inborn errors of development if any at your babies have been diagnosed.
This pregnancy is No
Number of children you have
Number of previous spontaneous miscarriages
Number of demanded abortions
Inborn errors of development of your baby/fetus in your previous pregnancies
 11. **Were there any inborn errors of development in your family relatives'?**
Yes No
 12. **If so, please, write down what kind of error was it and who had it (e.g. niece, sister, cousin from your father's side...)**
.....
 13. **Why have you been recommended for genetic counselling?**
Tick the correct answer.
 - The screening results of the First trimester (blood taking and ultrasound check between the weeks 11–14)
 - The results of the blood taking in the 16th week (called Triple Test)
 - The combination of both these screenings
 - Pregnancy at an older age
 - Your personal or family medical history (including your partner's), state what precisely.
 14. **Do you know why we have to draw out the amniotic fluid (the amniocentesis procedure)?**
 - Yes
 - To some extent
 - I do not know
 - A little
 - No
 15. **What kind of diseases are revealed by amniocentesis?**
 16. **Have you already gone through amniocentesis?** Yes No
 17. **If you are well informed, where have you got the information from?** (more answers are possible to be ticked off)
 - The doctor/ gynecologist you currently visit
 - The doctor you have visited for a special examination (e.g. the First trimester screening)
 - Family or female-relatives who have already gone through this procedure
 - Female friends, acquaintance
 - Internet
 - Specialized literature
 - Other sources, name where exactly:
 18. **What did you feel when you were recommended for genetic counselling for of the above reasons?**
You can tick more than one response.
 - Anger
 - Fear
 - Anxiety
 - Sadness
 - Despair
 - Nothing special, You just took it as a recommendation for another examination
 - You were not surprised, You expected this
 - You were glad to have as many examinations as possible to be sure your baby is healthy
 - Other feelings and thoughts, write down
 19. **If you experienced a feeling of fear, anxiety or another negative feelings during the time of waiting for the genetical consultancy, these feelings have:**
 - Improved
 - Got worse
 - Remained the same
 20. **Have you been looking for anyone's support?**
 - Partner
 - Parents
 - Wider family
 - Doctor
 - Female friend(s)
 - I haven't been looking for any support
 - Other, write down
 21. **Have you decided to go through the amniotic fluid drawing (amniocentesis)?**
Yes No I don't know
 22. **If you do, what is the reason for your decision?**
 23. **If you do not, what is the reason for your decision?**
 24. **Do You think the examination of the amniotic fluid can be substituted by another examination?**
Yes No I don't know
 25. **If so, write down what type the substitutive check-up do you mean:**
 26. **Do You have confidence in the results of amniocentesis?**
 - I definitely trust it
 - I somewhat trust it
 - I'm not sure
 - I somewhat do not trust it
 - I definitely do not trust it
- The question No. 27 will be answered only by the women who have been recommended the genetic consultancy based on the results of the screening programmes (First and Second Trimester)**
27. **How much do you trust the results of the screening (blood taking and ultrasound)?**
 - I definitely trust it
 - I somewhat trust it
 - I'm not sure
 - I somewhat do not trust it
 - I definitely do not trust it

Thank You for filling in the questionnaire. Please, hand it in to a nurse or a doctor at the Dept.of Medical Genetics where your consultancy will take place.

who underwent amniocentesis because of chromosomal aberration in previous pregnancy reported the highest stress score. Our findings were relatively surprising. We found no statistical differences between the feelings of women with abnormal first trimester screening test vs. those with abnormal second trimester screening test. This is despite the fact that the biochemical screening test of the second trimester is only 50-70% sensitive (5% false positivity), whilst the combined first trimester screening test is 95% sensitive²¹. This finding can be explained by the insufficient level of our patients' knowledge. Due to small numbers available (2 patients), we were unable to compare these findings with those in patients with proven chromosomal aberrations in previous pregnancies.

Leithner et al.⁷, Hamilton et al.⁸, Rudnicki et al.³⁴ and Borchering et al.³⁵ studied coping strategies of patients following an abnormal screening test result. Durand et al.²⁹ pointed at the important supporting role of partners and doctors in the decision making process whether to undergo amniocentesis. In our cohort, 178 of 271 women sought support from their partner, 84 from their parents and only 15 from their doctors, whilst 69 patients did not find any support. These findings are supported by the fact that both partners experience stress events differently during pregnancy^{13,29}.

We also investigated the level of a patients' confidence in individual screening tests and also in fetal karyotyping test. Our results showed that patients were significantly more confident about the combined first trimester screening test results. It was relatively surprising to find that the level of confidence in the combined first trimester screening was no different to that in the karyotyping test. We did not find any similar findings in the literature but our results could be a reflection in the low level of knowledge of women concerning individual tests. We also showed no statistical differences in the level of negative feelings between women who are referred following an abnormal first trimester versus second trimester screening test or on the basis of older age.

CONCLUSION

Our study showed that compared to international studies, Czech women show an insufficient level of knowledge in regards to pregnancy screening tests. Women confuse the possibilities of screening, invasive and ultrasound tests. Their expectations on invasive screening tests such as amniocentesis and subsequent karyotyping are unfounded. It is also important to note that doctors who take care of patients during pregnancy do not fulfil a sufficient role during stress situations. Referral for invasive tests during pregnancy is associated with largely negative feelings such as fear, anxiety, anger and hopelessness. Women's distress can be reduced if the information about indicated genetic consultation is conveyed correctly and can also help them make the right informed decision about their future course of action.

Practice implication

Our main recommendation, which is based on the findings of this study, would be to increase the level of women's knowledge of individual pregnancy screening tests. In addition to verbal communication, written information material and booklets would also be appropriate as patients and their partners can refer to them again. Gynaecologists should play a key role in this process.

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