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How women deal with the results of serum screening for Down syndrome in the second trimester of pregnancy

Martin J. N. Weinans^{1*}, Annemarie M. G. Huijssoon¹, Tjeerd Tymstra², Mignon C. F. Gerrits¹, Johan R. Beekhuis¹ and Albert Mantingh¹

¹*Antenatal Diagnosis Unit, Department of Obstetrics and Gynaecology, University Hospital Groningen, The Netherlands*

²*Department of Medical Sociology, Faculty of Medical Sciences, University of Groningen, The Netherlands*

To gain insight into how pregnant women experience serum screening for Down syndrome, we sent questionnaires to two groups of relevant subjects in the north of the Netherlands. The questionnaires addressed the following issues: decision-making process, knowledge and opinions. Questionnaire A was sent to women of 36 years of age and older ($n=99$) (group A) who were all 20 to 36 weeks pregnant at that time. In the Netherlands prenatal diagnosis is routinely available to these women. Questionnaire B was sent to women of younger than 36 years ($n=69$) (group B) who had received a screen-positive result and had subsequently undergone amniocentesis. About half of these women were still pregnant at that time. For these women, serum screening is only available on the basis of opting-in. The two questionnaires were largely identical. The response rates to questionnaires A and B were 82% and 91%, respectively. Group A (women of 36 years and older) considered that second trimester serum screening made a welcome contribution to the decision-making process about whether to undergo amniocentesis. Moreover, it reduced the amniocentesis rate considerably. The vast majority said they would apply for serum screening in a following pregnancy, but favoured the idea of first trimester screening. In group B (women of younger than 36 years), reassurance was the most commonly mentioned reason for undergoing serum screening. Almost all the women experienced some degree of anxiety when they were informed about the screen-positive result and 13% continued to be anxious, even after the favourable result of the amniocentesis. The majority of the respondents would also apply for serum screening in a following pregnancy and were of the opinion that this screening should be offered to all pregnant women in the Netherlands. Copyright © 2000 John Wiley & Sons, Ltd.

KEY WORDS: prenatal diagnosis; Down syndrome; maternal serum screening; decision making

INTRODUCTION

Prenatal screening for Down syndrome has been in practice for the past three decades. Until the mid-1980s, this screening was only offered to women on the basis of maternal age and previous history. The detection rate based on maternal age is about 25–30% for a false-positive rate of 5%, depending on the age distribution of the population. In 1988, a screening test for Down syndrome, using a combination of three biochemical markers (alpha fetoprotein—AFP, human chorionic gonadotrophin—hCG and unconjugated oestriol—uE₃) and maternal age, predicted a detection rate of over 60% with a false-positive rate of 5% (Wald *et al.*, 1988). This is more than double the detection rate of screening on the basis of maternal age alone. Since then, various authors have clearly demonstrated the efficacy of many different screening programmes (Wald *et al.*, 1992; Milunsky, 1992; Haddow and Palomaki, 1993). At present, biochemical screening for Down syndrome is well established in many countries.

When assessing the performance of maternal serum screening for Down syndrome in the Netherlands, the way in which the obstetrical care system is organized

has to be taken into account. It is strongly decentralized and subdivided into first- and second-level care. Maternal serum screening for Down syndrome has been available at our centre in the north of the Netherlands since 1990; it was evaluated by Beekhuis in 1993. A few thousand serum samples per year are analysed at our centre; about 60% are sent by midwives and general practitioners (first-level care), 30% are sent by other hospitals and 10% are from our own department. Owing to Dutch regulations (The *Population Screening Act* of 1992), serum screening is not offered routinely to all pregnant women. Prenatal diagnosis does not form part of routine care for pregnant women younger than 36 years. For these women serum screening is only available on the basis of opting-in, whereas women of 36 years of age and older participate on the basis of opting-out. At our centre, the result of serum screening in women of 36 years of age and older is given as a numeric value, whereas in women of younger than 36 years, the result is given as 'screen positive' or 'screen negative'.

Various studies have focused on women's experience of serum screening programmes for Down syndrome, including their opinions, motives, knowledge and decision-making processes (Marteau *et al.*, 1992; Roelofsen *et al.*, 1993; Santalahti *et al.*, 1998; Gekas *et al.*, 1999; Michie *et al.*, 1999). Nevertheless, certain questions have remained unanswered. The aim of this study on women who underwent serum screening was

*Correspondence to: M. J. N. Weinans, Department of Obstetrics and Gynaecology, University Hospital, P.O. Box 30.001, 9700 RB Groningen, The Netherlands. E-mail: m.j.n.weinans@og.azg.nl

to gain insight into the following issues: How much do they understand? Are they able to recall the information they received? What is their actual experience of this screening? What decisions do they make and what influences these decisions? What is the impact of the screening result on the further course of pregnancy?

SUBJECTS AND METHODS

Two semi-structured questionnaires were developed by interviewing 20 women in their homes about maternal serum screening for Down syndrome. Questionnaire A was sent to a sample of pregnant women of 36 years of age and older. Questionnaire B was sent to women of younger than 36 years who had undergone second trimester serum screening and were screen-positive. We chose 36 years as the cut-off point, because Dutch regulations restrict routine prenatal diagnosis to women of 36 years and older.

Questionnaire A

The first questionnaire was sent to 99 women of 36 years of age and older who had undergone maternal serum screening at around 16 weeks of gestation. Serum samples were sent by midwives, general practitioners, other hospitals and some were from our own department. Risk assessment was done at our centre. All these women were 20–36 weeks pregnant when they filled in and returned the questionnaire.

Questionnaire B

The second questionnaire was sent to 69 women younger than 36 years who had undergone amniocentesis at the University Hospital Groningen in the preceding six months because of a positive serum screening result for Down syndrome. None of these amniocenteses showed a chromosomal abnormality. Of these 44% women were still pregnant when they filled in and returned the questionnaire, while 56% had already delivered.

The two questionnaires were largely identical. Questionnaire B contained more questions about amniocentesis, whereas questionnaire A focused more on decision making after the result of serum screening. We invited the women to write down their personal experience of prenatal screening at the end of the questionnaire.

RESULTS

Questionnaire A (women of 36 years and older)

We received 81 correctly completed questionnaires (response rate 82%). It appeared that half of the respondents (52%) had not known about the existence of serum screening before their present pregnancy. Table 1 (left column) shows how they found out about

Table 1—How did you find out about the existence of serum screening? (You can choose more than one)

	≥ 36 years	< 36 years
General practitioner	12%	17%
Gynaecologist	62%	19%
Midwife	7%	57%
Relatives, acquaintances	15%	19%
Magazines, television	21%	13%
Other	15%	10%

the existence of serum screening. Gynaecologists played the most important role in this. Other questions about the information process demonstrated that only 7% of the respondents did not feel wellinformed about the serum screening, while 24% said that the serum screening result would tell them whether they were carrying a child with Down syndrome.

If serum screening had not existed, almost half of the respondents (47%) would have undergone amniocentesis, while the other half would have declined. A full 50% of the women had decided beforehand at which specific risk estimate they would have amniocentesis; 70% of them chose the risk level based on their age. Only 12% of the respondents could not recall the result of their serum screening in numbers. In one third of the respondents, the risk according to serum screening was higher than that based solely on their age. Almost half of the women (49%) had received the advice from the general practitioner gynaecologist or midwife, to have or to decline amniocentesis when they were informed about the serum screening result (Table 2). Eighty-two per cent of the respondents stated that the serum screening had helped them to make a well-founded decision about whether to have amniocentesis or not.

Twenty-one per cent of the respondents had a positive serum screening result (Down syndrome risk of 1 in 250, or higher); 76% decided to have amniocentesis. None of these amniocenteses showed a chromosomal abnormality. Seventy-nine per cent of the respondents had a negative serum screening result; they all therefore declined amniocentesis. The amniocentesis rate for the total group was 16%. In a subsequent pregnancy almost all the respondents (93%) said that they would apply for serum screening. At the end of the questionnaire, we asked whether it would be considered an advantage to have serum screening performed in the first trimester of pregnancy. The vast majority of the respondents (82%) thought that this would be an advantage. The most commonly mentioned reasons for preferring first trimester screen-

Table 2—Did you receive any advice to have or to decline amniocentesis from your general practitioner/gynaecologist/midwife when you were informed about the serum screening result?

No, I had to make a decision myself	51%
Yes, I was advised to have an amniocentesis	15%
Yes, I was advised to decline an amniocentesis	34%

ing were the shorter period of uncertainty and the possibility of first trimester termination of pregnancy before any fetal movements could be felt.

Questionnaire B (women younger than 36 years)

A total of 63 out of the 69 women responded (response rate 91%). Table 1 (right column) shows how they had found out about the existence of serum screening. It appears that the midwives played an important role. Only 6% of the women felt that they had not been wellinformed about the serum screening, whereas 29% of respondents said that the serum screening result would tell them whether they were carrying a child with Down syndrome.

We asked the women what their reasons were for wanting serum screening. The most commonly mentioned reason was reassurance regarding fetal health (71%), while 10% answered that they would undergo any prenatal test available. As one woman stated: 'This kind of screening has different sides. It is there, you can use it and it is difficult not to use because the procedure is so simple. But imagine that something is wrong and you didn't have this test. During my previous pregnancies the test was not available, making things a lot easier because I didn't have to decide.' Almost all the women (92%) experienced anxiety after receiving the positive result of serum screening. We asked the women whether it had been difficult to decide to have amniocentesis. Forty-one per cent of the respondents had found it difficult to decide to have amniocentesis, ranging from slightly to very difficult.

All the women in group B underwent amniocentesis. The outcome was favourable in all of them. We asked whether this favourable result had enabled them to feel more confident about their pregnancy. Thirteen per cent of them had continued to experience anxiety despite the normal amniocentesis result. Only 5% of the respondents regretted undergoing serum screening and 17% would not apply for serum screening in a subsequent pregnancy. Forty-three per cent of the women concluded that pregnancy became a more enjoyable experience thanks to these forms of prenatal diagnosis. Only 15% of the respondents disagreed with the statement that serum screening should be offered to all pregnant women in the Netherlands, regardless of their age.

DISCUSSION

The aim of this study was to gain insight into the user's opinions about maternal serum screening for Down syndrome in the Netherlands. Two distinctive groups of pregnant women were approached: women of 36 years of age and older (to whom this screening is routinely offered) and younger women (for whom serum screening is only available on the basis of opting-in).

It is well known that serum screening in women of

36 years of age and older has a positive effect on the amniocentesis rate (and the associated fetal loss) and that it also facilitates decision making about amniocentesis (Haddow *et al.*, 1994). In our study, the most important reason for amniocentesis was not only a positive serum screening result (Down syndrome risk of 1 in 250, or higher) but, in particular, a higher risk according to serum screening than a risk based solely on age. On the other hand, all women with a negative serum screening result decided to decline amniocentesis. The amniocentesis rate in our study diminished from 47% to 16%, a reduction of 66%.

Although the positive effects of serum screening predominate in the group of women aged 36 years and older, the negative effects are more pronounced in the group of younger women with a screen-positive result. The burden of serum screening is mainly psychosocial and emotional. Previous studies have shown that women with a positive serum-screening result experience anxiety while awaiting the result of subsequent amniocentesis (Marteau *et al.*, 1992). This was confirmed in our study. The majority of these women felt much more confident about their pregnancy after hearing the favourable result of amniocentesis. Despite a few weeks of distress, the majority of these women felt positively about serum screening in retrospect. They were of the opinion that serum screening should be offered to all pregnant women, regardless of their age. It is disturbing that in our study, 13% of women younger than 36 years who had a positive serum screening result continued to experience some degree of anxiety despite the normal amniocentesis result. Although our semi-structured questionnaire did not allow precise quantification of anxiety levels, this sustained anxiety is in line with the study of Marteau *et al.* (1992).

As we did not send questionnaires to women of younger than 36 years who had a true negative serum screening result, we can only speculate about their opinions. It seems likely that these women will have a positive attitude towards serum screening, because reassurance is the most commonly mentioned reason for having this screening test (Santalahti *et al.*, 1998). It would also be interesting to obtain information about the opinions of women with a false negative serum screening result.

Providing a sufficient level of counselling about serum screening and possible subsequent amniocentesis continues to be a challenging issue. What information does a woman or couple need in order to make well-founded decisions? In our study almost all women felt that they had been well-informed about the serum screening. Nonetheless, about one quarter of them supposed that the serum screening result would tell them whether or not they were carrying a Down syndrome child. This demonstrates lack of knowledge. Similar observations have been reported in previous studies (Goel *et al.*, 1996; Gekas *et al.*, 1999). Accurate information provided by health professionals could possibly raise knowledge levels. But, is it possible to reduce the problem of the anxiety that women experience when they receive a screen-positive result

merely by improving the counselling process? We agree with Santalathi *et al.* (1998) that this is unlikely because the reason for participating in serum screening is not to find out whether they belong to a risk group, but to obtain reassurance about the health of the fetus. We do not think that changing the current method of counselling would make the drawbacks of serum screening disappear.

The finding of Kornman *et al.* (1997) that women in our region favour first trimester screening over second trimester screening, was confirmed in this study. The most commonly mentioned reasons for preferring first trimester screening were the shorter period of uncertainty and the possibility of first trimester termination of pregnancy before any fetal movements could be felt.

In the Netherlands, the *Population Screening Act* prohibits offering routine serum screening to women of younger than 36 years. We think that this policy is questionable: Does it show respect for women's autonomy? Earlier studies have stressed that the autonomy principle of the individual should be considered a basic right, as expressed by women when asked their opinions about screening (Tymstra *et al.*, 1991). The majority of the women younger than 36 years in our study (they all had a screen-positive result) confirmed this by indicating that serum screening should be offered to all pregnant women in the Netherlands, regardless of their age. We agree with them: it is improper to exclude women younger than 36 years from serum screening.

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