

## ELECTRONIC LETTER

# Preconceptional couple screening for cystic fibrosis carrier status: couples prefer full disclosure of test results

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Carrier screening programmes, aimed at the identification of carrier couples in the general population, provide these couples with the opportunity to be informed about their risk and about the reproductive options that are available. It has been argued that the imperfect sensitivity of the carrier test, for example in the case of cystic fibrosis (CF), might cause anxiety in couples if a mutation is detected in one of the partners, while the other tests negative, but may still carry a rare mutation (in future referred to as +/- couples).<sup>1</sup> Such couples have a residual risk and may be distressed by the lack of clear options, such as prenatal diagnosis.

Wald<sup>2</sup> therefore proposed a couple based screening method, where both partners provide samples simultaneously. Only those couples in which both partners are found to be carriers are informed about their carrier status, while all others are told that they have no marked increased risk, circumventing the possible anxiety in a +/- couple. To a certain extent, Wald's proposal can be considered favourable. Simultaneous sampling prevents the anxiety and need for counselling that might arise among those who are tested positive while awaiting the partner's results, which can occur in stepwise screening.<sup>3-5</sup> In addition, sampling one partner later requires extra effort. A major disadvantage of couple screening without full disclosure of the test results is that information is withheld from identified carriers who might need it at a later date with a new partner or whose relatives might wish to be tested. Moreover, Wald's procedure can be considered as directive counselling, because the patient is not given the full information on which choices can be based.<sup>5</sup> The advantages and disadvantages of disclosure and non-disclosure of +/- results have generated much debate. Studies have reported anxiety and false reassurance among participants in both approaches. Some results support the non-disclosure strategy proposed by Wald,<sup>6-9</sup> although it remains controversial.<sup>3 10 11</sup> To investigate what the couples themselves prefer with regard to the disclosure of test results, couples participating in a preconceptional screening programme were offered the opportunity to choose whether they wished to be fully informed about the test results (including disclosure of +/-) or only informed about whether or not they had a 1 in 4 risk as a couple.

## METHODS

The sample consisted of 108 couples planning a pregnancy and participating in preconceptional CF carrier screening. In short, the couples responded to a letter of invitation from their general practitioner (n=5) offering screening to all subjects (aged 20-35 years) and their partners between May 1997 and November 1999. The letter invited couples to attend a 45 minute educational session organised by one of the authors (LH) on two evenings in one week. The response rate was approximately 10% of the eligible target population (couples planning to have children) (data not shown). Detailed information on the screening procedure and calculation of the response has been described in a previous paper.<sup>12</sup> During the educational session, the couples were provided with detailed

information about the clinical and genetic aspects of CF, and the implications and options for couples with positive and +/- test results. Furthermore, the couples were informed about the method of testing, including the imperfect sensitivity of the test (the test sensitivity in this study was approximately 87%). The verbal information was backed up with a 15 page booklet. At the end of the educational session, both partners of the participating couples provided mouthwashes simultaneously, and they were given a consent form to sign at home. Mutation analysis was performed only for couples who had returned their signed consent form by mail. The method of testing was stepwise, that is, the sample of one partner was tested first, and the other sample was only tested if the first one tested positive. An additional section of information in the brochure explained three ways in which the test results could be received, and the couples could indicate their choice on the consent form: (A) "We will be informed whether or not we are both carriers", (B) "We will be informed whether or not we are both carriers, or whether or not the first partner tested is a carrier", (C) "We have no preference for either A or B, the choice will be made at random". The booklet and a kit with which the mouthwash sample was to be collected were sent to the home address of couples who were not able to attend the session, but were interested in the screening. All couples completed sociodemographic assessment questionnaires, in which they were also asked to describe in their own words the motivation for their choice with regard to the way in which they wished to receive their test results.

## RESULTS AND DISCUSSION

Of the 108 couples, 41% were married. The mean age of the men was 30.8 years (range 20-45) and the mean age of the women was 28.6 years (range 20-44). The level of education was high for 37% of the couples (higher vocational training, university), intermediate for 49% (higher level of secondary school, intermediate vocational training), and low for 14% (primary school, lower level of secondary school, lower vocational training). There was no difference in socio-demographic characteristics between the eligible couples who participated in screening and those who did not, as was shown in a previous study comparing 76 of these participating couples and 53 non-participating couples.<sup>12</sup> This in contrast with the findings of others, showing, for example, that those with high levels of education were more likely have the test.<sup>13 14</sup>

In total, 87 couples (81%) attended the educational session and 21 couples (19%) received the information at home. Table 1 shows that most couples (94%) chose to be fully informed about the test results, including a +/- result (B). Four couples reported that they only wanted to know whether both partners were carriers (A), and two couples had no preference for the way in which they would be informed (C).

Half of the couples (n=51) who chose to be fully informed (B) explained their choice by stating that they did not want any information to be withheld from them. Other reasons

**Table 1** Choices of couples with regard to the communication of cystic fibrosis carrier test results

Choice	Total (%)
(A) Informed whether or not both are carriers	4 (4)
(B) Informed whether or not both are carriers, or whether or not the first partner tested is a carrier	102 (94)
(C) No preference for A or B	2 (2)

reported were that they wanted to be able to inform other family members (25%), or that they wanted to know for their own sake (13%). Five couples argued that the information could be used in a new relationship and seven couples gave no reason for their choice. Reasons why couples did not wish to know +/- results (A) were that they perceived the residual risk (1/870) of having a child with CF as low, and that it would not influence their decision to have children (two couples). One couple stated that knowing +/- results would cause anxiety, because no options are provided to reduce the uncertainty. One couple gave no reason for their choice. Of the four couples choosing (A), one couple did not attend the educational session.

The results of this Dutch study are in line with the findings of Miedzybrodzka *et al*<sup>15</sup> in the United Kingdom. Out of a total of 450 pregnant woman who were asked which screening method they preferred, if screening were available, 62% preferred stepwise screening with full disclosure of the test results, 26% preferred couple screening with non-disclosure, and 12% had no preference.<sup>15</sup> However, differences with other screening programmes and other countries will exist, so the question remains whether the attitudes of participants in screening to the disclosure of test results in other European populations will be similar.

In the present study, 42 subjects (20%) spontaneously stated that they would prefer to have results of both partners, suggesting that they wanted simultaneous testing of samples in addition to full disclosure of the test results. Since most couples (94%) wanted to be fully informed about the test results, it was not possible to compare demographic, educational, or outcome variables, such as satisfaction or understanding of the test results, between couples receiving the results according to methods A, B, or C.

The results of a randomised controlled trial evaluated by Marteau *et al*<sup>8</sup> showed that three years after prenatal testing, women who had undergone couple sampling, one partner tested in the first instance, and non-disclosure of the test results (the same as our method A) were 4.5 times more likely than those who were sampled and tested stepwise to recall accurately that the test results meant that it was unlikely that they were both carriers. It was suggested that the time and effort spent by staff on explaining the meaning of couple results might have caused the differences found between the two groups. In addition, approaching couples may have resulted in more communication between the partners in couple screening than in stepwise screening, in which, in most cases, only the women had been approached.<sup>8</sup>

Based on the findings of the present study and the results of others, simultaneous sampling of partners and full disclosure of the test results is recommended for CF carrier screening. Recently, the American College of Medical Genetics Subcommittee on Cystic Fibrosis Screening recommended either couple based screening with full disclosure of test results or stepwise screening, leaving this decision up to the judgement of the practitioner.<sup>16</sup> The results of this study show that some couples prefer simultaneous testing of partners, although this implies that the costs of testing will be doubled, and twice as many +/- couples will be identified, with no further options.

- Couple based carrier screening, in which both partners provide samples and a positive result is given only if both partners are found to be carriers, has generated much debate. The objective of this study was to find out how much couples wish to know about their screening test results.
- In total, 108 couples participating in cystic fibrosis (CF) carrier screening were offered the opportunity to choose whether they wished to be fully informed about the test results or whether they only wanted to know whether or not they had a 1 in 4 risk. The advantage of the latter choice is the avoidance of possible anxiety about a residual risk in those couples in which one partner is found to be a carrier and the other is not.
- Questionnaires were used to assess the reasons for the choice. Most couples (94%) wished to be fully informed, and the main reason was that they did not want any information to be withheld from them (50%). Other reasons given were to inform relatives (25%), for their own sake (13%), and for use in case of a new partner (5%). The results show that the great majority of couples want to be fully informed about their test results, including the results if only one partner tested positive and the other partner negative.
- These findings may be useful in the development of future carrier screening programmes.

Whatever method of screening is used, attention should be given to the pre-test and post-test counselling of couples, to ensure an informed choice and maximum understanding of the test results and to minimise anxiety.

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