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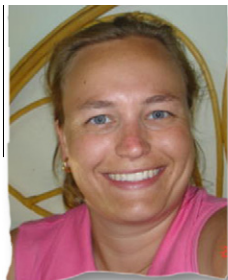
Knowledge and perceived risks in couples undergoing genetic testing after recurrent miscarriage or for poor semen quality

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Abstract Couples with recurrent miscarriage (RM) and men with poor semen quality may undergo genetic testing as part of the diagnostic work-up. This study explored their knowledge and perception of genetic testing, evaluated psychological wellbeing and identified associated variables. A prospective questionnaire study was conducted in seven clinical genetics centres and referring gynaecological departments in couples with RM or poor semen quality. Questionnaires were completed before disclosure of genetic test results. Main outcome measures were knowledge, perceived risk, anxiety and depression. Of 439 participants, 256 were not aware genetic testing was part of the diagnostic work-up. One-third (36% RM, 33% poor semen quality) indicated they had not received information about the genetic test from their doctor. Perceived risk of receiving an abnormal genetic test result was higher than objective risk. Anxiety was highly correlated with perceived risk. Women with RM were more anxious than women in the poor semen quality group or men ($P < 0.01$). These couples undergoing genetic testing have a suboptimal understanding of the nature of testing, overestimate the risks of receiving an abnormal result and some show high levels of anxiety. The results of this study can be used to improve patient counselling before genetic testing. 

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KEYWORDS: anxiety, chromosome analysis, depression, perceived risk, poor semen quality, recurrent miscarriage

Introduction

In reproductive medicine, couples with recurrent miscarriage (RM) and men with poor semen quality may undergo genetic testing as part of the diagnostic work-up. In couples with RM one of the partners may carry a balanced structural chromosome abnormality and their offspring is therefore at risk of unbalanced structural chromosome abnormalities leading to severe mental retardation and/or congenital malformations. The prevalence of a parental structural chromosome abnormality is estimated between 3% and 6% (de Braekeleer and Dao, 1990; Franssen et al., 2005; Hook et al., 1989). Clinical guidelines recommend parental karyotyping in couples with RM and subsequent invasive prenatal diagnosis when a chromosomal abnormality is found (ACOG, 2002; Jouniaux et al., 2006; NVOG, 2007; RCOG, 2003). Genetic testing in couples with RM is generally offered after two or three miscarriages.

Genetic testing in men with poor semen quality is generally performed if the total sperm count is <1 million spermatozoa per ejaculate. Men undergo karyotyping and are evaluated for deletions on the Y-chromosome before starting intracytoplasmic sperm injection (ICSI) treatment. A genetic cause is found in 10–15% of cases (Ferlin et al., 2006). Men with a chromosomal abnormality have a higher risk of aneuploid offspring after ICSI treatment. In case of Y-deletions, all sons conceived by ICSI will inherit the same deletion with possible consequences for their reproductive chances (Aittomaki et al., 2004; Silber and Repping, 2002).

As with any other medical intervention, patients should be properly informed before undergoing genetic testing. It is the duty of healthcare professionals to involve and support patients in rational decision making, since patients have the legal and ethical right to know what will happen to them. The information provided should at least cover the nature of the procedure and its consequences (UNESCO, 2006; World Medical Association (WMA), *Medical Ethics Manual*, 2009a). Compared with other medical interventions, genetic testing may pose additional challenges to physicians and patients. The principles of genetics and of genetic testing may, in general, be more difficult to convey (WMA, *Statement on Genetics and Medicine*, 2009b). The association between risk perception and other psychological variables are key issues for research on genetic counselling.

This study evaluated participants' understanding of and perceptions about genetic testing. It also assessed to what extent their knowledge, or the lack thereof, and perceived risks influenced the levels of anxiety and depression. It postulated that, even though couples with RM or with poor semen quality have different underlying mechanisms for their reproductive problems, both undergo genetic testing and might feel the same uncertainty and psychological distress before disclosure. The main aim of this study was to examine the consequences of genetic testing before disclosure; therefore both groups were studied concurrently.

Materials and methods

Study population and procedure

Couples with RM in whom parental karyotyping in both partners was performed or couples with poor semen quality in whom kar-

yotyping and DNA analysis in the male partner was performed were eligible for inclusion. RM was defined as two or more, not necessarily consecutive, miscarriages before 20 weeks of gestation. Poor semen quality was defined as a total sperm count less than 1 million spermatozoa per ejaculate.

Couples were identified between January 2006 and June 2009 in the clinical genetics laboratories of seven academic hospitals in the Netherlands. Only couples still unaware of their genetic test result were included. Exclusion criteria were pregnancy (gestational age > 12 weeks) at time of disclosure of the genetic test result, an already identified familial genetic abnormality, a serious or life-threatening disease or insufficient language proficiency. After being identified, the referring gynaecologist or urologist was contacted by the central researcher and was asked for consent to approach the couple and to determine eligibility.

Both partners of eligible couples were sent an identical questionnaire including a letter explaining the purpose of the study and asking for written informed consent. Every couple was assigned with a unique study number, identifying both partners as a couple. Following the standardized study protocol, 3–5 days after receiving the questionnaire the couples were contacted by telephone to provide additional information about the study. Both partners of the couple were asked to complete the questionnaire separately. A stamped return envelope was included.

Most couples had blood taken for the genetic test 6–8 weeks before being contacted and a scheduled appointment for disclosure of the genetic test result 1–3 weeks after receiving the questionnaire. All invited couples were unaware of the results of genetic testing at the moment of returning the questionnaire.

Before study initiation, the Institutional Review Board (IRB) of the Academic Medical Centre in Amsterdam had indicated that IRB approval was not needed for this study, according to the legal requirements in the Netherlands.

Pre-test counselling

In the Netherlands, pre-test counselling in couples with RM or poor semen quality is provided by the gynaecologist or urologist who requests the test. The healthcare professional is supposed to inform the couples and to mention advantages and disadvantages. In case a genetic abnormality is found, the physician ordering the test is supposed to discuss this result with the patients and to refer them to a clinical geneticist for further counselling and information.

Study instrument

A pilot study was conducted among 10 couples (20 participants) to evaluate clarity, ease of reading and feasibility of the questionnaire. The final questionnaire consisted of multiple-choice and open-ended questions and included two validated standardized psychological measurements (state-trait anxiety inventory (STAI), beck depression inventory (BDI-II)).

Socio-demographics and obstetric history

Participants were asked about their religion, education, occupation and obstetric history.

Knowledge and awareness

Knowledge of what is particularly tested with a genetic test, awareness of the genetic test that was performed and the information received during counselling was asked by multiple-choice questions. Participants were asked to tick only one answer.

Perceived risks

Participants were asked how they perceived their risk of receiving an abnormal genetic test result, their risk of another miscarriage, a stillborn child, a child with major congenital abnormalities, their chance of conceiving at least one child and their chance of conceiving their desired number of children. Participants marked their perceived risk on visual analogue scales, ranging from 0% to 100% (Caruso et al., 2009; Lerman et al., 1995). Participants were asked to indicate their perceived risk for each possible risk scenario in contrast with the risk for the general population (lower, comparable or higher).

The questionnaire relating to patients knowledge and awareness of the genetic testing and their perceived risks is given in the Appendix (available online only).

Anxiety

Anxiety was measured using the validated Dutch version of the Spielberger state–trait anxiety inventory (STAI) (Spielberger, 1983; Van der Ploeg, 2000). The STAI consists of two subscales, the 20-item state anxiety scale and the 20-item trait anxiety scale. The STAI differentiates between the temporary condition of ‘state anxiety’ and the more general and long-standing quality of ‘trait anxiety.’ Responses are rated on a four-point scale (1–4). Sum scores on this measure range from 20 to 80, with higher scores representing higher levels of anxiety. Scores on the STAI were compared with a Dutch reference population.

Depression

Depression was measured using the validated Dutch version of the 21-item second edition of the Beck depression inventory (BDI-II) (Beck and Steer, 1987; Van der Does, 2002). Respondents are asked to rate how they felt in the last 2 weeks. Responses are rated on a four-point scale (0–3). Sum scores on this measure range from 0 to 63, with higher scores reflecting more depressive symptoms. A score of 0–9 indicates minimal depression, 10–18 indicates mild depression, 19–29 indicates moderate depression and 30–63 indicates severe depression. Scores on the BDI-II were compared with a Dutch reference population.

Statistical analysis

Statistical Package for the Social Sciences (SPSS version 15.0, USA) was used to analyse the data. Descriptive statistics were generated to describe the total sample of participants and the subsamples of participants with RM and poor semen quality in terms of socio-demographics and obstetric history. Differences were tested with t-test statistics and ANOVA for parametric data, and Mann–Whitney, Kruskal–Wallis and Chi-squared tests statistics for non-parametric data. Correlations were tested with Spearman’s rho or Pearson’s *r* where appropriate. A general linear model

was used to identify factors related to the STAI scores and to identify factors associated with the perceived risks. A logistic model was used to evaluate factors associated with the BDI-II. Various factors were included: education (categorical), religion (categorical), time trying to conceive (categorical), age (continuous), number of previous miscarriages (continuous) and number of previous pregnancies (continuous). Two-tailed *P*-values <0.05 were considered to indicate statistical significance.

Results

During the study period (2006–2009), 482 potentially eligible couples were identified in the seven participating clinical genetics laboratories (Figure 1), 198 couples with RM and 284 couples with a man with poor semen quality. Of all those couples, 41 couples were excluded, because of a language barrier (*n* = 10), current pregnancy (*n* = 5), a previous disclosed genetic test result (*n* = 10), serious illness (*n* = 4), no consent from the referring physician (*n* = 6), a familial genetic abnormality (*n* = 1) or other reasons (*n* = 5). Consequently the questionnaire was sent to 441 couples (882 potential participants). Of these subjects, 439 (222 men and 217 women) returned the questionnaire (response rate 50%), 172 had a history of RM (39%) and 267 a history of poor semen quality (61%). Almost all participants were part of an included couple; only two women in the RM group and seven men in the poor semen quality group participated without their partner.

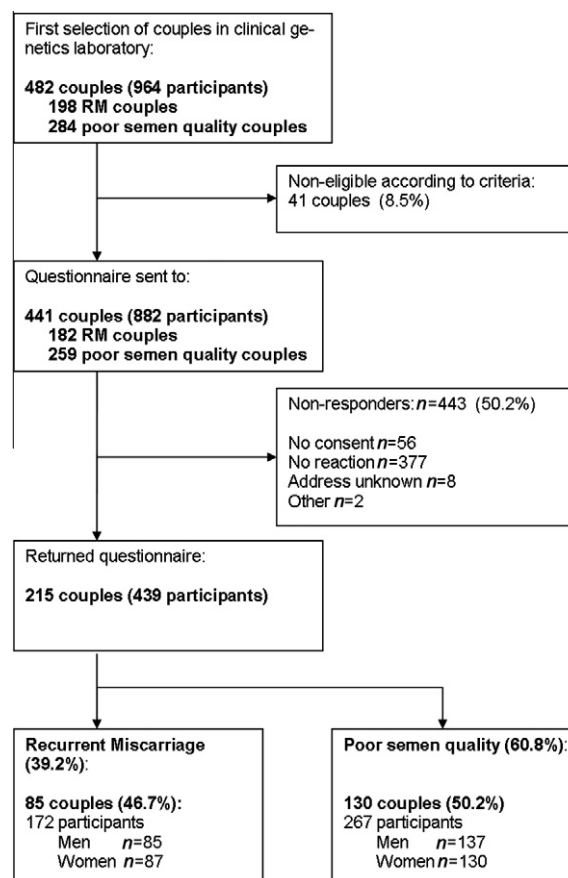


Figure 1 Flowchart summarizing inclusion and response.

The mean age of the RM group was 35.4 years versus 35.1 years in the poor semen quality group (Table 1). Most participants were of Dutch nationality, had finished secondary education and reported no religious affiliation. Information on obstetric history is presented per couple. The poor semen quality group had been trying to conceive significantly longer than the RM group ($P = 0.02$) (Table 1). On average, the RM group would have liked to have 2.3 children, slightly higher than the mean of 2.1 children in the poor semen quality group ($P = 0.01$). The RM group reported a median of three pregnancies and two miscarriages, whereas the poor semen quality group obviously recorded none.

More than half of participants stated that, before their consultation, they were not aware genetic testing was part of the diagnostic work-up of RM or poor semen quality (101 (59%) in the RM group and 155 (58%) in the poor semen

quality group; Table 2). One-third of participants stated that they did not receive any information about the genetic test from their referring doctor during the consultation (36% RM, 33% poor semen quality). Only 40% (45% poor semen quality) of the participants with RM understood the meaning of the genetic test, 39% stated they had no idea (42% poor semen quality). In the RM group, 69% of participants felt it was not possible to ask questions about the genetic test during the consultation and 12% could not remember the genetic test being discussed at all. In the group of men with poor semen quality, the percentages were 60% and 21%, respectively (Table 2). In contrast, around 60% of the participants did not feel they needed any more information during their visit (58% RM, 64% poor semen quality). There were no significant statistical differences between participants with RM or poor semen quality in knowledge or awareness. Preferences for information were examined by gender. More women

Table 1 Baseline characteristics and obstetric history of participants with recurrent miscarriage and poor semen quality ($n = 439$).

Baseline characteristics	Recurrent miscarriage ($n = 172$)	Poor semen quality ($n = 267$)
Gender		
Men	85 (49)	137 (51)
Women	87 (51)	130 (49)
Age (years)	35.4 ± 5.1	35.1 ± 6.4
Highest education		
Primary	4 (2)	14 (5)
Secondary	89 (53)	154 (58)
Higher	72 (43)	94 (35)
Other	4 (2)	3 (1)
Religious affiliation		
None	89 (55)	159 (60)
Christian	55 (34)	71 (27)
Other	18 (11)	34 (13)
Total score BDI (median (IQR)(range))	5 (0–11) (0–44)	3 (0–7) (0–41)
Total score STAI state	36.6 ± 12.2	34.6 ± 10.1
Male	33.1 ± 8.9	32.9 ± 9.2
Female	40.0 ± 13.8	36.3 ± 10.8
Total score STAI trait	36.4 ± 11.5	33.2 ± 10.3
Male	32.8 ± 9.0	32.0 ± 9.7
Female	40.0 ± 12.6	34.4 ± 10.8
Obstetric history per couple	Recurrent miscarriage ($n = 87$)	Poor semen quality ($n = 130$)
Time trying to conceive		
<6 months	1 (1)	3 (2)
6 month–1 year	17 (20)	6 (5)
1–2 year	33 (39)	55 (42)
>2 year	33 (39)	66 (51)
Unknown	3	0
All previous pregnancies (median, IQR)	3 (2–4)	0
Previous miscarriages (median, IQR, range)	2 (2–3) (1–6)	0
Previous live-born children (median, IQR)	0 (0–1)	0
Desire to have children (%), IQR)	95 (95–100)	95 (95–100)
Desired no. of children	2.34 ± 0.99	2.12 ± 0.79

Values are n (%) or mean ± standard deviation, unless otherwise stated.

BDI = beck depression inventory; IQR = interquartile range; STAI = state-trait anxiety inventory.

Table 2 Knowledge and information about the genetic test related to the fertility work-up.

Criteria	Recurrent miscarriage (n = 172)	Poor semen quality (n = 267)
Aware of standard genetic testing		
No	101 (59)	155 (58)
Yes	70 (41)	110 (42)
Information received about genetic testing		
No	61 (36)	87 (33)
Yes	110 (64)	177 (67)
What is tested? (only 1 answer possible)		
Antibodies	8 (5)	8 (3)
Clotting factors	11 (6)	1 (0)
Changes in DNA or chromosomes	68 (40)	117 (45)
Something else	17 (10)	26 (10)
Don't know	66 (39)	111 (42)
Possible to ask questions		
No	117 (69)	160 (61)
Yes	32 (19)	47 (18)
Don't remember the genetic test discussed	21 (12)	57 (22)
Wanted to ask questions	n = 151 ^a	n = 211 ^a
No	81 (54)	114 (55)
Yes	70 (46)	93 (45)
More information needed		
No	88 (58)	131 (64)
Yes	63 (42)	75 (36)
Offered the choice to have genetic test		
No	53 (35)	63 (31)
Yes	60 (40)	73 (35)
Don't know	38 (25)	70 (34)
Chosen for genetic testing?		
No	21 (14)	10 (5)
Yes	101 (67)	155 (75)
Don't know	29 (19)	42 (20)

There were no statistically significant differences between the two groups.

^aQuestions only answered by people who could remember the genetic test being discussed during the consultation.

indicated that they preferred additional information, but this was not statistically significant, nor were other preferences.

The average perceived risk of the probability of an abnormal genetic test result was 15% in both groups (Figures 2 and 3). The participants with RM estimated their risk of subsequent miscarriage around 50%. The RM group estimated their chances of conceiving at least one child (75%) much higher than the poor semen quality group (50%) ($P < 0.01$). The individual perceived risks for all risk scenarios were highly correlated ($P < 0.01$). The range of r_s was 0.136–0.694.

Most participants felt that their risk indicated on the different scenarios was equal to that of the general population (data not shown). There were two exceptions. The risk of another miscarriage was perceived as higher by the RM group. The chances of conceiving at least one child and conceiving the desired number of children were perceived as lower in the poor semen quality group compared with a general population.

There was a high level of agreement between partners of a couple in knowledge, awareness and risk perceptions. Spearman's rho varied in the recurrent miscarriage group between 0.274 ($P = 0.01$) and 0.588 ($P < 0.001$), and in the poor semen quality group between 0.360 and 0.599 ($P < 0.001$).

Scores on the BDI and the STAI trait were highly correlated ($r_s = 0.714$, $P < 0.001$). This was also true for scores on the BDI and the STAI state ($r_s = 0.733$, $P < 0.001$).

The mean \pm standard deviation score on the state anxiety subscale was 36.6 ± 12.2 in the RM group and 34.6 ± 10.1 in the poor semen quality group (Table 1). A statistically significant difference was observed between the RM and the poor semen quality group on the trait anxiety subscale ($P < 0.01$), with participants with RM having higher scores. Scores on the trait and state anxiety subscales are presented for both men and women. Between men in the RM group and in the poor semen quality group, there were no statistical significant differences in scores on either the state anxiety or trait anxiety subscales. In women the state

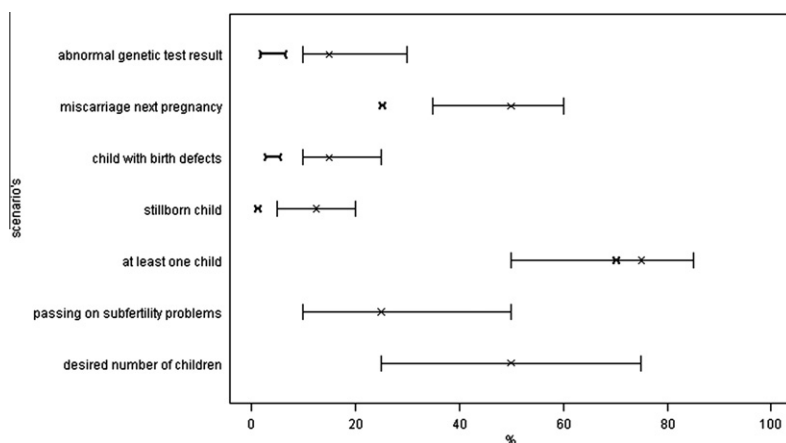


Figure 2 Perceived and absolute risks for different scenarios in couples with recurrent miscarriage. Thin line = perceived risk (median and interquartile range); thick line = absolute risk. Source: Brigham et al. (1999) and Franssen et al. (2006).

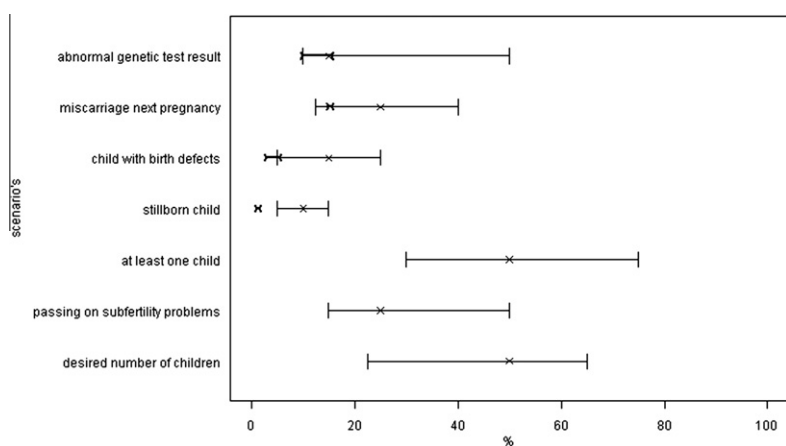


Figure 3 Perceived and absolute risks for different scenarios in couples with male partner with poor semen quality. Thin line = perceived risk (median and interquartile range); thick line = absolute risk. Source: Brigham et al. (1999), Franssen et al. (2006) and Ferlin et al. (2006).

anxiety score was significantly higher in the RM group than in women in the poor semen quality group (40.0 versus 36.3, $P = 0.04$). This was the same for scores on the trait anxiety scale (40.0 versus 34.4, $P < 0.01$).

To put the results of the state anxiety and trait anxiety subscales in perspective, a reference population of 121 Dutch couples visiting a fertility clinic for preconception counselling was used (De Weerd et al., 2001). The scores on the state anxiety and trait anxiety subscales for men in both RM and poor semen quality groups in the current study were not different from those in the reference population. The scores of women with a partner with poor semen quality also did not differ significantly. The scores of women with RM were significantly higher on both the state anxiety and the trait anxiety subscales. The means were 40.0 versus 35.5 in the state anxiety subscale ($P < 0.01$) and 40.0 versus 35.6 on the trait anxiety subscale ($P < 0.01$). Using linear regression, the state anxiety and trait anxiety scores were found to be highly correlated with the perceived risks: the higher the perceived risk, the higher the score. This was especially robust for the perceived risk of an abnormal genetic test result ($P < 0.01$) in both groups and for the majority of clinical outcomes (risk of another miscarriage,

risk of a stillborn child and risk of a child with birth defects). The range of r_s was 0.177–0.298 in the RM group and 0.234–0.317 in the poor semen quality group.

The median score on the BDI-II was 5.0 for the RM group and 3.0 for the poor semen quality group. These scores were comparable to the scores of a Dutch reference population, consisting of 505 people (Van der Does, 2002), and were classified in the lowest category of the BDI-II 'none-to-minimal depression'. In a logistic regression model, no relationships were found between any of the potential determinants and the BDI-II score.

Discussion

This study of couples undergoing genetic testing after RM or due to poor semen quality found that most couples had a suboptimal knowledge of the genetic test and overestimated their risks of receiving an abnormal genetic test result before disclosure of the test result. Women with a history of RM had higher levels of anxiety compared with a reference population and compared with women with a partner with poor semen quality. In good clinical practice

it is essential that patients are well informed about the tests and procedures they are exposed to. The pre-testing information needs to be structured and concise to enable shared decision making (Aittomaki et al., 2004).

As far as is known, this is the first study to explore the perceived risks and knowledge about genetic testing in couples with RM or with poor semen quality before disclosure of the genetic test result. Although couples with RM or with poor semen quality have different underlying mechanisms for their reproductive problems, both undergo a genetic test and face the same uncertainty before disclosure.

This study was conducted in seven of the eight academic hospitals in the Netherlands, which means it invited a representative sample of patients undergoing genetic testing at outpatient clinics of gynaecology because of reproductive problems. So far, few studies have included men as participants, as most studies addressed only attitudes and emotional responses of women undergoing fertility treatments (Engelhard et al., 2006; Quinn et al., 2009; Verhaak et al., 2007).

The current results suggest that participants did not have proper knowledge about the nature of the genetic test. They also felt they could not discuss this issue with their physician. The exact reason for this cannot be inferred from this study. It might be that physicians do not inform their patients adequately. It is also possible that patients fail to understand the information given to them or do not remember it correctly. It is known that knowledge and understanding of genetics is low among the general public since this is often complex information (Henneman et al., 2004). More than half of participants answered that they did not need any more information at the pre-test counselling. Combining this outcome with the finding that only a minority knew about the purpose of the genetic test, it seems that patient awareness of genetic counselling is low. In the current study, all participants overestimated their perceived risks for the different risk scenarios presented; their risk of receiving an abnormal genetic test result, their risk of another miscarriage, a still-born child, a child with major congenital abnormalities, their chance of conceiving at least one child and the chance of conceiving the desired number of children. This overestimation was regardless of educational status, gender or age. Most studies in hereditary cancer, such as breast and ovarian cancer, also show an overestimation of patients of perceived risks of getting cancer (Sivell et al., 2008). Whether this is a representation of a truly perceived risk or more of a coping strategy is unclear (Sivell et al., 2008). Other studies have shown that individuals, both patients and physicians, experience difficulty in understanding probability and relative risk data (Edwards et al., 2001; Gigerenzer and Edwards, 2003; Hoffrage et al., 2000). A higher perceived risk may lead to more anxiety in patients. More realistic perceptions of the possible risk could therefore lead to less anxiety in patients (Sivell et al., 2008). On the other hand, it might also be that higher anxiety levels lead to higher perceived risks (Lerman et al., 1995). Higher perceived risks have also been linked to a lack of knowledge about the disease (Partridge et al., 2008). Research has shown that the process by which individuals construct their risks is complex and influenced by many factors, such as environmental factors, occupation, stress and worry, in addition to their own experiences or family history factors (Sivell et al., 2008).

Besides this, subfertility in itself has an enormous impact on psychological wellbeing of couples. The unfulfilled desire to have children and the possible threat of permanent infertility have been shown to lead to increased levels of depression and anxiety in women (Verhaak et al., 2001). In the current study, women with a history of RM were more anxious before disclosure of the genetic test result than women with a partner with poor semen quality: they recorded higher scores on both state and trait anxiety. Recurrent pregnancy loss has been described as a traumatic event, leading to symptoms of anxiety and depression and feelings of guilt (Bagchi and Friedman, 1999; Goldbach et al., 1991). Maybe women with RM blame themselves for the miscarriages which can lead to increased anxiety, while women with a partner with poor semen quality do not. Women with RM may need extra attention and counselling during their consultations. Why women with RM also have higher scores on trait anxiety is unknown. This might be the result of a selection bias where women with a higher trait anxiety were more willing to participate. In contrast, the current study did not observe higher levels of anxiety in men with poor semen quality compared with men with RM. Only a few studies have focused on the emotional wellbeing of infertile men. It has been suggested that men speak less of personal issues and, in case of poor semen quality, will keep emotional distress to themselves and also that men are psychologically affected as much as women but are less open about it (Collins et al., 1992; Mahlstedt, 1994).

Several limitations of this study should be considered. Firstly, the response rate was about 50%. Only one telephone call to include eligible participants could be made, since in the study design there was little time before disclosure of the genetic test result. There are no data on the non-responders and for most of them it is not known why they did not participate. It is therefore possible that a form of selection bias occurred where participants who felt more attracted to the subject of the study were more willing to participate. Nevertheless, comparing the patient characteristics in this study to those of a large earlier study in couples with RM, it seems that at least the participants in the current study are a good representation of the (Dutch) clinic-based population (Franssen et al., 2005). Secondly, there are only self-reported data of participants (there was no access to the medical records of participants) and therefore it is unknown what information was given during the consultations by their clinicians. There is no real standardization in informing and counselling couples who undergo genetic testing. Last but not least, although couples were asked to complete the questionnaire independently, it cannot be ruled out that some respondents' answers may have been influenced by their partners'.

It is important to understand why patients have a suboptimal knowledge of genetic testing and why they overestimate their genetic risk. Future research should therefore include observing the consultation process and the topics discussed and the patients' knowledge afterwards. The knowledge about genetics and genetic testing among gynaecologists should also be explored. Furthermore it would be interesting to study if patients who have realistic expectations about the genetic test and their risks experience less anxiety before disclosure of the test result and if realistic

perceived risks reflect whether patients truly understand their risk.

Results of this study could be used to improve patient counselling before genetic testing, leading to well-informed patients. Potentially, this could be established if gynaecologists take more time to discuss the pros and cons of genetic testing and explain clearly the objective risks the couples have in the different outcome scenarios, such as the risk of an abnormal genetic test result or the risk of another miscarriage. The information given in these consultations should be standardized and gynaecologists probably need more training in providing genetic counselling. Since gynaecologists have limited time per patient and have to inform couples about other diagnostic tests (such as a hysterosalpingogram and blood tests for various antibodies) at the same time, it might be an alternative to work with specialized counsellors providing the genetic pre-test counselling. To improve counselling in general it is important to realize that patients might not understand all the information given to them and do not always feel free to ask questions. In conclusion, improved knowledge about genetic testing in couples with RM and poor semen quality is needed before a genetic test is performed. This could lead to more realistic expectations of patients about the consequences of the genetic test and to less anxiety before disclosure of the genetic test result.

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Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at [doi:10.1016/j.rbmo.2011.07.002](https://doi.org/10.1016/j.rbmo.2011.07.002).

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