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Article

Professionals' knowledge, attitude and referral behaviour of preimplantation genetic diagnosis for hereditary breast and ovarian cancer

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KEY MESSAGE

Professionals who are aware of PGD are more likely to consider PGD for *BRCA*; professionals who are knowledgeable about PGD are more inclined to discuss this option with patients and make a referral. Awareness and knowledge of PGD should be optimized to inform reproductive decision making among patients with *BRCA*.

A B S T R A C T

Hereditary breast and ovarian cancer caused by a *BRCA1/2* mutation is the most frequent indication for preimplantation genetic diagnosis (PGD) in the Netherlands. The extent to which involved professionals are informed about this option, however, is unclear. The few available international studies mostly represent a limited range of professionals, and suggest that their knowledge about PGD for hereditary cancer syndromes is sparse and referral for PGD is based on limited understanding. A cross-sectional survey assessing awareness, knowledge, acceptability and PGD-referral for *BRCA*

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was completed by 188 professionals involved in the field of breast and ovarian cancer or reproduction. One-half of professionals were aware of PGD for *BRCA*, and most had a low to moderate level of knowledge. A total of 86% considered PGD for *BRCA* acceptable and 48% had referred patients with *BRCA* for PGD. Awareness and knowledge was higher among professionals who worked at a university hospital (compared with a general hospital). Knowledge of PGD was positively associated with discussing and referring for PGD, and PGD acceptability was associated with previous awareness. Although PGD counselling is the primary responsibility of the geneticist, other involved professionals may be gatekeepers as patients rely on them for raising awareness and referral.

Introduction

About 5–10% of all cancers are caused by a hereditary predisposition (Garber and Offit, 2005). Women with a *BRCA1/2* mutation face an elevated risk of 27–57% of developing breast cancer and 6–40% of developing ovarian cancer by the age of 70 years (Brohet et al., 2014; Chen and Parmigiani, 2007). Preventive possibilities are limited to chemoprevention and prophylactic surgery, which can be both physically and psychologically demanding. Although therapeutic options are available, hereditary breast and ovarian cancer (HBOC) caused by a *BRCA1/2* mutation accounts for a disproportionally large amount of life years lost as it occurs at a relatively young age (Roukos and Briasoulis, 2007). As this is an autosomal dominant predisposition, there is a 50% risk of passing it on to the next generation. In the Netherlands, couples with a *BRCA1/2* mutation and a wish for a biological child have three reproductive options: a natural pregnancy, implying acceptance of the risk of passing on the *BRCA* mutation; prenatal diagnosis (diagnosis during pregnancy and possible termination of pregnancy in case of a female carrier); and preimplantation genetic diagnosis (PGD) (selection of IVF and intracytoplasmic sperm injection [ICSI] embryos, free of the *BRCA* mutation before implantation in the uterus).

As use of PGD was permitted for late-onset genetic cancer syndromes in the Netherlands in 2008, HBOC caused by a *BRCA1/2* mutation has been the most frequent indication for PGD, with 25 couples that started a PGD treatment for *BRCA* in 2013, 36 couples in 2014 and 45 couples in 2015 (PGD Netherlands, year reports, 2013; 2014; 2015). Ethical concerns about PGD for late-onset cancer syndromes such as *BRCA* have risen internationally among patients and professionals owing to the condition's adult onset character, incomplete penetrance and availability of (albeit physically and emotionally traumatic) preventive and therapeutic options (Julian-Reynier et al., 2009; Klitzman et al., 2013; Rich et al., 2014).

Although PGD is nowadays available as a reproductive option for couples with *BRCA* in many countries, published data show that knowledge among involved health professionals is sparse and most would refer for PGD based on limited understanding of the procedure and its applications (Abbate et al., 2014; Caldas et al., 2010; Klitzman et al., 2013). Moreover, professionals' intentions to refer eligible patients for PGD counselling often exceed their actual referral behaviour (Brandt et al., 2010; Klitzman et al., 2013). The few available studies focusing specifically on hereditary cancer syndromes (Brandt et al., 2010; Julian-Reynier et al., 2009; Quinn et al., 2014) show that professionals' acceptability of PGD is often influenced by the nature of the predisposition and the patient's personal history of cancer. The previously mentioned studies focused on various outcomes, such as assessing PGD knowledge by awareness or measuring acceptability of PGD depending on patients' family history of cancer and reproductive history among gynaecologists, gynaecological oncologists, obstetricians or oncology nurses; only one study included clinical geneticists, the specialists who are primarily involved in PGD (Julian-Reynier et al., 2009). Therefore, we assessed awareness, knowledge and acceptability of PGD for *BRCA* among health professionals, and their referral behaviour (including clinical geneticists and genetic counsellors), and investigated possible associations of these outcomes with clinical and demographic factors. This study was carried out as part of an overarching project aimed at enhancing guidance and psychological support for couples with *BRCA* and a child wish in the Netherlands.

Materials and methods

Participants and procedures

Participants were recruited in collaboration with the following Dutch associations of health professionals that are involved in the field of hereditary breast and ovarian cancer, reproduction, or both: The Association of Clinical Genetics Netherlands (VKGN), The Dutch Association of Genetic Counsellors (NVGC), The Dutch Association for Obstetrics and Gynaecology (NVOG), and The Dutch Association for Oncology (NvVO). The associations sent one mass mail inviting their members to participate in the study. No reminders were sent. The approached professionals were gynaecologists, clinical geneticists, genetic counsellors, medical oncologists and fertility physicians. The single inclusion criterion was being a medical specialist involved in the field of reproduction, oncology, or both, whereas the single exclusion criterion was insufficient understanding of the Dutch language as the survey was in Dutch. The email invitation contained brief information about the study, contact details of the researcher and a link to the online questionnaire. It was clearly stated that participants gave their informed consent by initiating the questionnaire. Procedures were approved by the Medical Ethics Committee of the Maastricht UMC+, reference number: METC 12-4-075, dd 18-06-2012.

Questionnaire

To explore an appropriate basis for the questionnaire content and to ensure relevance of the questions for the approached professional groups, seven in-depth telephone interviews were conducted with several medical professionals (two clinical geneticists, two genetic counsellors, one gynaecologist, one gynaecologic oncologist and one medical oncologist) before the start of the study. The duration of these interviews was about 30 mins, and they addressed awareness, knowledge, attitude, referral behaviour and informational needs of PGD for *BRCA*. The interviews were audio taped, transcribed verbatim and

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