Editorial: genetic aspects of preconception consultation in primary care

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Welcome to this special theme issue of the *Journal of Community Genetics* which focuses on the topic of preconception care. It aims to review this subject from the perspective of primary health care, while recognising that the types of health-care practitioner included within the primary healthcare system differ significantly between countries covering, for example, family practitioners, obstetricians, internal physicians and midwives. The core features of preconception care are the assessment of risk to the future child and mother and provision of information and support about potential options to manage any identified risks. The key element of course is that this occurs prior to conception since this allows couples a greater range of reproductive choices and proactive management of existing medical or lifestyle factors which could affect a future pregnancy. The themed issue covers in detail several important genetic aspects of preconception care and looks ahead to future scenarios as new genetic technologies rapidly increase the range of genetic risks which could be identified preconceptionally.

Even with the predicted growth in DNA-based testing, the fundamentals of good medical and psychosocial assessment as part of a preconception consultation will remain. In the context of identifying genetic risks, the family medical

history continues to play a key role (Bennett 2012). Bennett provides an excellent overview of this, reminding us that the family medical history can also give insight into shared environmental exposures and offer important psychosocial clues as well. One of the challenges in primary care is the time required to obtain a full three-generational pedigree which may be necessary to assess fully any genetic risks. This highlights an important potential role for electronic medical records which can be updated readily and allow patients to enter their own family history in advance of their consultation in primary care.

Comprehensive preconception care requires assessment of the woman's personal health, health behaviours and past medical history as well as the couple's family medical history. Several common chronic diseases or their pharmacological treatments increase the risk of adverse pregnancy outcomes and congenital anomalies and ideally require optimization of management, including careful consideration and potential changes to the treatment regimen, before conception. Diabetes and epilepsy are important examples of this and may also require advice on higher dosage of periconceptional folic acid supplementation. Preconception care also allows the assessment of immunisation status, and potential risk of exposure to common pathogens such as rubella, influenza and varicella which can have serious consequences in pregnancy, including teratogenic effects. Lifestyle risk factors, in particular smoking, alcohol and illicit drug use should also be explored and cessation recommended, with referral for treatment as appropriate. However, as Riedijk and colleagues discuss, achieving behaviour change, even in the context of planning a pregnancy, can be challenging (Riedijk et al. 2012). Mulvihill discusses the specific issue of past exposure to mutagens; this is increasingly relevant as survival from childhood cancers improves as well as rarer exposures to radiation or other environmental pollutants. Evidence from survivors of cancer and the

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Japanese atomic bombs suggests that one can be relatively reassured about the absence of germ-cell mutations caused by chemotherapy or radiation exposure (Mulvihill 2012).

Hamamy discusses specific issues of preconception care in consanguineous marriages (Hamamy 2012). Consanguinity is a common cultural trend particularly in North Africa, West Asia and South India; emigrants from these countries often continue this practice and so it is important for all practitioners to be aware of the implications of consanguineous marriage and provide initial preconception counselling. The family medical history is again an important initial step in this process. In the absence of a known genetic disorder in the family, the risks are still increased but actually lower than what a couple might expect: first cousin marriages have roughly double the risk of a child with a birth defect with an absolute risk of approximately 5 %. Consanguineous couples with a family history of a genetic disorder would require more detailed assessment by a specialist genetic counselling service.

In addition to assessment of the couple's personal and family medical history, genetic carrier screening options should also be considered as part of comprehensive preconception care. The selection of specific tests should be guided by carrier frequencies and the couple's ethnic ancestry, as discussed by Metcalfe: cystic fibrosis in those from Northern Europe; haemoglobinopathies in people from Southern Europe, the Middle East, Africa, India and South East Asia; and Tay Sachs in those of Ashkenazi Jewish descent (Metcalfe 2012). More recently, studies have explored offering carrier screening for Fragile X syndrome and spinal muscular atrophy to general populations. Several studies have specifically looked at outcomes of offering genetic carrier screening in primary care both preconception and prenatally. Most have demonstrated that these tests can be effectively offered in primary care without causing significant adverse psychological outcomes. Uptake of the test is affected by the method of offering the test as well as the specific setting such that active opportunistic testing in a family planning clinic had higher uptake compared, for example, to a letter of invitation from general practice. Other important outcomes such as informed choice, as opposed to simply test uptake, should also be taken into account.

Read and Donnai discuss the range of options available to a couple if a significant genetic risk is identified (Read and Donnai 2012). Non-directive genetic counselling is central to this to allow a couple to reach a fully informed decision. Recent developments in pre-implantation genetic diagnosis (PGD) are an important rationale for preconception counselling as it offers a potentially more acceptable approach to couples wishing to avoid pre-natal diagnosis and termination of an affected pregnancy. However, as Read and Donnai discuss, PGD is not an 'easy option' given its reliance on IVF technology and associated significant

psychological stress and financial cost. Advances in non-invasive pre-natal diagnosis may soon offer a safer and more acceptable method than amniocentesis or chorionic villous sampling, but only for the detection of mutations of paternal origin or numerical chromosome anomalies. It does not of course avoid difficult decisions about termination of an affected pregnancy. The use of donor gametes, adoption or remaining childless should also be offered to allow a couple to make fully informed reproductive choices.

Preconception counselling raises important ethical challenges which are clearly elaborated in the paper by De Wert et al. (2012). The authors distinguish the ethics of individual preconception counselling from that of population carrier screening. Individual counselling can be viewed as offering couples autonomy and reproductive choice; the alternative 'prevention view' of individual counselling risks placing pressure on couples to make the perceived 'right choice' and terminate an affected pregnancy. Preconception carrier screening raises broader ethical concerns about the resurgence of eugenics and the 'expressivist argument' that such population screening programmes express a discriminatory view against disability. In this context, it is important therefore to ensure that carrier screening programmes can demonstrate a positive balance of benefits over harms for participants, and seek to support informed choice not simply high test uptake. The potential psychosocial harms, which are critical to consider in the context of this ethical framework, are further discussed in the paper by Riedijk et al. (2012).

Current genetic carrier screening programmes are limited to a few specific genetic conditions. The rapid advances in 'next generation sequencing' could significantly change this, as described by Ropers (2012). Examples provided include a diagnostic test panel of approximately 90 genetic defects associated with X-linked intellectual disability and a second panel covering mutations in 500 genes for severe recessive childhood disease.

These technological advances raise the important question of how health services can provide adequate counselling for this growing array of genetic tests available to couples contemplating pregnancy. This theme issue of the journal is about preconception care in primary care. As several authors discuss, there are inherent difficulties of delivering preconception care, not least that perhaps up to half of pregnancies are unplanned (Riedijk et al. 2012). While in some communities with a high prevalence of specific autosomal recessive conditions, there is a culture of school-based or pre-marital testing (Metcalfe 2012); in many countries, preconception counselling is far from a cultural norm. A further issue is the capacity for primary care to offer preconception counselling. As discussed by Ten Kate (2012), a study of preconception counselling in primary care found that 42 % of couples required further action by the GP and 4 % referral to a clinical geneticist

based upon identified risks. In the Netherlands, preconception care has become more integrated into primary care partly through the establishment of midwifery-led clinics (Riedijk et al. 2012). If the costs of next-generation sequencing fall as predicted (Ropers 2012), offering preconception counselling will only become more complex but there are insufficient specialist genetic services available to provide this counselling. New models of providing preconception care in the community need to be developed and evaluated if we are to offer couples the opportunity to make informed decisions about the growing array of genetic tests that will be available soon.

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