UPSALA JOURNAL OF MEDICAL SCIENCES, 2016
VOL. 121, NO. 4, 289–294
http://dx.doi.org/10.1080/03009734.2016.1218575

Taylor & Francis
Taylor & Francis Group

ORIGINAL ARTICLE

OPEN ACCESS

Swedish parents' interest in preconception genetic carrier screening

Maria Ekstrand Ragnar[a,b], Tanja Tydén[b], Ulrik Kihlbom[c] and Margareta Larsson[a]

[a]Department of Women's and Children's Health, Uppsala University, Sweden; [b]Department of Public Health and Caring Sciences, Uppsala University, Sweden; [c]Centre for Research Ethics and Bioethics, Uppsala University, Uppsala, Sweden

ABSTRACT

Introduction: Genetic technologies advance rapidly. It is possible to undergo genetic carrier screening before pregnancy to examine genetic risks to future offspring. We aimed to investigate parents' interest and motives towards preconception genetic carrier screening (PCS) as well as factors associated with interest in PCS.

Material and methods: Our study sample consists of 777 parent couples within the longitudinal Swedish Pregnancy Planning study. Women responded to questionnaires at three occasions: in early pregnancy, late pregnancy, and one year after childbirth. Male partners responded to one questionnaire one year after childbirth.

Results: One-third of the parents were positive (30% versus 34% of women and men, respectively), less than a third were negative (26% versus 28%), and 45% versus 38% were uncertain about whether to consider PCS before a future pregnancy. No differences in PCS interest were found between women and men ($P = 0.091$), but a higher proportion of women were concerned about negative consequences (53% versus 46%, $P < 0.003$) and were 'opposed to such a way of child selection' (31.8% versus 25.2%, $P = 0.002$). Factors associated with PCS interest were experiences of prenatal diagnostics and positive attitudes towards finding out or choosing sex of one's child (women), and prenatal diagnostics, self-rated poor health, and pregnancy planning (men).

Conclusion: Both women and men had relatively high uncertainty towards PCS, but women were more concerned about negative consequences. The future extent of the clinical utility of PCS is currently unknown, but parents' interests and doubts are important aspects to consider.

ARTICLE HISTORY
Received 25 April 2016
Revised 6 July 2016
Accepted 26 July 2016

KEYWORDS
Interest; motives; parents; preconception genetic carrier screening

Introduction

Genetic technologies are advancing rapidly. With safer, faster, and cheaper tests, the possibility for healthy couples to undergo preconception carrier screening (PCS) for autosomal recessive conditions has increased markedly in recent years (1). In order for an autosomal recessive disorder to develop (for example cystic fibrosis (CF), spinal muscular atrophy (SMA), thalassemia, and sickle cell disease), the abnormal gene must be present in both biological parents, and, if so, there is a 25% risk of them having a child with the autosomal recessive disease they both carry. It has been estimated that the birth prevalence of severe recessive disorders is between 0.25% and 0.5%, which means that approximately 1–2 in 100 couples are at risk of having a child affected with a recessive genetic condition (2). Healthy carriers usually do not know of their carrier status unless they have a known family history of recessive genetic disorder, or if they have had an affected child.

Preconception carrier screening (PCS) is usually defined as the detection of carrier status *before* pregnancy, in couples or persons who *do not* have a known increased risk of being carriers, to determine the risk of having a child with a recessive genetic disorder (1). Identifying carriers of autosomal recessive disorders before pregnancy has the potential to benefit prospective parents by making them aware of the possible genetic risks to a future child, and of the reproductive options available. These options include not only prenatal diagnosis, followed (or not) by pregnancy termination in case of an affected fetus or by coming to terms with the risk, but may also include the possibilities of using preimplantation genetic diagnosis, donor sperm or oocytes, seeking adoption—or refraining from having children (3,4).

Traditionally, PCS has been limited to a small number of specific tests and generally offered to certain (often ethnic) high-risk populations (5). However, advancements in genomics have opened new possibilities for carrier screening in whole populations without a prior risk or family history; in the US for example, the American College of Medical Genetics recommends all couples regardless of ancestry or geographic origin to be offered screening for SMA (6), and the American College of Obstetricians & Gynecologists recommends all women of reproductive age (regardless of ancestry or geographic origin) to be offered screening for CF (7). Besides this, the rapid development in genetic technologies enables broader test panels to be used. This allows screening for the carrier status of a large number of inherited conditions at one go (8–10). The new technology has also opened up for

CONTACT Maria Ekstrand Ragnar Maria.Ekstrand@kbh.uu.se Department of Women's and Children's Health, Uppsala University, Uppsala, Sweden.

commercial companies to offer a wide array of genetic screening tests and services, either directly to the consumer, or via referrals from private health care providers (5).

Previous research has shown generally accepting views towards PCS. However, this research has mostly been limited to single-gene conditions and performed among target populations for certain conditions—for example, individuals already affected with a genetic recessive disorder and their parents and relatives (11). There is still limited knowledge about public perceptions towards population-based PCS and PCS using expanded panels. However, one study found that public opinions towards expanded PCS were multifaceted, deriving from limited knowledge, feelings of not being at risk, worries that testing might cause unnecessary stress, and/or financial concerns regarding cost of testing (12).

PCS is not yet in practice in Sweden, and couples without a family history of recessive (and/or other) genetic disorders are currently not offered screening within the public health care system. However, anyone can order PCS tests through international commercial companies online. Thus, little is known about interest in PCS among potential parents-to-be. This study aimed to investigate parents' interest in PCS, stated motives why or why not to consider such screening, and factors associated with interest in PCS. We hypothesized that interest in PCS would be associated with parents having undergone prenatal diagnostics, having requested information about the sex of the fetus prior to delivery, and/or having responded favorable to a hypothetical question about fetal sex selection. Furthermore, we postulated that interest in PCS would be associated with a high degree of pregnancy planning, self-rated poor health, previous miscarriage(s), and socio-demographic background characteristics such as a high level of education. To date, there has been no exploration of the factors associated with PCS interest or factors that are likely to motivate individuals to consider PCS.

Materials and methods

Setting, participants, and procedure

This study was part of the Swedish Pregnancy Planning (SWEPP) study—a longitudinal cohort study examining lifestyle and health in connection to pregnancy and childbirth among nearly 5,000 women and nearly 1,000 partners, previously described by Stern et al. (13).

In Sweden, antenatal care is offered free of charge to all women. As long as the pregnancy is normal, antenatal care is provided exclusively by midwives. The study invited a total of 215 antenatal clinics, of which 153 (71%) agreed to participate. The antenatal clinics ranged from small to large, in the countryside and in the cities, in nine Swedish counties (about 2,500,000 inhabitants).

Pregnant women were recruited in consecutive order at registration in the antenatal clinic after having received verbal and written information about the study by their midwife. Of 5,494 women invited, 4,969 agreed to participate. The women received questionnaires to fill out at the clinic or at home and return by post in a prepaid envelope. The women filled out one questionnaire (Q1) in early pregnancy (n = 3,389) and one (Q2) around gestational week 34 (n = 2,584). A third questionnaire (Q3) was sent out 12 months post-partum to 2,000 women who had replied previously to both Q1 and Q2. Along with Q3, a partner questionnaire (Q3P) containing a selection of questions from Q1–3 was enclosed (n = 2,000), and the women were asked to hand it to their partner/the other parent (of the child that was born 12 months earlier). In total 817 partners/parents filled out and returned the Q3P. Recruitment took place during the period September 2012 through July 2013, and data collection was completed in March 2015.

Our study sample consists of 777 parent couples (n = 777 women, and n = 777 men) who had responded to *all* questionnaires (Q1–Q3 and Q3P) including the two study-specific questions about PCS of relevance for the study aim. We chose to exclude female partners (n = 15) since recessive inherited conditions concern biological parents only.

Ethics

The study was approved by the Regional Ethical Review Board in Uppsala, Sweden (reference number 2010/085). Participation was voluntary, and participants were informed that the care given at the antenatal clinic was not related to their participation in the study. Informed consent was obtained from all participants.

Questionnaires

Q1 contained 148 items, whereas Q2, Q3, and Q3P contained 114 and 156 items, respectively, most of which were multiple choice questions, covering a range of pregnancy-related topics. Researchers, clinicians, and laypeople reviewed the questionnaires, and a pilot study was conducted, after which some items were adjusted.

In the present paper we use questionnaire items relevant to the aim of this study: questions about PCS, socio-demographic background characteristics, questions about personal health, health of the child, and reproductive history.

Questions about socio-demographic background characteristics covered: the woman's age, partner's age, country of birth, level of education, occupation, and household income. Questions about personal and child health covered: self-reported health status, smoking, and whether or not the child had any congenital conditions. Reproductive health and history covered: parity, miscarriages, IVF treatment, level of pregnancy planning, intentions of future childbearing, experience of prenatal diagnostics, knowledge about the sex of the fetus prior to delivery, and attitudes towards fetal sex selection.

The level of pregnancy planning was measured using a single item, 'How planned was your current pregnancy?', and a five-step Likert scale with response alternatives ranging from 'very planned' to 'very unplanned', previously described by Tydén et al., Backhausen et al., and Stern et al. (13–15).

The questions about PCS were introduced with a short text as follows:

Some congenital disorders can be passed on. The mutated gene(s) might be carried by one or both parents.

Both parents could be carriers without being or becoming affected by genetic disease, BUT if a child inherits the gene from both parents there is an increased risk that the child will be affected. In the future, couples will be able to test genetic carrier status before pregnancy occurs.

Participants were thereafter asked whether or not they would consider taking such a test ('yes/no/don't know'), what motive(s) would be of importance to them in their decision-making, and whether they believed such a test could lead to negative consequences if it were offered to prospective parents in general ('yes/no/don't know'). The statements about motives for or against PCS were developed based on the literature and covered topics commonly discussed in the context of preconception carrier screening. The motives statements were: 'I do not want the future child to suffer', 'I am opposed to such a way of child selection', 'It would be an act of responsibility', 'It is expected of me by others', 'I do not want my child to suffer', 'I want to exclude the risk of having a child with a severe genetic disorder', 'Other motive(s)'.

Data analysis

Data were entered and analyzed using IBM SPSS Statistics version 22 (IBM, Armonk, NY, USA). Categorical data are presented as frequencies and percentages, and continuous data by range, means, and standard deviations. Differences regarding women's and men's perceptions of and stated motives for preconception genetic screening were analyzed using McNemar–Bowker's test for categorical variables. For all statistical analyses, a two-sided P value <0.05 was considered significant.

A binary logistic regression was used to analyze the association between the dependent variable (interest in PCS) and the independent variables. Some variables were recoded and dichotomized. Data regarding household income were collapsed into two categories; ($\leq$39,999/$>$40,000 Swedish krona per month), as was occupation including studies ($<$50%/$\geq$50%). The level of pregnancy planning was categorized as 'planned' (very or fairly planned) or 'unplanned' (neither planned nor unplanned, fairly or very unplanned), level of education as 'high' (university/college) or 'low' (no complete education, elementary school or high school/vocational education), self-rated health as 'good' (very/fairly good) or 'poor' (neither good nor bad, fairly/very poor), and interest in PCS as 'yes' or 'no/uncertain'.

Results

Background characteristics, including participants' reproductive health and history and interest in PCS, are presented in Table 1. One-third of the parents were positive towards PCS (30% versus 34% of women and men, respectively), 25% versus 28% were negative, and 45% versus 38% were uncertain about whether they would consider a genetic test before a future pregnancy.

No differences were found between women and men regarding interest in PCS ($P = 0.091$). A higher proportion of women than men were concerned about negative consequences if PCS were to be offered to prospective parents (53%, $n = 409$ of women versus 46%, $n = 356$ among men, $P < 0.003$).

Comparison *within* the couples regarding motives for and against PCS are presented in Table 2. A higher proportion of women compared to men ticked avoiding the (future) child to suffer ($P < 0.000$), and 'opposed to such a way of child selection' ($P = 0.002$).

Binary logistic regression analysis showed that increased age, being born outside Sweden, self-rated poor health, having undergone prenatal diagnostics, wanting to know the sex of the fetus prior to delivery, and having positive attitudes towards fetal sex selection was positively associated with women's interest in PCS. Among the partners, self-rated poor health, having had a planned pregnancy and having undergone prenatal diagnostics was associated with interest in PCS. See Table 3.

Discussion

Interest and motives

Our most interesting result was the varying interest among parents towards PCS; about one-third would like to undergo PCS before a future pregnancy, a little less than a third would not, and 45% of women versus 38% of men were uncertain towards such screening.

PCS is a new concept in Sweden and is neither publicly debated nor offered as standard care. Even though available through commercial companies, there is no direct-to-consumer marketing on the Swedish market at present. Consequently, it is reasonable to believe that knowledge and awareness regarding PCS among the general public, as well as among Swedish health care providers, is low. Still, a third of the parents in our study claimed to be willing to undergo PCS. However, it is possible that even more parents would have been willing to consider PCS, had they been more aware about the concept, and especially if PCS was endorsed by health care professionals, commercially marketed, and/or offered within the public health care system. It has been argued that such 'routinization' could diminish potential controversies and even justify the introduction of new medical or genetic technologies (16). As a comparison, screening for Down syndrome is nowadays widely accepted among Swedish parents-to-be and more or less viewed by many as part of standard care.

Although one-third of the parents showed interest in PCS, the majority was negative or hesitant. Low interest or uncertainty must, however, not be explained by lack of knowledge alone. It could also—unrelated to one's pre-existing knowledge—indicate a general resistance against mass screening for carrier status, or reluctance to take part in seeking information that such screening would entail. In our study, 31.8% of women and 25.2% men motivated their non-interest in PCS with being 'opposed to such a way of child selection', and one in five had no demand for receiving information about carrier status. With expanded test panels, and variation in penetrance and severity of the traits that one can be tested for, risk estimation will become quite difficult, both for the prospective parents as well as for health care

Table 1. Background characteristics among women ($n = 777$) and male partners ($n = 777$). Categorical data are presented as frequencies and percentages, *n* (%), and continuous data are presented as means, standard deviations (SD), and range.

	Women		Male partners	
Variable	*n*	Value	*n*	Value
Mean age (years)	759	29.78	775	35.30
SD		4.639		5.584
Range		17–47		16–58
Household income (≤39,999/>40,000 SEK/month), *n* (%)	751	478/273 (63.6/36.4)	770	439/331 (56.5/42.6)
Occupation <50%/≥50%, *n* (%)	777	307/470 (39.5/60.5)	777	140/637 (18.0/82.0)
Education	766		775	
No completed education/elementary school, *n* (%)		23 (3.0)		33 (4.3)
High school/vocational education, *n* (%)		285 (37.2)		401 (51.7)
University/college, *n* (%)		458 (59.8)		341 (44.0)
Born outside Sweden, *n* (%)	773	66 (8.5)	772	56 (7.3)
Self-rated health good/poor, *n* (%)	775	599/176 (77.3/22.7)	773	517/256 (66.9/33.1)
Smoking, yes/no, *n* (%)	776	32/744 (4.1/95.8)	774	57/717 (7.4/92.7)
Nullipara/multipara, *n* (%)	510	111/399 (21.8/78.2)		–
Parity	500			
1, *n* (%)		241 (48.2)		–
2–3, *n* (%)		207 (41.4)		–
≥4, *n* (%)		52 (10.4)		–
Miscarriage, yes/no, *n* (%)	769	177/592 (23.0/77.0)		–
IVF, yes/no, *n* (%)	777	33/744 (4.2/95.8)	777	33/744 (4.2/95.8)
Prenatal diagnostics, yes/no, *n* (%)	777	314/463 (40.4/59.6)	777	314/463 (40.4/59.6)
Congenital condition (in child born 12 months ago), yes/no, *n* (%)	777	8/769 (1.0/99.0)	777	8/769 (1.0/99.0)
Pregnancy planning	776		775	
Unplanned *n* (%)		156 (20.0)		142 (18.3)
Planned *n* (%)		620 (80.0)		633 (81.7)
Wanting to know sex of child, yes/no, *n* (%)	758	375/383 (49.5/50.5)	760	395/365 (52.0/48.0)
Sex selection attitudes	760			
Positive, *n* (%)		34 (4.5)		–
Negative/uncertain, *n* (%)		726 (95.5)		–
Interest in PCS	777		777	
Yes		233 (30)		261 (33.6)
No		198 (25.5)		219 (28.2)
Uncertain		346 (44.5)		297 (38.2)
Want more children, *n* (%)	776		776	
Yes		424 (54.6)		338 (43.6)
No		203 (26.2)		278 (35.8)
Uncertain		149 (19.2)		160 (20.6)

Table 2. Motives for and against PCS, comparison within couples ($n = 710$).

	Women/male partners $n = 710/710$ (%)	McNemar *P* value
Not wanting (future) child to suffer	364/286 (51.3%/40.3%)	**0.000**
Exclude the risk of having a child with a severe genetic disorder	321/326 (45.2%/45.9%)	0.811
An act of responsibility	165/194 (23.2%/27.3%)	0.058
Expected by others	4/12 (0.6%/1.7%)	0.057
Opposed to such a way of child selection	226/179 (31.8%/25.2%)	**0.002**
Not wanting to know/such information	152/155 (21.4%/21.8%)	0.894
Other motives	48/60 (6.8%/8.5%)	0.261

professionals counseling them (17). So, even if PCS promises to enhance reproductive autonomy through offering more reproductive options (18), people may instead feel burdened when facing decisions with uncertain but significant long-term effects on their lives. Worries about child selection and/or doubts about receiving information about potential carrier status are of course important aspects to consider if, or when, PCS is further implemented in Sweden.

We found no difference between women and men regarding their interest in PCS. However, a higher proportion of women, compared to men, were concerned about negative consequences if PCS were to be offered to prospective parents in general. Advancements in genetic technologies and PCS often raise ethical concerns and have been frequently discussed in the literature. Besides issues focusing on informed consent and autonomy (1,17), the ethical discussion includes, for example, concerns about a supposed renaissance of 'eugenics' (aiming at improving the genetic quality of the human population) (19), medicalization (potentially eroding people's confidence in the solidity of their health) (20), and discrimination (including carrier stigma, insurance discrimination, and avoidance of the birth of a child with a potentially severe disorder) (18,21,22).

The most common motives *for* PCS, ticked by nearly half of the respondents, were not wanting a future child to suffer or not wanting to face the risk of having a child with a severe genetic disorder. Very few stated that their motivation for opting for PCS would be feeling expectations from others. This finding was to be expected as no PCS programs have been implemented in Sweden so far. However, implementation of new genetic screening practices may generate various kinds of pressure among targeted users (23–25). Hence, such a development could of course be imaginable, should PCS ever become part of Swedish standard care.

Factors associated with PCS

We found no clear pattern of factors associated with PCS interest, except for joint experiences of prenatal diagnostics

Table 3. Characteristics associated with parents' interest in PCS—a binary logistic regression analysis ($n = 1,554$).

Variables	Women ($n = 777$)			Partners ($n = 777$)		
	OR	95% CI	*P* value	OR	95% CI	*P* value
Age	**0.95**	**0.91–0.99**	**0.04**	1.02	0.99–1.05	0.14
Country of birth (Sweden[a]/outside Sweden)	**1.98**	**1.08–3.64**	**0.02**	1.22	0.67–2.23	0.51
Education (high[a]/low)	1.05	0.71–1.56	0.78	1.18	0.85–1.65	0.31
Household income (high[a]/low)	0.77	0.53–1.13	0.18	0.91	0.65–1.27	0.58
Self-rated health (good[a]/poor)	**1.56**	**1.04–2.33**	**0.02**	**1.50**	**1.08–2.08**	**0.01**
Pregnancy planning (planned[a]/unplanned)	1.02	0.66–1.56	0.92	**0.59**	**0.38–0.91**	**0.01**
Prenatal diagnostics (yes[a]/no)	**0.57**	**0.40–0.81**	**0.00**	**0.63**	**0.46–0.87**	**0.00**
Wanting to know sex of child (yes[a]/no)	**0.56**	**0.39–0.80**	**0.00**	0.79	0.58–1.09	0.16
Gender selection (positive[a]/negative attitude)	**0.26**	**0.12–0.57**	**0.00**			
Previous miscarriage (yes[a]/no)	0.83	0.55–1.26	0.39			

[a]Reference category.

and positive attitudes towards the possibility of finding out—or even choosing—the sex of one's child (among the women). In contrast to our hypothesis, level of education or history of miscarriages did not seem to influence parents' interest in PCS; however, poor health and high degree of pregnancy planning did (by male partners). PCS interest was also more likely among women with non-Swedish origin. The varying associations to PCS interest are not easily explained, but probably reflect the diversity in PCS attitudes among the parents in our study.

Clinical implications

The future extent of the clinical utility of PCS in Sweden is currently unknown. It is likely that awareness about PCS will soon increase among the general public, resulting in increased screening demand among some, and a remaining reluctance among others. Diverse opinions among the target group will require great efforts to individualize counseling, provide complete and transparent information, and to ensure autonomy for prospective parents in their reproductive choices. Primary care professionals, midwives, and gynecologists will have an important role to play in helping potential parents navigate the rapidly changing landscape of genetic technologies and screening services. No doubt, another issue for reflection will be how to prioritize resources within preconception care. If a growing number of people request PCS in the future, this will probably have noticeable effects in various areas, including economic aspects, health care delivery, and genetic information services, as well as insurance issues, to mention a few. Preconception genetic screening options will continue to advance. Understanding the role of women and men's interests and doubts, as well as health care professionals' perceptions of PCS, is vital in order to prepare for future developments of PCS in Sweden.

Strengths and limitations

Our study included a large sample of both women and men recruited via antenatal clinics from different settings, in both rural and urban areas in Sweden. As shown previously, our female study population is representative for women attending antenatal care in Sweden, except for women born outside Sweden that are underrepresented (13). Regarding the male partner, we had to rely on the participating women for distribution of questionnaires, and consequently we have limited data on the external dropout rate and characteristics of those who chose not to participate.

Since the community awareness regarding PCS is likely to be low, people in general may have difficulties relating to hypothetical questions about PCS. An introductory text explaining the core concept of PCS was therefore presented to the respondents in the questionnaire, directly followed by the questions and the response alternatives. The introductory text was purposely rather brief. The text included neither severity nor onset of potential recessive disorders, nor was a distinction made regarding different forms of PCS (such as screening for single genes or using expanded panels). This could affect the way respondents interpreted and responded to the questions.

The present study design does not allow any further exploration of what kind of concerns respondents might have had towards PCS, nor does it enable deeper understanding of the motives for or against PCS. This needs to be further explored, preferably by using a qualitative study design among potential parents-to-be.

Conclusion

Both women and men had relatively high uncertainty towards PCS, but women were more concerned about negative consequences. Factors associated with interest in PCS were, among others, experiences of prenatal diagnostics and positive attitudes towards finding out or choosing the sex of one's child. The future extent of the clinical utility of PCS is currently unknown, but patients' interests and doubts are important aspects to consider.

Acknowledgements

We thank Andreas Rosenblad for statistical advice and Jenny Stern, Jennifer Drevin, and Maja Bodin for distribution of questionnaires and data collection.

Disclosure statement

The authors report no conflicts of interest.

Funding

Medical Faculty, Uppsala university, Uppsala, Sweden.

ORCID

Tanja Tydén http://orcid.org/0000-0002-2172-6527

References

1. Henneman L, Borry P, Chokoshvili D, Cornel MC, van El C, Forzano F, et al. Responsible implementation of expanded carrier screening. Eur J Hum Gen. 2016:e1–e12.
2. Ropers HH. On the future of genetic risk assessment. J Community Genet. 2012;3:229–36.
3. The American College of Obstetricians and Gynecologists. Frequently asked questions. FAQ179 Pregnancy [internet]. Available at: http://www.acog.org/~/media/For%20Patients/faq179.pdf [retrieved 14 April 2016].
4. Borry P, Henneman L, Lakeman P, Kate LP, Cornel M, Howard H. Preconceptional genetic carrier testing and the commercial offer directly-to-consumers. Hum Rep. 2011;26:972–7.
5. Bajaj K, Gross S. Carrier screening: past, present, and future. J Clin Med. 2014;3:1033–42.
6. ACMG practice guidelines: carrier screening for spinal muscular atrophy. Genet Med. 2008;10:840–42.
7. American College of Obstetricians and Gynecologists Committee on Genetics ACOG Committee Opinion No. 486: Update on carrier screening for cystic fibrosis. Obstet Gynecol. 2011;117:1028–31.
8. Srinivasan B, Evans E, Flannick J, Scott Patterson A, Chang C, Pham T, et al. A universal carrier test for the long tail of Mendelian disease. Reprod BioMed Online. 2010;21:537–51.
9. Kingsmore S. Comprehensive carrier screening and molecular diagnostic testing for recessive childhood diseases. PLoS Curr. 2012 May 2:e4f9877ab8ffa9.
10. Edwards JG, Feldman G, Goldberg J, Gregg AR, Norton ME, Rose NC, et al. Expanded carrier screening in reproductive medicine – points to consider: a joint statement of the American College of Medical Genetics and Genomics. Obstet Gynecol. 2015;125:653–62.
11. Ioannou L, McClaren BJ, Massie J, Lewis S, Metcalfe SA, Forrest L, et al. Population-based carrier screening for cystic fibrosis: a systematic review of 23 years of research. Genet Med. 2014;16:207–16.
12. Shiroff J, Nemeth L. Public perceptions of recessive carrier testing in the preconception and prenatal periods. J Obstet Gynecol Neonatal Nurs. 2015;44:717–25.
13. Stern J, Salih Joelsson L, Tydén T, Berglund A, Ekstrand M, Hegaard H, et al. Is pregnancy planning associated with background characteristics and pregnancy-planning behaviour? Acta Obstet Gynecol Scand. 2016;95:182–9.
14. Tydén T, Stern J, Nydahl M, Berglund A, Larsson M, Rosenblad A, et al. Pregnancy planning in Sweden–a pilot study among 270 women attending antenatal clinics. Acta Obstet Gynecol Scand. 2011;90:408–12.
15. Backhausen MG, Ekstrand M, Tydén T, Magnussen BK, Shawe J, Stern J, et al. Pregnancy planning and lifestyle prior to conception and during early pregnancy among Danish women. Eur J Contracept Reprod Health Care. 2014;19:57–65.
16. Foster MW, Royal CD, Sharp RR. The routinisation of genomics and genetics: implications for ethical practices. J Med Ethics. 2006;32:635–8.
17. Kihlbom U. Ethical issues in preconception genetic carrier screening. Ups J Med Sci. 2016 Jul 8:1–4 [Epub ahead of print].
18. De Wert G, Dondorp W, Knoppers B. Preconception care and genetic risk: ethical issues. J Community Genet. 2012;3:221–8.
19. Scully JL. Disability and genetics in the era of genomic medicine. Nat Rev Genet. 2008;9:797–802.
20. Verweij M. Medicalization as a moral problem for preventative medicine. Bioethics. 1999;13:89–113.
21. Chattopadhyay S. 'Rakter dosh'—corrupting blood: the challenges of preventing thalassemia in Bengal, India. Soc Sci Med. 2006;63:2661–73.
22. Kingsmore S, Lantos J, Dinwiddie D, Miller N, Soden S, Farrow E, et al. Next-generation community genetics for low- and middle-income countries. Genome Med. 2012;4:25.
23. Lawson K. Perceptions of deservedness of social aid as a function of prenatal diagnostic testing. J Applied Soc Psych. 2003;33:76–90.
24. Tremain S. Reproductive freedom, self-regulation, and the government of impairment in utero. Hypatia. 2006;21:35–53.
25. Gregg R. "Choice" as a double-edged sword: information, guilt and mother-blaming in a high-tech age. Genet Med. 2013;15:482–3.