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ORIGINAL ARTICLE

Swedish healthcare providers' perceptions of preconception expanded carrier screening (ECS)—a qualitative study

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Abstract Reproductive autonomy, medicalization, and discrimination against disabled and parental responsibility are the main ongoing ethical debates concerning reproductive genetic screening. To examine Swedish healthcare professionals' views on preconception expanded carrier screening (ECS), a qualitative study involving academic and clinical institutions in Sweden was conducted in September 2014 to February 2015. Eleven healthcare professionals including clinicians, geneticists, a midwife, and a genetic counselor were interviewed in depth using a semi-structured interview guide. The questionnaire was constructed after reviewing the main literature and meetings with relevant healthcare providers. The interviews were recorded, transcribed verbatim, and content analyzed for categories and subcategories. Participants nurtured many ethical and non-ethical concerns regarding preconception ECS. Among the ethical concerns were the potential for discrimination, medicalization, concerns with prioritization of healthcare resources, and effects on reproductive freedom. The effects of implementation of preconception ECS, its stakeholders, regulations, and motivation are some of non-ethical concerns. These concerns, if not addressed, may affect the uptake and usage of carrier screening within Swedish healthcare system. As this is a qualitative study with a small non-random sample size, the findings cannot be generalized. The participants had little to no working experience with expanded screening panels. Moreover, the interviews were conducted in English, a second language for the participants, which might have limited the expression of their views. However, the authors claim that the findings may be pertinent to similar settings in other Scandinavian countries.

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Introduction

Preconception expanded carrier screening (ECS) is the process by which a couple tests for their carrier status of autosomal recessive (AR) diseases without having a positive family history (Castellani et al. 2010). If they prove carrier positive, there is a 25 % risk of them having a child with the autosomal recessive disease they both carry with every pregnancy. Though each recessive genetic disease is rare by itself, together they make up approximately 20 % of infant deaths and 18 % of total infant hospitalization. On average, every person has 2.8 AR mutations in their DNA that cause severe illnesses (Kingsmore 2012).

There is a distinction between preconception genetic screening, preconception ECS, and preconception genetic testing. The latter has been offered to couples with positive family history of a recessive genetic disease for many years, for example, sickle cell disease in USA. Preconception genetic screening among high-risk communities for severe recessive genetic disorders dates back to the late 1970s, e.g., Tay-Sachs among Ashkenazi Jews in USA and thalassemia major in Sardinia, both of which are voluntary programs. In some countries such as Iran and Saudi Arabia, the screening for autosomal recessive trait, in this case, thalassemia major, is obligatory by law. Religious authorities have also been involved in supporting preconception genetic screening programs, for example, in Cyprus, the Greek Orthodox Church requires a certificate from couples confirming that they underwent preconception screening for thalassemia to be approved for marriage (Zlotogora 2009). The Health Council of

the Netherlands in its report on preconception care recommended offering potential parents and/or risk groups, genetic screening for cystic fibrosis, and hemoglobinopathies preconceptionally (Health Council of the Netherlands 2007).

Lately, expanded screening tests have become more reliable, faster, and cheaper which possibly will allow governments to consider mass screening for carrier status with expanded panels among the general population (Kingsmore 2012; University Medical Center Groningen 2015). Recent technologies, such as next generation sequencing, devises test panels that can screen for up to 500 autosomal recessive diseases (Kingsmore 2012). Currently, direct-to-consumers private companies offer genetic screening panels to users (Borry et al. 2011).

This new approach, which is characterized by offering tests with large panels to the whole population without a prior risk or family history, raises specific ethical concerns. On one hand, it has been argued that preconception ECS increases couples' reproductive choices (for example, IVF, continuing with the pregnancy, adoption) and thereby enhances reproductive autonomy, or it can prevent suffering by potentially reducing the birth of affected children. On the other hand, there is fear of discrimination and stigmatization against disabled and carriers of diseases, and there is a risk of extending such discrimination to relatives of couples who opted out of screening (De Wert et al. 2011). This has been observed among Ashkenazi ultra orthodox community whose genetic carrier status stigma extended to their offspring though they may have not been tested. Some were shunned as potential spouses or are expected to pay a higher dowry in marriage (Raz and Vizner 2008). On a societal level, it is feared that the mass screening for recessive diseases may eventually precipitate eugenic practices (Scully 2008; De Wert et al. 2011). Parental responsibility and parental virtue have been discussed in relation to reproductive genetic screening and preconception care (Clarkeburn 2000; Wasserman 2005; Mcdougall 2007; Savulescu and Kahane 2009; Van Der Zee and Beaufort 2011).

The American College of Medical Genetics and Genomics (ACMG) in its policy statement, 2013 and a consensus document of American College of Medical Genetics and Genomics, American College of Obstetricians and Gynecologists, National Society of Genetic Counselors, Perinatal Quality Foundation, and Society for Maternal-Fetal Medicine 2015 recommended the need to screen for genetic diseases that are pertinent to the population where residual risk for negative screen parents is known. If not available, companies should update their residual risk information upon availability of such data prospectively (Grody et al. 2013; Edwards et al. 2015). Moreover, the tests offered should have a "validated clinical association between the mutation(s) detected and the severity of the disorder" (Grody et al. 2013). The consensus document advised preconception/prenatal expanded screening for traits that may lead to cognitive impairment, affect the quality of life, or require medical intervention (Edwards et al. 2015).

Recognizing the challenge of obtaining users' informed consent with expanded screening panel testing, the experts recommend users to undergo a pre-test educational session, where the common characteristics of the tested diseases and drawbacks of screening are explained. In addition, clinicians were encouraged to guide users to access more detailed information on diseases via other resources, for example, online resources, pamphlets, and videos (Grody et al. 2013; Edwards et al. 2015). Furthermore, post-test genetic counseling has been emphasized and education of both healthcare providers and patients has been advised (Edwards et al. 2015).

Moreover, the European Society of Human Genetics and EuroGentest have recently issued guidelines for assessment of next generation sequencing diagnostic testing, a technology expected to be used in expanded screening panels. The document includes guidelines pertaining to diagnostic and clinical utility, informed consent and information sharing to patients and physicians, distinction between research and medical care diagnostics, and reporting of results (van El et al. 2013; Matthijs et al. 2016).

It is noteworthy to mention that the context for this study is the Swedish healthcare system. Sweden has been a welfare country with tax-funded healthcare system. The policies that drive healthcare aim at "universality, equity, and gender equality." The county councils are in charge of delivery and funding healthcare services. This has started to change in the 1990s when "Swedish choice revolution" was introduced allowing county councils (21 in number) to hire private companies to provide healthcare services. In 2009, a new law was issued that permitted patient's right to choose their healthcare provider. Thus, only those healthcare providers that meet the requirements of users are selected and contracted. In 2010, private healthcare providers contributed to 10 % of total healthcare and 25 % to primary healthcare. However, there are variations in extent of privatization between the 21 county councils (Burström 2015).

National laws and guidelines emphasize that the prescription of care is dependent on patients' need rather than their socioeconomic status. However, this may come into conflict with local county governance, which is reliant on economics and profits (Fredriksson 2012).

There are very few studies evaluating the perspectives on preconception genetic screening with expanded panels. Examples are Cho and colleagues who examined genetic professionals' views on reproductive expanded carrier screening in six academic institutes in USA (Cho et al. 2013). Ready and colleagues assessed knowledge and attitudes toward expanded screening panels of participants in American Society for Reproductive Medicine 2010 and American College of Obstetricians and Gynecologists 2011 Annual Meetings

(Ready et al. 2012), and lastly Schoen et al. discovered discrepancy in expanded genetic screening usage between private and public patients in New Jersey (Schoen et al. 2014). What information to include in an informed consent for prenatal genetic screening was investigated qualitatively and quantitatively among pregnant women (Ormond et al. 2007, 2009).

To our knowledge, no studies on preconception ECS have been conducted in Scandinavia, which creates a knowledge gap we are hoping to bridge in this study.

The aim of this study is to explore and describe Swedish healthcare professionals' perceptions of preconception ECS with focus on the ethical aspects.

Methodology

The study had a descriptive qualitative design. Since this is a new topic that has not been researched in Sweden, we decided to use a qualitative method in order to explore in depth practitioners' views on preconception ECS.

Participants

Initially, individual informal meetings with a clinical geneticist and a gynecologist were carried out, which provided the basis for the selection of medical professions to include in the study as well as a preliminary list of healthcare professionals to contact for the study. The sampling was strategic, and inclusions were made following the recommendations given during the informal interviews and snowballing thereafter. In total, 18 healthcare professionals were contacted and 11 agreed to participate in the study. Individual in-depth interviews were conducted with healthcare professionals at major hospitals and universities in Sweden during September 2014–February 2015. The interviewees included three gynecologists, three obstetricians with subspecialty in fetal medicine, two clinical geneticists, one pediatrician, one genetic counselor, and one midwife (Table 1). We have not included any general practitioners (GPs) in our sample since they are not concerned with women and pregnancy-related health. In the Swedish Primary Healthcare, there is a women's clinic run by midwives and gynecologists/obstetricians, and they are the first line of contact with women for contraceptives, pregnancy, and gynecological concerns.

Data collection

The interviews were conducted in English by the first author. A Swedish translator was offered to help with the interview process but was declined by all interviewees. All of them had little or no problem conducting the interviews in English. The interviews lasted between 30 and 60 min and were all recorded. They were transcribed verbatim by a professional transcription company.

After reviewing the recent literature on ethical issues of new genetic reproductive technologies, the first author drafted an interview guide, which was revised by the second and third authors. The guide was semi-structured with open-ended questions and divided into four parts. The first set of questions explored the interviewee's background, and the second to last sections examined potential effects of preconception ECS program on couples/parents, healthcare system, and lastly the society at large (Table 2).

Analysis

The transcribed text was analyzed employing content analysis as described by Graneheim and Lundman (2004), and the focus was primarily on the manifest content to interpret the data. The authors inquired about ethical concerns related to reproductive technologies as presented in the literature (condensed interview guide Table 2). Nevertheless, participants were prompted to express their own views and concerns not necessarily raised by the interview guide. During the analysis, the focus was solely on the text generated by the respondents, which directed the process of analysis.

Initially, the transcripts were read through, meaning units were marked and preliminary codes assigned. Next, via a word processing document, meaning units were collected, condensed, and abstracted to codes, subcategories, and categories (Graneheim and Lundman 2004). The analysis process is described in Table 3.

The second author analyzed three interviews and together with the first author compared the results; any discrepancies were discussed and consensus reached. Furthermore, during the process of analysis, all authors met regularly, discussed any inconsistencies, and came to agreement regarding categories and subcategories. The final version of the analysis was agreed upon by all authors. All these measures were taken to assure credibility of data and analysis.

Ethical considerations

This study did not require ethical review clearance from a Research Ethics Committee according to Swedish regulations (Eriksson 2014; The Ministry of Education and Cultural Affairs 2003). However, the research process complied with international guidelines with regards to research involving human subject (WMA 2013). Participation was voluntary, and participants were interviewed after acquiring their informed consent. They were asked to participate via e-mail where an overview of the study was attached including the main aim

Table 1 Characteristics of participants

Specialty	Gynecologist	Obstetrician—fetal medicine	Clinical geneticist	Pediatrician	Genetic counselor	Midwife	Total
Number	3	3	2	1	1	1	11
Gender	Male		Female		Total		
Number	4		7		11		
Duration of practice		0–10 years	11–20 years		Above 20 years	Total	
Number of years professionals		5	2		4	11	

and assurance of confidentiality as well as a request of approval for recording the interview. The interviewees were free to terminate their participation anytime during the study.

Results

The analysis generated six main categories namely implementation of preconception ECS, medicalization, prioritization, discrimination, uncertainty, and reproductive autonomy, and under each, a couple of subcategories ensued (Table 4). It is noteworthy to mention that the categories and subcategories were not entirely mutually exclusive, and in few instances, some quotes/codes fell into more than one category. Except for the genetic counselor, midwife (who does research on prenatal genetic testing), and clinical geneticists, no one had experience of preconception genetic screening and the idea of expanded screening panels.

Implementation of preconception ECS

Under this category, the different aspects of implementing a preconception ECS program were grouped together such as who the stakeholders are, what motivates the implementation, the possible effects of preconception ECS, and lastly regulation of the process. The informants raised these ideas repeatedly during their interviews.

Stakeholders

Regarding stakeholders, respondents had varying views. Some thought healthcare professionals, such as pediatricians and clinical geneticists, should be in charge of preconception ECS implementation. Others considered, in addition to physicians, the involvement of politicians and prospective parents as well. One participant stated

> First of all I think the professions working with it should discuss it actually… to get some kind of consensus, what we think… and then again we have to discuss with politicians, how should the health care resources be shared… and then of course, it is good to know how future prospective parents think about this, what they want of course (informant 9).

Another respondent stated

> I think it's important that [healthcare] professions should decide this. It should not be private companies, wanting just to sell the test to make money (informant 2).

Table 2 Condensed interview guide

Section	Main questions
Background information	Specialty, duration of practice, awareness of preconception ECS.
Challenges to parents/couples	1.Do you think preconception ECS offer more reproductive choices?
	2.Do you think that preconception ECS would be "the responsible thing to do" for any couple thinking of getting pregnant?
	3.Do you think couples might see preconception ECS as "complicating an already natural process" such as pregnancy?
	4.Do you think if preconception ECS were to be offered as part of healthcare, would couples feel a pressure to be tested?
Challenges to healthcare system	5.What do you think would be the main challenges for healthcare professionals if preconception ECS were offered?
	6.What are the positive aspects for healthcare professionals if a preconception ECS program were to be done?
	7.What should we be screening for in preconception ECS? What kind of diseases?
Challenges to the society	8.What do you think would be the actual motives behind offering preconception ECS as part of healthcare system? What do you think of such motives? Why?
	9.Do you think governments should include preconception ECS as part of basic healthcare system? Do you think it is feasible to have such a program?
	10.Do you think implementation of a preconception ECS program would affect the society in any way? How?
	11.Do you think preconception ECS can precipitate eugenic practices?
	12. Do think preconception ECS may cause discrimination against the disabled?

Table 3 Example of the analysis process

I think a frequency of a disease is a good reason to put it on the screening, you know....because it will all depend on the frequency of the disease. If you have a low frequency disease, then it has to be very severe if you want to put it on the screening. If it is a high frequency, maybe you want to put it on the screening even though it is not lethal, for example. So I think that the frequency matters.	The frequently occurring diseases should be put on PCS panel even if they are not lethal. For low-frequently diseases, only the very severe should be screened.	Criteria of disease to test in PCS.	What should we test for?	Uncertainty
It is offered almost as a routine on the…at the antenatal clinic… mödravårdscentral…and it is built into the system as something…if not compulsory..it is almost a clinical routine even though it should not be offered like that but it is	Antenatal screen built into the system as clinical routine although it should not be offered like that	Risk of turning PCS into a clinical routine	Pressure to test	Reproductive freedom
You are healthy unit the age of 40, 50 years of age and in 40, 50 years of age it may be a cure for that disorder. So should we terminate pregnancies or avoid to become pregnant because of something that may not be a problem 50 years from now. That kind of problem I think, or if it is…let us say…some sort of increased risk for developing cancer later in life, how should we counsel patients regarding that if…once again if this problem is solved in 20 years time.	You are healthy till 40 or 50 years of age, a cure maybe found during the time, should we terminate pregnancy with such diseases?	Test for late (adult) onset disease (difficult boundaries)	What should we test?	Uncertainty

Effects

Informants raised issues regarding possible effects of preconception ECS, which can be grouped as effects on healthcare system and on families. Regarding effects on healthcare system, some stated that there would be minimal effect because only few parents will be both screen positive, while others indicated that it could cause overload on the healthcare system. This was negated by one respondent who stated

> Many of the cases we see today would not be here because they would not have that kind of problem anymore, so… it would be a relief for us to try to fix that or to have these kind of affected pregnancies. So for us in fetal treatment programs and so on, it will be… it would make our life easier as they would not… those patients… those unborn patients would not emerge anymore (informant 8).

Table 4 Results: categories and subcategories

	Categories	Subcategories
1	Implementation of preconception ECS	Stakeholders
		Effects
		Motivations
		Regulations
2	Medicalization	Striving for control
		Increased anxiety
		Shift of paradigm
3	Prioritization	Costs
		Health equity
4	Discrimination	Eugenics
		Stigmatization
5	Uncertainty	What should we test for?
		Interpretations of results
		Need for information
6	Reproductive freedom	Pressure to test
		Responsibility

More than one participant indicated the effect of preconception ECS on possibly decreasing incidence of abortion, as one respondent stated

> I think it is a very good thing because you can prevent to do an abortion due to the prenatal examination showing something wrong, then if you can already check this up before the pregnancy, I think it is a very big pro (informant 6).

Other potential effects stated were a need for more genetic counselors as well as an increase demand by parents for prenatal diagnosis and pre-implantation genetic diagnosis.

For families, potential effects of preconception ECS implementation would be "*decrease suffering of children*" and "*starting a family will be better planned in the future.*"

One respondent added

> One positive thing might be fewer babies born with severe illness that die in the hospital, you see, problematic period for the parents and so on. This could be a positive thing (informant 1).

Motivations

Respondents expected different political motivations for implementing preconception ECS, the recurrent one being reducing healthcare cost, even if politicians would not state this as an actual motive. Reducing suffering, demands from parents, decreasing incidence of abortion, and conforming to Scandinavian or EU guidelines were other motives for implementation as mentioned by participants.

One participant said

> If expecting parents will ask for it I think that is a very important thing. And also, if it were implemented abroad, people would read about it. But then again…

> we have to discuss why. Should we do it just because we can and because people are asking for it? I think there must be clinical relevance, there must be an advantage of it and one advantage is of course that you could decrease invasive, prenatal testing… because, there is a risk of miscarriage (informant 9).

Regulations

If preconception ECS were to be implemented, interviewees indicated the need for regulations, as one interviewee stated

> This is good even though there must be a very rigorous framework, I think. And it must be… if one offers this, it must be relevant conditions that you screen for (informant 8).

Another respondent said

> Well, I think we often implement methods before they are evaluated in a serious way. So that is my problem (informant 4).

Medicalization

Many participants expressed worry that preconception ECS would increase medicalization of life and parenthood. These worries have below been categorized into how preconception ECS would let individuals "strive for control" and how it may trigger "a shift in paradigm" in the sense that preconception ECS may alter the view on life, for example, from a non-genetic to a genetic perspective. A third subcategory generated was increased anxiety of potential parents.

Striving for control

Respondents viewed preconception ECS as a way for parents to take control of their lives, a notion viewed by some in a negative way, and attributed to a certain social class. One healthcare professional said,

> You feel sadness because nowadays people are very concerned about having control of everything. There is a lot of well-educated people living in the city who do not want anything to let go to their destiny. They want to have control. This is a sign of the new society with well-educated people that do not let nature have it its own way (informant 10).

Another participant's view

> I think that: what is next then? …I mean there is no way we can avoid difficulties in life. I mean we cannot just test away [eradicate the risk of] everything, so you cannot have a Preconception ECS and then you will be so sure that your child will not be suffering from life adversities because he can be hit by a car at the age of one or maybe he is …bullied in school or [has] learning disabilities. I mean…. we cannot test away everything (informant 2).

Increased anxiety

Some respondents mentioned the potential of preconception ECS to cause anxiety among parents because they might discover some aspects of themselves they did not know, such as being a carrier of a genetic disorder. Or, parents might worry about the decisions to make when they are carrier of a non-severe disease.

A respondent mentioned

> that will definitely lead to that we will be more aware of diseases and problems, you know… suddenly you got a lot of information about disorders and diseases you did not even know about and heard about… that maybe you can be more pathological in your thinking, you know like worried about things, maybe (informant 11).

Another stated

> How can you explain to this individual what consequences this will have in the future or in a child to be. I think… this sound to me as very difficult and it sounds to me that it might cause a lot of worry because you have some information about yourself that you do not… probably, all of us have some problems with our genes in some way (informant 6).

Shift of paradigm

In addition, respondents expected a shift of paradigm with preconception ECS. For example, with regards to choosing a partner instead of proclaiming love, a partner has to possess a suitable "*genetic certificate*" on the "*dance floor*" (informant 8). Some went as far as calling the implementation of preconception ECS programs as "*science fiction*" (informants 1 and 6).

As one interviewee said

> [It is] the start of kind of a genetic revolution and there will be so many other things that you will know about

your genome in the future, and a lot of choices and things that will depend on that (informant 11).

Another reiterated

I do not know really but in my primary thoughts… I think this is a little science fiction. I think it is a little strange building of a society when you have to schedule [screen] all genetics of all young people. So, I think it is a little scary thought for me (informant 6).

Prioritization

Respondents were concerned about the cost of preconception ECS and whether its implementation may affect the budgeting of other healthcare areas. Many indicated that preconception ECS should not be a priority and that money should be geared toward more vital diseases such as cancer. Under this category, two main subcategories were grouped: cost and health equity.

Cost

Many respondents were doubtful that preconception ECS would save money for the healthcare system. They believed that it would be expensive and laborious.

One participant said

If it is a disease, one per thousand… then I really wonder, is it a good thing to do… with in mind everything else we have to do within the health care. We have to take care of our elderly people and we have to take care of people with cancer. Is this then the thing we shall focus on and what is the pros and cons and what is the costs for both economical and individual (informant 6).

Another informant stated

In a state or in a health care system where you have to reduce the costs, I can find thousand other alternatives to reduce costs but politics will never take that to a priority list, but reducing these very unusual genetic diseases… I think it is no….no way to go. No way to save money, no (informant 3).

Health equity

Few informants were worried that preconception ECS would precipitate a health equity problem since those who can afford it will buy preconception ECS from private companies while those who could not will not be able to access it through healthcare creating a gap between the have and the have not.

One respondent declared

I think society should be proactive because it [Preconception ECS] will happen anyway and if the society is not proactive people will start buying these services and then we will have even worse than discrimination, we will have segregation; the ones who can pay for it and the ones who cannot pay for it. Healthcare should be equal, it should not depend on who you are and where you come from and how much you know and how much money you have. You're entitled as good healthcare as anyone (informant 2).

Another participant said

…that some people are paying on their own for these tests and someone cannot afford it, then it's not a really good situation. It is a situation that we do not see that much in Sweden. It is not equal healthcare in some way if you can call this healthcare. So then it will be better if the healthcare offer this (informant 1).

Discrimination

This category addresses the potential of preconception ECS to create discrimination against individuals; it was a way to "*sort out people*" either with diseases or carrier status or parents who opt not to undergo preconception ECS. One respondent was worried that it can precipitate discrimination against minorities where some diseases are more common than in the general population. Under discrimination, two subcategories were clustered: eugenics and stigmatization.

Eugenics

When asked about eugenics, several informants confirmed the risk for it if preconception ECS was to be implemented on a population level while few did not agree. The reason being, many of the traits are not monogenic and therefore you cannot screen for them, for example, IQ or height. Some claimed that it is not a risk in modern Sweden; however, one informant stated

In international politics nowadays, a lot of fascists and extreme right wing/nationalistic parties are coming up. Though it is something that seems to be very far away and belonging to history, it's now being a reality again. I don't see it right now in our country but you always have to not forget about that (informant 11).

Among those who agreed that preconception ECS could precipitate eugenics, one informant said

> You cannot say there is no risk for any Eugenic practices, of course there is such a risk, like with any genetic information it is very sensitive information about individuals that you have to think about how you are going to test and treat that type of information (informant 11).

Stigmatization

Generally, respondents did not make the distinction between stigmatization and discrimination except for one respondent who distinguished between the two, the former being "*they see you as one kind of person, for example that you live in a breast cancer family*" but discrimination has more "*negative value*" in it "*for example, it is harder for you to get a job*" (informant 9).

Uncertainty

According to respondents, there were many issues that pertained to uncertainty with regards to preconception ECS. To many of the interviewees, preconception ECS was a new approach they never heard of. They had queries regarding the type of diseases to test for and interpretations of preconception ECS results, and many acknowledged the general need for more information, all of which make the subcategories for this category.

What should we test for?

Deciding on what to test in preconception ECS proved a challenge to many respondents. Some agreed to focus on only severe and lethal diseases, while others had difficulty in defining severe diseases. For example, should severe diseases include high incidence autosomal recessive diseases such as cystic fibrosis (CF)? To some, CF was deemed severe because of its relative high prevalence in Sweden, though patients can lead a relatively good quality life until their middle age.

One respondent said

> I think it is difficult because I think it is relative, because different diseases may mean different things to different families and different children (informant 6).

Another respondent explained

> I do not think this is a pure medical issue. Some parents cope very well with having a disabled child and some are horrified if they find out that their baby is suffering from club feet for instance and terminate the pregnancy due to that, or a cleft lip or whatever and I do not think it is that easy to say what is relevant for each individual… but still, I think society has to have some sort of standard, yes (informant 8).

There were fears of "slippery slope" and worries of where to "draw the limit" as to what preconception ECS screens for, for example, screening for adult onset diseases or cancers may not be acceptable. To some respondents, the extent of adversity to the family's and affected child's quality of life were determining factors in what diseases preconception ECS should screen for.

Interpretation of results

Some respondents, particularly clinical geneticists, were especially worried about interpretation of results. They agreed that concentrating on monogenic, well-described AR diseases is the best way, because other methods, such as wide genome sequencing, would generate wealth of information, much of which has no clinical utility and may cause confusion.

As one respondent explained

> [If] you just sequence all the severe recessive genes, you would find a lot of variants that you do not know how to interpret [if they are pathological or not]. Or if you just test for known, severe mutations that have already been described in children with these disorders [only known pathological]. So I think that depends on how you do the testing, because if you screen for anything you would find a lot of things that you do not know what it means (informant 5).

Another respondent was more specific and explained

> You can design the test so that you can only find mutations that have been described and we know for sure they are mutations. But you can also design tests that you see everything in the genes and then you can also find gene variants, genetic variants within the gene that you cannot say for sure if it is deleterious mutations or if it is an uncommon normal variant, then you have problems. Because if you find many of those you will most likely, confuse or scare the patient: [so] we found something [in the woman] that we do not know [if it] is a mutation or not and [the] husband is a carrier of a certain mutation that we know is pathogenic, maybe they feel they are forced to do prenatal test (informant 1).

Need for information

The need for information was almost a unanimous request. There is a need for information to educate healthcare

professionals on preconception ECS and potential parents to make informed decisions. Also, information regarding communicating risk to couples was brought up by the informants and was viewed as challenging.

As one respondent summed it up

> Information, definitely, first before the test information… what should you have, what is the aim of the testing, what is the use of it, how useful is it, and a follow up after the test (informant 6).

Few questioned the feasibility of imparting large amount of information about screened diseases to potential parents. Others queried who should give the information, gynecologists or midwives, or a specific preconception clinic, and when the information should be given. Some called for research to assess whether preconception ECS should be part of basic healthcare and the societal acceptance of it.

As a participant said

> I think you have to do some kind of research project, to test it in a society and see what the effects will be and what people think about it… and I think you have to evaluate it also from an ethical view (informant 4).

Reproductive freedom

During the interviews, the word "autonomy" has not been presented except occasionally; however, respondents in many other words expressed the notion, either by the use of words like "*informed decisions*" or "*reproductive choices*" or "*reproductive freedom*." Many agreed that preconception ECS may increase reproductive choices; nevertheless, some respondents believed that the opposite would happen because if parents test positive, their "*choice to become a parent together with that woman was taken away.*" Under this category, informants raised the potential for "*pressure to test*" and the notion of responsibility.

Pressure to test

Some informants were concerned about the potential for pressure to test, either by the society's expectations or the view that preconception ECS is a recommendation of healthcare professionals. One respondent's view on pressure to test was

> Yea in some way yes, when you are offering something to the public so just by offering there might be small but significant push you should have this (informant 1).

Another respondent added

> In my perception, it is not that they feel pressured to be tested, but they think it is a good idea since it is offered by the health care system, so in that case it has to be something that is recommended and good. So often they do not think through it a lot on beforehand,/—/I will have it because someone thinks it is good, and they come here without understanding what they really accepted (informant 8).

Responsibility

Participants brought up the notion of responsibility in relation to preconception ECS, and one participant speculated the type whether it is responsibility toward society or future child or toward healthcare system. Assigning responsibility upon prospective parents to undergo preconception ECS may be viewed as a form of "compulsion" as one respondent stated

> No, I think that it should not be some kind of responsibility for the couple, it should be a voluntary offer, nothing else. It must be some kind of individual decision and cope with your personal ethics (informant 4).

Another interviewee stated

> I mean, we are not responsible and so I would not like to increase the burden of being responsible in the society. I think there should be also room for unplanned and spontaneous pregnancy and irresponsible behavior because in most cases these parents turn out to be as good parents as any other (informant 2).

Discussion

The respondents raised many concerns both ethical and non-ethical regarding preconception ECS for couples at a population level. Such screening programs have not yet been implemented in Sweden. The findings have been grouped into concerns with implementation of preconception ECS, medicalization, prioritization, discrimination, uncertainty, and reproductive freedom.

There are only few studies that examined expanded genetic screening outside Scandinavia, and none have addressed the ethical concerns perceived by healthcare professionals if used preconceptionally. Therefore, there are only few studies to compare our results to. This gap of knowledge needs to be managed with further research as recently emphasized by conjoint statement of the American College of Medical Genetics and Genomics, American College of Obstetricians and Gynecologists, National Society of Genetic Counselors,

Perinatal Quality Foundation, and Society for Maternal-Fetal Medicine (Edwards et al. 2015).

One of the advantages of preconception ECS, as stated by The Health Council of the Netherlands, is that it allows parents to make reproductive decisions before a child is conceived, unlike prenatal and neonatal genetic screening currently in place. Prenatal screening provides an option for women to abort an affected fetus, while in neonatal screening, the child is already born (Health Council of the Netherlands 2007). In a study surveying gynecologists and obstetricians Fellows of the American College of Obstetricians and Gynecologists, two thirds of respondents indicated that the best time to use expanded carrier screening should be preconceptionally when a couple is planning a pregnancy (Benn et al. 2013). This was reiterated by respondents in our study who held the opinion that preconception ECS may reduce abortion incidence. In addition, participants also stated the potential for reducing workload off some physicians, for instance fetal medicine professionals and decrease in number of births of children with severe ailments.

Our results showed that healthcare professionals fostered ethical concerns regarding preconception ECS, which have been already discussed by other commentators (De Wert et al. 2011; Borry et al. 2011) such as medicalization, reproductive autonomy, discrimination, and eugenics and newly arising issues such as prioritization of health resources, health equity, uncertainty, and issues with the implementation of preconception ECS.

Respondents indicated that finding resources to pay for preconception ECS would be expensive and burdensome for the Swedish healthcare system. This comes in contrast to results of a focus group study examining US genetic professionals' views on expanded screening, where it was perceived as "great financial value" compared to monogenic screening (Cho et al. 2013). The reason can be that Sweden has a comprehensive healthcare coverage, and if such programs are implemented, it is expected to be government funded, whereas in USA, they are out of users' pockets, and therefore, offering extended screening panels could be more cost-effective for them. In addition, respondents in our sample presume that preconception ECS is cost-effective only if mathematically its price is less than the overall cost for taking care of people currently existing with diseases being screened for.

Currently, private companies are offering as well as determining what genetics diseases to include on screening panels for users in the USA, which are not necessarily compliant to professional guidelines (Langlois et al. 2015). Moreover, there is some evidence of discrepancy in the prescription of expanded screening panels between private (almost 10-fold higher) and public doctors (Schoen et al. 2014). The commercial offering, on one hand, may precipitate a health equity problem since only those who can afford the screening test buy it. On the other hand, it creates an extemporaneous model of healthcare policy development, where commercial companies, users, and legal practice direct the financial compensation and operation of healthcare, which, in turn, shape the standard of care. In contrast, the evidentiary model is based on evidence from research assessing clinical and normative issues resulting in harmony between the public and professional views. As such, standard of care is defined and this shapes the financial compensation and operation of healthcare (Wilfond and Nolan 1993).

Respondents in our study have raised concerns regarding both health equity and the role of private companies in preconception ECS. Moreover, they emphasized the need to properly evaluate such programs before implementation with regards to purpose, potential benefits and risks, relevance and acceptance in Swedish society, and overall cost. And, it should not be driven by political agenda or commercial benefits. The stakeholders of preconception ECS implementation should primarily be healthcare professionals and then users and politicians. Commercial companies have not been mentioned as a stakeholder, though they are manufacturing the screen panels for preconception ECS. It can be argued that the interviewees in our study preferred an evidentiary model of health policy development.

Informants expressed uncertainty concerning which diseases to test for and interpretation of results and emphasized the need for information for healthcare professionals and users. These same issues have been reiterated in other empirical and non-empirical articles (Borry et al. 2011; Cho et al. 2013; Benn et al. 2013; Edwards et al. 2015). For many reasons, adequate information is crucial in preconception ECS. Firstly, it can guide the decision on what diseases to screen for, how to interpret the results, and lastly allow couples to make informed reproductive choices, an integral component of reproductive autonomy.

One of the challenges in preconception ECS is informed consent, as expressed by respondents. Guidelines and policy statements have given some direction as how to handle the consent process and form (Grody et al. 2013; Edwards et al. 2015), which do not necessarily reflect users' preferences. Empirical studies (focus group and survey studies) inquiring about patients' preferences for information to constitute consent forms for expanded carrier screening showed their need for comprehensive information about the test and diseases being tested (Ormond et al. 2007; Ormond et al. 2009).

Medical supervision of the whole process and genetic counseling pre- and post-testing were recommended by participants in this study as well as professional guidelines (Grody et al. 2013; Edwards et al. 2015). One reason is to combat anticipated parental anxiety since it is expected that a parent will be a carrier of some severe autosomal recessive trait (Kingsmore 2012). In addition, with expanded screening panels, there will be information overload about diseases, some of which are very rare (Grody et al. 2013).

There have been philosophical debates on parental responsibility and whether partaking in screening programs and preconception care is considered a parent's responsibility (Savulescu and Kahane 2009; Van Der Zee and Beaufort 2011). The respondents in our study reasoned differently, as some respondents expressed that assigning responsibility upon parents to undergo screening might pose a threat to their reproductive autonomy. In addition, seeking to have more control and create a "perfect society" is not realistic and can predispose to discrimination and non-acceptance of the disabled. Though the informants were in favor of reducing harm to the severely affected children and affirmed a certain level of life quality to children-to-be, they were reluctant to screen for moderately severe and adult onset diseases. This has also been discussed by Clarkeburn (2000) and Wasserman (2005).

Though the study identified many issues to keep in consideration, some matters were raised in the literature but not addressed by participants, such as how to handle incidental findings (Juth and Munthe 2011), sharing of genetic status with family members, and offering cascade screening (where genetic screening is offered for proband members of family) (De Wert et al. 2011). Moreover, Scully (2008) cautioned of diminished appreciation of genetic diversity that results in variable phenotypes, an aspect that was not raised as a concern by respondents in our study. Hence, further research, both empirical and philosophical, is needed in these areas. In addition, further quantitative studies of healthcare professionals are required in order to identify whether our results can be generalizable.

The study also reveals further need for quantitative and qualitative research to examine prospective parents' and politicians' views of preconception ECS.

Discussion of the methodology and limitations

To ensure trustworthiness of the analysis, the aspects of credibility, dependability, and transferability of data were considered in our study (Graneheim and Lundman 2004). For credibility, we interviewed healthcare professionals of various genders and work experience. We focused on medical specialties, which are likely to encounter couples requesting preconception ECS or are directly involved if preconception ECS programs are implemented, as advised during the initial informal meetings mentioned above. In addition to different specialties and gender, our sample includes healthcare professionals who have been practicing their profession for varying durations. Moreover, we have chosen to do qualitative in-depth interviews because the topic of our research is new and has not been examined among Swedish healthcare professionals. Data saturation was reached for the aspects addressed in the paper, and further interviews generated no new concepts or ideas (Guest et al. 2006).

According to Graneheim and Lundman, the concept of dependability is part of trustworthiness of data and refers to the extent of change in the data over time either due to expected modifications done by researchers during the process of interviewing or during the analysis (Graneheim and Lundman 2004). Our data was collected within 6 months and analysis done in 3 months (February to April, 2015). Both durations seem reasonable, and there was little change in the interview guide. Any inconsistencies with regards to categories and subcategories were discussed, and consensus reached via open dialog between researchers.

The last aspect of trustworthiness is transferability and denotes the degree by which results can be transferred to other data sets (Graneheim and Lundman 2004). We believe that the findings are pertinent to similar settings; however, since this is a qualitative research with a non-random and small-sized sample, the results cannot be generalized. In addition, interviews were conducted in a foreign language, so there is a risk of respondents not expressing themselves as easily. We do claim, however, that our study has captured important aspects of the ethical concerns regarding preconception ECS among healthcare providers in settings similar to the Swedish context.

Conclusion

Ethical issues raised in the interview study include medicalization, concerns with reproductive autonomy, parental responsibility, discrimination against diseased/carrier status, prioritization of health resources, and uncertainty concerning what to test for and how to interpret the results.

This study gives insight to some of the concerns, both ethical and non-ethical, of Swedish healthcare professionals with regards to preconception ECS. Though such programs have not yet been implemented, the study attempts to identify important aspects in relation to preconception ECS and the associated bioethical aspects before they are put in place, so the concerns can be addressed or taken into consideration during the implementation process, should it take place.

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Author's roles A. M.: main contributions to conception, design, acquisition of data, analysis, and interpretation of data; drafting the manuscript and revising it, and final approval of the version to be published.

U. K.: assist with the conception and design. Revising it critically for important intellectual content, and final approval of the version to be published.

A. T. H.: contributions to the design, assist with analysis and interpretation of data; revising it critically for important intellectual content, and final approval of the version to be published.

Compliance with ethical standards

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Conflicts of interest Author AM declares no conflict of interest. Author UK declares no conflict of interest. Author ATH declares no conflict of interest.

Ethical approval This study involved human participants. All procedures were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards.

Informed consent Verbal informed consent was obtained from all individual participants included in the study.

This article does not contain any studies with animals performed by any of the authors.

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