Vulnerable populations and genetic disorders: a socio-science approach to the application of genetic technology in China
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您好！我是睢素利，荷兰阿姆斯特丹大学社会科学研究院的博士研究生。这个问卷涉及的是我目前的研究课题。我非常感谢您阅读和回答我这个问卷。您可以自愿参与，自愿回答问题，您可以随时放弃回答问题。所有的问题都列出了可选择的项目。答案没有对错之分，您可以根据自己的情况和自己的判断给出您自己的答案。问题假设了一些不好的情况，希望您可以谅解，也希望不会给您带来不悦。非常感谢您阅读和回答我的问卷！如果您觉得不便，您可以不回答那些您不愿意回答或者您觉得不便回答的问题。非常感谢您！

I am Suli Sui, PhD candidate of ASSR Amsterdam University. I would appreciate it very much that you read and answer my questions, which are for my current research. All the questions are optional. There are no wrong or right the answers. What I want is your own opinion based on your experiences and conditions. You can choose more than one answer and you can refuse to answer the question/questions if you do not want to answer. You can withdraw this questionnaire or end the interview at anytime you wish. In some questions, certain situations are supposed. I hope you do not mind

46 The questionnaire/interview outline was conducted in China using the Chinese version. The English version was translated by the author.
these supposed unpleasant situations. Thank you so much!

时间 time: ____________________________
地点 location: ________________________________

访谈对象基本信息 interviewee personal information:

* 您的年龄 age________
* 性别 gender________
* 职业背景 professional background________________________
* 教育背景 educational background __________________________
* 长期住处 大城市/小城市/县城/农村 live place of interviewee big
city/small city/county/ village________
* 您是否生育过 是/否 birth experience yes / no 如果有，您有____个孩子
（____男孩____女孩） if yes, number and gender of your children, children
boy______ girl____
* 您/您妻子是否有过终止妊娠的经历？ (your/your wife or partner) abortion
experience yes/no ______次?(if yes, the number of abortions)________
* 如果您（或者您妻子）在孕期，第几次怀孕________怀孕多久了？_____
If you/your wife are pregnant, this is the ___ week of my ___ pregnancy”?

第一部分：遗传咨询和检测

Part One: Genetic counselling and genetic testing
1. 您知道或者听说过遗传咨询吗？
   A. 听说过，知道一些
   B. 只是听说过这个词，具体是什么不是很清楚
   C. 没有听说过

1. Are you familiar with or have you heard of genetic counselling?
   A. I know something about it.
   B. I have heard this word, but do not understand it clearly.
   C. I have never heard of it.

2. 如果您知道或者提说过遗传咨询，是从什么渠道得知的？
   A. 听其他医生介绍
   B. 网络报纸媒体
   C. 亲戚朋友病友等介绍
   D. 其他介绍

2. If you already knew of or have heard of genetic counselling, where did you get the information?
   A. From the local doctors.
   B. Website on Internet, newspaper, TV and other media.
   C. From friends, relatives and patients.
   D. Other.

3. 您觉得什么样的人应该进行遗传咨询？
   A. 有家族遗传病史的
   B. 生育过有病的孩子的
C. 有多次自发或者不明原因的流产的
D. 大龄的准备怀孕的夫妇
E. 为了优生目的育龄夫妇都应该进行

3. What kind of person do you think should apply for genetic counselling?
A. Those with a family history of genetic disorder.
B. Those who have had experience with an affected child/children.
C. Those who have experience of spontaneous abortion.
D. A couple prepared to have child at the age of more than 35.
E. For the sake of eugenics, all couples who want to have a baby.

4. 您觉得遗传咨询是：
A. 为有遗传病的家庭提供咨询
B. 咨询关于遗传病的知识
C. 寻求医治遗传病的方法
D. 确诊某种遗传病
E. 为有遗传病的家庭提供生育指导
F. 为有遗传病的家庭提供心理支持
G. 介绍和提供遗传检测

4. What do you think genetic counselling is?
A. Counselling for families with a history of genetic disorder.
B. Consultation for genetic knowledge.
C. Seeking a way to medical treatment.
D. The diagnosis of a genetic disorder.
E. Reproductive advice for a family with a history of genetic disorder.
F. To offer Psychological support for a family with a history of genetic disorder.
G. To introduce and offer genetic testing.
5. 如果您曾经看过遗传咨询门诊，您觉得您咨询给您的收获是

如果您没有，您认为您期望得到

A. 知道了疾病的遗传特征
B. 知道了哪里可以确诊
C. 了解到一些关于治疗的信息
D. 得到为病人进行检测的建议
E. 得到关于家族其他成员进行检测的建议

5. If you have visited a genetic counselling clinic, what is the main result you think you have gained? ________ If you have no such experience, what do you expect from the genetic clinic?

A. To understand the inherited character of the disorder.
B. To know where to get access to the diagnosis.
C. To gain information about its medical treatment.
D. To get advice about undertaking testing for the patient.
E. To get advice about undertaking testing for family members.

6. 听说过基因检测吗？

A. 没有听说过
B. 只是听说过这个词，具体是什么不是很清楚
C. 听说过，大概知道是检测基因的
D. 听说过，知道是检测遗传病基因的

6. What do you know about genetic testing?

A. Nothing.
B. I have only heard the word, but do not understand it clearly.
C. I have only heard a bit, but I think it tests genes.
D. I know it tests for genetic disorders.

7. If you have heard of genetic testing, were did you get the information?
A. Doctors (progestational consultation).
B. Website, newspaper and other media.
C. Relatives and friends.
D. Friends who have been patients.

8. Did you ever think to take a predictive genetic testing to know the risk for a certain illness (suspicous position testing)?
A. I never heard of it.
B. I think it is ok to have a try, but did not try it myself.
C. I have heard of it, but I don’t know what it is.
D. I think it is not helpful and do want to take it.

9. If you are a pregnant woman, and you know about the prenatal diagnosis/prenatal genetic testing, would you ask for the testing initiatively?
A. Yes.
B. No.
C. Maybe yes, maybe no, do not know yet.

10. What are the reasons you asked to undertake the test?
A. I have a family history of genetic disorder.
B. To feel relieved.
C. I think there is no harm and it may be good.
D. It would be a heavy burden to have an affected child.

11. You think you would not or don't want to ask for the testing initiatively. What is the main reason?
A. Price is too expensive
B. To feel relieved.
C. 觉得检测结果也不一定可靠，会增加心理压力

D. 觉得生孩子还是顺其自然的好

E. 自己也没有家族遗传病史，没有必要检测

11. What are the reasons you do not ask to undertake the test?
A. The price is so high.
B. I feel the technology is not very reliable.
C. I feel the result is not reliable and adds psychological burden.
D. I feel it is better to follow ‘nature’.
E. I do not need the test because I have no family history.

12. 如果您自己不知道或者不会主动要求进行检测，您的医生给您介绍了产前诊断/产前基因检测的技术（没有建议您做），您可以自由选择做或者不做，这种情况下，您会怎样：
A. 自己决定选择做或者不做
B. 和丈夫商议，听丈夫的意见
C. 和父母商议，听父母的意见
D. 问医生，听医生的意见
E. 和家人商议，但觉得更信赖医生的意见
F. 了解其他就诊的孕妇有没有做，如果很多人都做了，那自己也就选择做，如果很多人都没有做，自己也就不做了
G. 和家人商议，共同做决定

12. If you want to undertake genetic testing, how will you make the final decision?
A. I will make the decision myself.
B. I will consult with husband, and follow my husband’s opinion.
C. I will consult with my parent, and follow my parent’s opinion.
D. I will ask the doctor, and follow the doctor’s advice.
E. I will consult with the family, but feel the doctor’s opinion is more reliable.
F. I will ask other pregnant women to see if many took it, and then follow the majority.
G. I will consult with the family, and make the decision together.

13. If you had decided to take test, will you or ask the doctor to explain the potential risk and that some unexpected results may brought by the test?
A. Yes.
B. Maybe, but not sure.
C. If the doctor did not mention them, maybe I would not think to ask.
D. If the doctor did not mention them, maybe I would not inquire.
E. I would not ask. I feel the doctor will explain if there is something that should to be explained then. There is no point to ask for more.

14. 假设您准备生育，下面的何种情况您会考虑进行产前基因检测
A. 知道家族有遗传史
B. 知道自己是携带者（做过携带者测试）
C. 已经有一个发病的孩子
D. 医生建议（建议的原因_________________）

E. 知道有检测技术，自己希望了解自己的遗传信息

14. Suppose that you are preparing to conceive. What conditions might cause you to apply for the prenatal genetic testing?
A. Having a family history of genetic disorder.
B. Already knowing that I am a carrier (have taken the carrier test).
C. I already have an affected child.
D. Doctor’s advice (the reason for this advice ________________).
E. I hope to know more about my genetic information.

15. 假设您准备生育，并且觉得自己是需要检测的情况 费用问题会使您不愿意进行检测吗（DMD 确诊或者携带者检测，一份血样 150 元，一般至少需要父母母亲和孩子的血样；产前检测 2000-3000 元）：________
A. 不会
B. 觉得应该不会吧
C. 需要考虑一下费用
D. 觉得费用贵，但可以负担
E. 费用贵，但贵也愿意负担检测费用

15. Suppose that you are preparing to conceive, and you consider that you should to take the test. Will the price then be an obstacle to the testing (for example, DMD prenatal genetic testing will cost about ¥2000 to ¥3000)?
A. No.
B. I feel it will not.
C. I think the price will be considered.
D. I feel the price is high, but can afford it.
E. The price is so high, but I would like to afford.
16. 假设您准备生育，并且觉得自己是需要检测的情况，检测技术问题会使您不愿意检测吗？对检测担心吗？
A. 对技术不是很担心，期待好结果
B. 有些害怕不好的结果
C. 不知道检测的准确性，还是有些担心，害怕万一有误

16. Suppose that you are preparing to conceive, and you consider that you should to take test. Will you then worry about the technology as an obstacle to testing?
A. I do not very worry much about the technology and expect a good result.
B. I worry about a bad result.
C. I worry whether the result will be accurate, and am afraid of the risk of a mistake.

17. 假设你是需要检测的情况，丈夫怎样看待您要做的检测或者你觉得丈夫会怎样看待您要做的检测？
A. 强烈支持
B. 支持
C. 不反对

17. Suppose that you think you should have the test. How might your partner respond? Or what do you think the response might be?
A. I think I would be strongly supported.
B. I think I would be supported.
C. I think I would not be opposed.

18. 假设你是需要检测的情况，家人怎样看待您要做的检测或者你觉得家人会怎样看待您要做的检测？
A. 强烈支持

B. 支持

C. 不反对

18. Suppose that you think you should have the test. How might your family respond? Or what do you think their response might be?
A. I think I would be strongly supported.
B. I think I would be supported.
C. I think I would not be opposed.

19. 如果检测结果显示胎儿是某种遗传疾病的携带者（比如地中海贫血，DMD严重性肌营养不良），不知道胎儿性别，您会选择：_______

A. 终止妊娠

B. 继续妊娠

19. Suppose that the test result shows that the foetus is a carrier of a hereditary disease (for example: thalassaemia, Duchenne muscular dystrophy DMD), and you do not know the gender of the foetus. Would you:
A. terminate the pregnancy?
B. continue the pregnancy?

20. 如果检测结果显示胎儿是某种遗传疾病的携带者（比如地中海贫血，DMD严重性肌营养不良，并且您知道胎儿是男胎，您会选择：_____

A. 终止妊娠

B. 继续妊娠

20. Suppose that the test result shows that the foetus is a carrier of a hereditary disease (for example: thalassaemia, Duchenne muscular dystrophy DMD), and you already know the foetus is male. Would you:
A. terminate the pregnancy?
B. continue the pregnancy?

21. If the test result shows that the foetus is a carrier of a hereditary disease (for example: thalassaemia, Duchenne muscular dystrophy DMD), and you already know the foetus is female. Would you:
A. terminate the pregnancy?
B. continue the pregnancy?

22. If the result of prenatal genetic testing shows that the foetus is affected by DMD, are you sure you will abort the foetus?
A. I certainly will. The purpose of taking the test is to avoid having an affected child, so I would not keep an affected foetus.
B. I will, but will feel sad about it.
C. I will feel very sad and reluctant to do it, but feel there would be no choice.

23. If the prenatal test result is positive for DMD carrier (girl), what will you do?
A. abort, because I want a healthy child
B. 流产，为将来着想，孩子要结婚生育，不希望孩子面临困境和压力

C. 流产，希望疾病在自己家族消失

D. 保留，携带者不会发病，孩子是健康的

E. 保留，孩子是健康的，将来可以通过检测要健康的孩子

23. If the result of prenatal genetic testing shows that the foetus is a DMD carrier (in this condition, the foetus is female), what will you do?
A. I would have an abortion, as I want to have a healthy child.
B. I would have an abortion, as I do not want to the child face difficulties in the future.
C. I would have an abortion, and hope that the disease disappears from the family.
D. I would continue with the pregnancy, as the carrier is healthy.
E. I would continue with the pregnancy, as the carrier is healthy and I could later have a healthy baby using the testing in the future.

24. 如果没有产前检测或者检测结果不能确定胎儿有没有遗传疾病，胎儿有 50% 的概率患有某种遗传疾病，您会选择：

A. 终止妊娠
B. 继续妊娠

24. If there is no prenatal genetic test available or the prenatal genetic test cannot offer a definite result, and you are told that there is 50 per cent risk rate that the foetus is affected by a certain disease, what would you do?
A. Terminate the pregnancy?
B. Continue the pregnancy?

25. 如果没有产前检测或者检测结果不能确定胎儿有没有遗传疾病，胎儿有 25% 的概率患有某种遗传疾病，您会选择：
A. 终止妊娠

B. 继续妊娠

25. If there is no prenatal genetic test available or the prenatal genetic test cannot offer
a definite result, and you are told that there is 50 per cent risk rate that the foetus is
affected by a certain disease, what would you do?
A. Terminate the pregnancy?
B. Continue the pregnancy?

26. 您觉得自己终止妊娠一个有患病概率的胎儿(即便概率很低)最大的原因是:
A. 担心巨额的医疗费用
B. 孩子的生活质量和以后的前途
C. 自己的巨大付出(时间 金钱 精力 心理压力)
D. 社会的压力 可能或必然要面对的歧视
E. 其他

26. What would be the main reason that you might abort a foetus which has a certain
probability/risk of being affected (even if the probability is low)?
A. Worry about high fee for the medical treatment for an affected child.
B. Worry about the life quality and future of the child.
C. Possibly the huge cost in time, money and energy, and the psychological pressure.
D. Possibly the social pressure and potential discrimination in the future.
E. Others, such as ________________________________.

27. 如果孩子的医疗费用和生活有良好的社会保障(比如，政府福利、社会医疗
保障体系)，是否会影响您针对上述问题的回答：_______
A. 会
27. If there is good social security for medical treatment (such as social welfare, healthcare system), would it influence your answer to the above questions? ______
A. Yes.
B. No.
C. Maybe.

28. How would you feel about terminating a foetus that is possibly an affected one?
A. I would certainly feel pain, but feel there is no other way
B. I would feel pain, but it would be more painful to have an affected child and the child would also suffer.
C. It is best to abort an affected foetus. It would benefit the individual, family and the society.
D. Many ‘unexpected’ foetuses have been aborted, not mention affected ones.
E. I would feel painful about aborting the foetus, but perhaps I would feel regret if I kept it.
F. I would feel that people would think I was crazy if I keep it.
29. 如果怀了一个“可能有缺陷”孩子，是否会觉得内心愧疚？

A. 会的，觉得对不起丈夫

B. 觉得自己很不幸很倒霉

C. 不会太愧疚，但估计会考虑终止妊娠

29. Suppose that you are pregnant and possibly the foetus is at risk one of being affected. Would you feel regret or shame?

A. I will feel so sorry for my husband.

B. I would feel so unlucky.

C. I would not feel very sorry, and maybe will decide to terminate the pregnancy.

30. 如果您生育过一个有遗传疾病的孩子，您会决定再生一个孩子吗？

A. 会，我希望至少有一个健康的孩子

B. 不会，我害怕再有一个患病的孩子

C. 不确定

30. If you had an affected child, would you want to have more children?

A. Yes, and I want to have at least one healthy child.

B. No, and I would feel afraid to have another affected one.

C. Not sure.

31. 如果您生育过一个有遗传疾病的孩子，您知道有产前检测或其他技术，可以帮助您（利用产前检测发现并流产可能发病的胎儿），您会决定再生孩子吗？

A. 会

B. 不会

31. If you had affected child, , and you knew that genetic testing could help you (for example, taking a test to find and abort a possibly affected foetus), would you
decide to have more children?
A. I would.
B. I would not.

32. If you had an affected child, and you that technology such as umbilicus blood transplantation and bone marrow transplantation, which could possibly cure the affected one if there is a tissue matching sibling, would you decide to give birth to a child as a ‘saviour sibling’?
A. I would.
B. I would not.

33. Considering question 30, what would be the main reason that you might decide to give birth to a ‘saviour sibling’?
A. To cure the affected child.
B. To have a chance to cure the affected child.
C. In the hope of having a healthy child, even if there is no chance of curing the affected one.
34. 假设家庭里有一个患病的孩子，您会觉得您面临的压力：

A. 经济上费用的支出
B. 精力上时间的投入
C. 精神上缺乏社会关怀和救济
D. 心理上不公平的对待和可能的歧视

34. Suppose that you have an affected child. What would you consider to be the greatest difficulty?
A. The economic burden.
B. The cost in energy and time.
C. The lack of support and help from society.
D. The psychological burden, such as unfair treatment and potential discrimination.

第二部分  DMD (Duchenne muscular dystrophy) 严重性肌营养不良

肌营养不良症 (MD) : 属 X-连锁隐性遗传。Duchenne 型营养不良症 (DMD)，也称严重性假肥大型营养不良症，仅见于男孩，母亲若为基因携带者，50%男性子代发病，常起病于 2-8 岁，初期感走路笨拙，易于跌倒，不能奔跑及登楼，站立时脊髄前凸，腹部挺出，两足撇开，步行缓慢摇摆，呈特殊的“鸭步”步态，当仰卧起立时非常困难，需要先翻身俯卧，再双手攀缘两膝，逐渐向上支撑起立。目前对 MD 没有有效的药物和治疗方法。通常患病者在 20 岁左右死亡。

Duchenne muscular dystrophy (DMD) is an X-linked recessive disease. The female is carrier and transmits the disease genetically to her offspring, but does not develop the disease. DMD is caused by the lack of dystrophin, which is a protein found in the
cell membrane of muscles. Boys are normal at birth and only begin to show signs of the muscle wasting disease when they are 3 or 4 years old. They experience increasing difficulty walking because of progressive weakness, with loss of ambulation, and are often wheelchair bound by 11–12 years of age. The muscle deterioration is continuous and affected children usually die in their late teens, or early twenties, because no effective treatment is yet available.

1. Have you heard of Duchenne muscular dystrophy (DMD)?
   A. I have heard about it.
   B. I have heard about it, but do not know much about it.
   C. I have never heard of it.

2. If you already knew of or have heard of DMD, where did you get the information?
   A. From the local doctors.
   B. Website on Internet, newspaper, TV and other media.
   C. From friends, relatives and patients.
   D. Other.
3. 如果您婚前知道自己是携带者，您觉得是否会告诉你的男朋友（或者未来的男友）

A. 我想会的，我觉得不应该隐瞒。应该在认识初期就告诉
B. 先不告诉，寻找合适机会再说
C. 还是不让他知道的好
D. 不知道该怎么办，不告诉不道德，告诉又顾虑多

3. If you know you were a carrier of DMD, would you let your boyfriend know?
A. Yes, I would tell him at the beginning. I would not ‘cheat’ on him.
B. I would not tell him at the beginning, but maybe later I would choose the right time to tell him.
C. I would not tell him. I would prefer that he did not know.
D. I would not know how to do. It would be wrong not to tell him but I would feel afraid to tell him.

4. 如果你女朋友是 DMD 携带者，你觉得她是否应该告诉你？

A. 是的，这么重要的事情她应该告诉我
B. 我宁愿不知道

4. If your gir friend knows she is a carrier of DMD, do you think she should tell you?
A. Yes. It is so important and she should let me know.
B. I would prefer not to know.

5. 如果你知道你的女朋友是 DMD 的携带者，你会有怎样的反映

A. 继续恋爱
B. 终止恋爱关系
C. 真要慎重考虑
D. 和父母商议，参考父母的意见
E. 和父母商议，听从父母的意见
F. 暂时不告诉父母

5. Suppose that your girlfriend told you she is a carrier of DMD. What would be your response?
A. I would continue the relationship with her.
B. I would end the relationship with her.
C. I would think about it very seriously. It is so difficult to say what I would do.
D. I think that I should and would consult with my parents.
E. It would be better not to tell my parent because I think they would want me to end the relationship with her.

6. 如何看待医生（咨询师）善意的想病人丈夫”隐瞒” 携带者的病史__________
A. 医生应该告诉病人自己，让别人选择是否和怎样告诉自己的亲人
B. 有时善意的隐瞒比真实的告知更好，可以避免对女性最直接的伤害
C. 医生应该告诉病人丈夫
D. 您相信医生知道告知与否以及怎样告知是最好的

6. What do you think about ‘truth telling’ in the counselling clinic?
A. The doctor should tell the client/family the truth about the situation.
B. The doctor should tell wife the truth.
C. The doctor can choose what is best and how to tell it in certain situations.
D. Sometimes, ‘well-intentioned’ truth-covering is better than truth-telling.

7. 假如你是丈夫，你生育了一个 DMD 的孩子。后来你知道你的妻子是 DMD
A. 的携带者，并且你知道是因为妻子的基因问题。您会觉得：__________

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A. I would feel angry and so unlucky.
B. I would blame my wife and think it was her fault.
C. I would blame my wife in mind but not say so.
D. I would accept the fact, and think it was bad fate that could not be helped.
E. Considering the health of offspring, perhaps I would decide to divorce.
F. Perhaps I would decide to divorce.
G. I would face the fact with my wife and console her.
H. I would not want to divorce now, but would not know what to do, or what will happen tomorrow.

8. Suppose you were the father of a child affected by DMD, and you knew your wife to be a DMD carrier, and you knew that your child’s illness was due to your wife’s genes. What would you feel and do?
A. I would feel angry and so unlucky.
B. I would blame my wife and think it was her fault.
C. I would blame my wife in mind but not say so.
D. I would accept the fact, and think it was bad fate that could not be helped.
E. Considering the health of offspring, perhaps I would decide to divorce.
F. Perhaps I would decide to divorce.
G. I would face the fact with my wife and console her.
H. I would not want to divorce now, but would not know what to do, or what will happen tomorrow.
C. 没有办法，自己也没有办法代替孩子受罪，还是要面对现实

D. 很害怕丈夫对自己的态度改变，害怕丈夫嫌弃自己

E. 不想拖累丈夫，或许会考虑离婚

F. 如果丈夫提出离婚，我也没有办法只有同意

G. 想再生一个健康的孩子

H. 不知道会怎么样，说不清楚未来，但很害怕，觉得无助

I. 不管怎样，要尽力为孩子看病，希望孩子活得快乐些

8. Suppose you were the mother of child with DMD and you knew you are a DMD carrier, and that you knew the disease of your child was due to your genes. What would you feel and do?

A. I would feel so sorry and guilty.

B. I would blame self and feel self-condemned.

C. I would feel so sad and really wish I could suffer instead of the child.

D. I would worry about the attitude of my husband, worry that he would dislike me

E. I would consider the feelings of husband and the family and maybe decide to divorce.

F. If my husband wants to divorce, I think I have no way but agree.

G. I would hope to bear another healthy child for my husband.

H. I would not know what to do and would feel frightened and helpless.

I. Despite the situation, I would love the child and try my best to treat and care for the child and hope it would be happy.