Perception of predictive testing for Huntington’s disease by young women: preferring uncertainty to certainty?

Marleen Decruyenaere, Gerry Evers-Kieboom, Herman Van den Berghe

Abstract
Opinions on the implications of predictive testing for Huntington’s disease were evaluated in a group of 169 women (aged 21–35 years) with interest in psychosocial issues, but with no special pre-existing knowledge or training in genetics.

Predictive testing for Huntington’s disease (HD) is considered to be a test case for predictive testing for other late onset diseases, monogenic as well as multifactorial disorders. In the hypothetical situation of having a 50% risk for developing HD, about half of the group expressed interest in a predictive test. As to the question of giving results of predictive tests to third parties, the group would be very reluctant to inform the employer or the insurer, but not their own family. Prenatal testing for late onset diseases was considered acceptable by half of the women; only one quarter of the total group would terminate a pregnancy of a child that might develop a late onset disease.

The assessment of attitudes towards predictive testing was carried out within the context of a global evaluation of perceived advantages and disadvantages of genetic counselling. The attitudes towards predictive testing were systematically associated with perceiving ‘having more certainty about the future’ as an advantage of genetic counselling and with rejecting ‘knowing everything in advance’ as a disadvantage.

Predictive DNA testing has become a reality for several autosomal dominant late onset diseases, such as myotonic dystrophy and Huntington’s disease (HD). As a consequence a unique situation has emerged in that one is able to inform asymptomatic persons that they will get a severe disease, for which there is no cure, with an onset years or even decades after the communication of the test result. It is highly probable that predictive tests for other late onset diseases will be available in the near future (for example, for various forms of cancer, cardiovascular diseases, and psychiatric disorders). It is the main aim of the present paper to assess the attitudes and opinions of young women about predictive testing for Huntington’s disease.

This evaluation was carried out within the context of the women’s perception of the advantages and disadvantages of genetic counselling. According to professionals, genetic tests give people more autonomy and control over their future, especially over procreation. It gives them the opportunity of preventing suffering in children and parents. Predictive testing for late onset diseases often resolves the stressful uncertainty about becoming ill or not, and may help in planning the future more easily. Genetic tests have, however, also entailed negative feelings, such as anxiety and depression, guilt and grief. Positive results of carrier testing may lead to stigmatisation or discrimination. Knowing with certainty that one will develop a serious late onset disease may be like living with a ‘time bomb’. Even in the case of a negative result of a predictive test, maladaptive coping has been observed.

Opinions about predictive testing have not previously been studied in the general population; they have only been evaluated in persons with a high risk of developing a specific disease. In the period immediately preceding the actual test programme in Belgium, 66% of a group of persons at risk for HD reported that they would request the predictive test. The proportion of persons actually entering the predictive testing programme in Belgium is smaller, however. This finding is in keeping with the experience in other centres.

The present paper evaluates young women’s intentions with regard to predictive testing as well as their opinions about informing third parties of the results of such tests. The problems of balancing the person’s right to privacy against the rights of third parties, such as relatives at risk, insurance companies, and employers, are well known in clinical genetics. This is one of the most difficult issues in the ethics of genetic counselling. Wertz and Fletcher stated that “geneticists in all nations are vividly aware of the potential damage from third party access to results, especially access by insurance companies”. The same authors also reported a reluctance of geneticists to giving access to results of genetic tests to employers. Moreover, genetic counselling does not only inform people about their own risk, but also (directly or indirectly) about their brothers’ and sisters’ risk. Therefore, we also evaluated the young women’s opinion with regard to informing relatives.

Methods

QUESTIONNAIRE
The description of the questionnaire is limited to the variables that are discussed in the present paper. A detailed description of the entire
questionnaire can be found in Decruyenaere et al. 17

Perceived advantages and disadvantages of genetic counselling
We asked to what extent the subjects agreed or disagreed with possible advantages and disadvantages of genetic counselling on a five point scale (1 = strongly disagree, 2 = mildly disagree, 3 = don't know, 4 = mildly agree, 5 = strongly agree). The formulation of these pros and cons was inspired by a survey of the VSOP, a patient organisation for genetic diseases in the Netherlands (see table 1 for a list of the advantages and disadvantages).

Attitudes towards predictive testing
We presented a descriptive text about HD (appendix 1). The possibility of a predictive test was explained; we simplified the situation and did not mention the limitations of the linkage test. Then we asked about their intentions to request the predictive test in the hypothetical situation of being at 50% risk for HD. We also assessed the acceptability of prenatal testing for late onset diseases (appendix 2) and their attitudes towards pregnancy termination, should they be pregnant with "a child that would get an adult onset disease (at 35 to 50 years), involving physical and mental deterioration" (appendix 3). Finally, we asked about informing third parties about the result of a predictive test and about informing their brothers and sisters about their increased risk (see Results for the answers).

The variables were measured by means of multiple choice questions and the data were analysed by means of SAS (1988). To study the relationship between the attitudes towards predictive testing and the variables, which were measured on an ordinal scale, we used the Kendall rank correlation coefficient (tau).

Results

DESCRIPTION OF THE SAMPLE
The mean age of the women was 30-35 years (table 2). The group covered the whole range of educational levels; however, compared with the general population the higher levels were slightly overrepresented. About half of the group (56%) had a job, 31% were housewives, and 12% were unemployed. Most of them were married (60%) or had a partner (17%). About 71% of the respondents had at least one child; the average number of children was two. Thirty-five percent were still considering a(nother) pregnancy. The majority of the respondents were religious (Roman Catholic), with varying degrees of church attendance.

PROS AND CONS OF GENETIC COUNSELLING
The agreement and disagreement with the so-called advantages and disadvantages of genetic counselling is presented in table 1. The categories 'strongly agree' and 'mildly agree' are grouped into one category and the same was done for 'strongly disagree' and 'mildly disagree'. When comparing advantages and disadvantages, 62% thought that the pros of genetic counselling outweighed the cons.

ATTITUDES TOWARDS PREDICTIVE TESTING
About half of the group (53%) expressed interest in a predictive test in the hypothetical situation of being at 50% risk for developing HD in the future, while 25% answered that they were not interested. A rather large proportion (16%) did not know and 6% gave no answer. Interest in predictive testing for HD was significantly correlated with agreeing with most of the advantages and disagreeing with the disadvantages of genetic counselling (appendix 4). We only discuss the highest correlations. We found a correlation of tau = -0.42 (p = 0.0001) with 'knowing everything
Table 3  Attitudes towards informing third parties about the results of a predictive test (n=169).

<table>
<thead>
<tr>
<th></th>
<th>Yes with permission</th>
<th>Only with permission</th>
<th>No</th>
<th>Missing</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Partner</td>
<td>73 (43.2%)</td>
<td>78 (46.2%)</td>
<td>3 (1.8%)</td>
<td>15 (8.9%)</td>
<td>169 (100%)</td>
</tr>
<tr>
<td>Children</td>
<td>30 (17.6%)</td>
<td>97 (57.4%)</td>
<td>13 (7.7%)</td>
<td>29 (17.2%)</td>
<td>169 (100%)</td>
</tr>
<tr>
<td>GP</td>
<td>80 (47.3%)</td>
<td>64 (37.9%)</td>
<td>4 (2.4%)</td>
<td>21 (12.4%)</td>
<td>169 (100%)</td>
</tr>
<tr>
<td>Employer</td>
<td>1 (0.6%)</td>
<td>13 (7.7%)</td>
<td>106 (63.9%)</td>
<td>47 (27.8%)</td>
<td>169 (100%)</td>
</tr>
<tr>
<td>Insurer</td>
<td>2 (1.2%)</td>
<td>18 (10.7%)</td>
<td>104 (61.5%)</td>
<td>45 (26.6%)</td>
<td>169 (100%)</td>
</tr>
</tbody>
</table>

in advance': people expressing interest in predictive testing for HD were less likely to think that knowing everything in advance is a possible disadvantage of genetic counselling. Agreeing that 'having more certainty about the future' and 'being better able to plan for the future' were advantages was also associated with a positive attitude towards predictive testing for HD (tau = 0.33 and tau = 0.35 respectively; p < 0.0001). We found no association with sociodemographic variables.

Prenatal testing to predict whether a child will have a late onset disease was considered 'acceptable' by 50% of the group and 'not acceptable' by 32%. Ten percent did not know and 7% did not answer. Here again, we found significant correlations between agreeing with the advantages and disagreeing with the disadvantages (appendix 4). Accepting prenatal testing was associated with agreeing that 'having more certainty about the future' is an advantage (tau = 0.36, p < 0.0001) and with rejecting 'knowing everything in advance' as a disadvantage of genetic counselling (tau = −0.35, p < 0.0001). We found no association with sociodemographic variables.

Twenty-eight percent of the total group (44% of those who considered prenatal predictive testing acceptable) would terminate the pregnancy should they be pregnant with 'a child with a serious late onset disease', 67% would not terminate, and 5% were undecided. The respondents who more frequently practised religion were more likely to reject abortion in this situation (tau = −0.16, p < 0.05). The intention to terminate the pregnancy was most strongly correlated with considering 'prevention of suffering of the parents' as an advantage (tau = 0.32, p < 0.0001) (appendix 4).

We asked the respondents who they thought should be allowed to receive information about the results of a predictive test (table 3). It is striking that a very low proportion of the women would allow the employer and the insurer to be informed. The more frequently practising religious respondents were more likely to permit (unconditionally) their partner and their children to be informed (tau = 0.18, p < 0.05 and tau = 0.15, p < 0.05 respectively). The higher the level of education, the more often they would inform the children (tau = 0.16, p < 0.05). We further confronted them with the hypothetical situation of being informed during genetic counselling about the increased risk to their brothers and sisters and asked their opinion about informing them. Slightly more than half of the respondents were in favour of referring them to a genetic centre to seek the information they need (table 4).
group of respondents would terminate a pregnancy should they be pregnant with 'a child who would get a late onset disease (at 35 to 50 years), involving physical and mental deterioration.' Other studies have reported that a proportion of those who would want prenatal testing (for HD) would not choose to terminate a pregnancy with an affected fetus. “As a result, it is likely that many children will grow up knowing, or learn, that they will eventually develop HD. The psychological impact of this information on the child is unknown”.

The majority of the group seemed to perceive genetic counselling as an opportunity rather than as a threat. Counselling was by about two-thirds of the group perceived as a means to gain more control and certainty about their future, especially about their offspring. Having more control over the health of their offspring was perceived as the most important advantage of genetic counselling by almost three-quarters of the subjects. Preventing suffering of the parents as well as of the handicapped child was also considered as an advantage of genetic counselling. As Van den Berghè stated: “Man in this society of today can no longer find any sense in suffering, especially when it hits him, on a genetic basis, in his existencial prolongation which is the child.” The danger of stigmatisation was perceived as a potential disadvantage by nearly two-thirds of the respondents. Getting information one can not change, anxiety, and the possibility of misuse of scientific genetic research were also considered as potential disadvantages by more than half of the group. In general, the advantages of genetic counselling were considered to outweigh the disadvantages.

The attitudes towards predictive testing were systematically associated with perceiving 'having more certainty about the future' as an advantage of genetic counselling and with rejecting 'knowing everything in advance' as a disadvantage. The possibility of having (some) certainty about the future and the need to know things in advance seem to play an important stimulating role in the attitudes towards predictive tests. It would be interesting to explore the personality profile corresponding with a person's need for certainty and with the need to know things in advance. This exploration may also be relevant to future predictive testing for other types of diseases, in particular when therapeutic interventions are available after a test result or when post-test changes in behaviour or life style can decrease the risk of developing symptoms. Nevertheless, we should keep in mind that knowing the consequences of specific behavioural patterns does not guarantee that the necessary behavioural changes will be made.

The Centre for Family Science in Brussels is acknowledged for its participation in this study. We are very grateful to all students as well as to the teachers and the director, Mrs G Jennes. This project was supported by the Inter-University Network for Fundamental Research sponsored by the Belgian Government (1991–1995).

16 Werts DC, Fletcher JC. An international survey of the attitudes of medical geneticists towards mass screening and access to results. Pahl Health Rep 1989;104:335–44.

Appendix 1 Informative text on HD. Huntington’s disease is a genetic disease, which progressively affects parts of the brain. The principal features are physical and mental deterioration. The first symptoms usually appear between 35 and 50 years. The majority of the patients die 10 to 20 years later.

Huntington’s disease is an autosomal dominant disease. That means that all children of an affected parent, boys as well as girls, have a 50% risk (thus 1 chance in 2) of developing the disease. The uncertainty about their health and the children’s future is very burdensome.

For some years, a predictive test has been available. Long before the manifestation of the first symptoms, it is possible to find out whether or not one has inherited the anomalous gene by means of a blood sample. If one has not inherited the anomalous gene, then one knows with certainty that one will not get the disease. If one has inherited the anomalous gene, then one knows with certainty that one will get the disease and that one can pass the disease on to the children.
Appendix 2 Prenatal testing for late onset diseases.
What is your opinion about prenatal testing to predict the serious diseases that the unborn child will get when grown up?
(1) This is completely unacceptable.
(2) This is fairly unacceptable.
(3) I don’t know.
(4) This is fairly acceptable.
(5) This is completely acceptable.

Appendix 3 Pregnancy termination.
In which of the following situations would you terminate the pregnancy?
One of the situations was: When pregnant with a child that would get an adult onset disease (at 35 to 50 years), involving physical and mental deterioration.

Appendix 4 Intercorrelations (Kendall-tau) of attitudes towards (A) predictive testing for HD, (B) prenatal testing for adult onset diseases, and (C) pregnancy termination should they be pregnant with a child who would get an adult onset disease, involving physical and mental deterioration, with agreeing with the so called advantages and disagreeing with the so called disadvantages of genetic counselling (n = 151 to 159).

<table>
<thead>
<tr>
<th></th>
<th>(A) Predictive test for HD</th>
<th>(B) Prenatal test</th>
<th>(C) Pregnancy termination</th>
</tr>
</thead>
<tbody>
<tr>
<td>So called advantages</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Having more certainty about the future</td>
<td>0.33***</td>
<td>0.36***</td>
<td>0.17*</td>
</tr>
<tr>
<td>Being able to plan better for the future</td>
<td>0.35***</td>
<td>0.27***</td>
<td>0.15*</td>
</tr>
<tr>
<td>Preventing suffering of the parents</td>
<td>0.24***</td>
<td>0.25***</td>
<td>0.32***</td>
</tr>
<tr>
<td>Preventing suffering of the handicapped child</td>
<td>0.17*</td>
<td>0.18**</td>
<td>0.17*</td>
</tr>
<tr>
<td>More financial resources for other aspects</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Control over the health of the offspring</td>
<td>0.05</td>
<td>0.15*</td>
<td>0.08</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>So called disadvantages</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Anxiety about undergoing a genetic check up</td>
<td>-0.17*</td>
<td>-0.11</td>
<td>-0.02</td>
</tr>
<tr>
<td>The danger of stigmatisation</td>
<td>-0.23**</td>
<td>-0.23***</td>
<td>-0.04</td>
</tr>
<tr>
<td>Making people unhappy</td>
<td>-0.22**</td>
<td>-0.15*</td>
<td>-0.07</td>
</tr>
<tr>
<td>Getting information one can not change</td>
<td>-0.21**</td>
<td>-0.16*</td>
<td>-0.07</td>
</tr>
<tr>
<td>Knowing everything in advance</td>
<td>-0.42***</td>
<td>-0.35***</td>
<td>-0.15*</td>
</tr>
<tr>
<td>The danger of misuse of genetic research</td>
<td>-0.10</td>
<td>-0.27***</td>
<td>-0.16*</td>
</tr>
</tbody>
</table>

Answer categories:
(A) I would not use it (= 1), I don’t know (= 2), I would use it (= 3).
(B) See appendix 2.
(C) Not terminate (= 1), terminate (= 2).

Level of significance of the correlation:
* significant at level 0.05.
** significant at level 0.01.
***significant at level 0.001.