Huntington's Disease Society of America

Psychological Issues of Testing Positive for HD

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Presenter Disclosures

Kimberly A. Quaid, Ph.D.

The following personal financial relationships with commercial interests relevant to this presentation existed during the past 12 months:

No relationships to disclose
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Predictive Testing for Huntington Disease

- Linked markers to HD gene found in 1983
- Group of health care professionals, patients, and HD advocate organizations met to devise testing protocols and guidelines
- Testing guidelines published in leading journals and by advocate organizations
- Protocols instituted in centers offering testing both nationally and internationally

Genetic Testing Protocols

- Neurological examination
- Pretest counseling
- Results given in person
- Follow-up available
  - Clinic visits
  - Referrals for supportive therapy

Estimates of Uptake

- In Europe, less than 20% of the at-risk population takes the test
- In Australia, estimates are 13-15%
- In the United States, about 10-12% of at risk population seeks testing
- In contrast to:
  - Breast cancer – 50%
  - Familial adenomatous polyposis – 80%

Who Decides to Test?

- More likely to have higher educational levels than general population
- More likely to be woman

Short Term Psychological Impact

- Wiggins et al. (1992) The psychological consequences of predictive testing for Huntington disease. New England Journal of Medicine, 327:1401-1405.
- Tibben et al. (1997) Three year follow up after presymptomatic testing for Huntington disease in tested individuals and partners. Health Psychology 16:20-35.
- Codori et al. (1997) Predictors of psychological adjustment to genetic testing for Huntington disease . Health Psychology 16:36-50.
- Quaid and Wesson (1995) Exploration of the effects of predictive testing for Huntington disease on intimate relationships. American Journal of Medical Genetics. 57:46-51.

Short Term Psychological Impact

- Decruyenaere et al. (1996) Prediction of psychological functioning one year after the predictive test for Huntington disease and impact of the test result on reproductive decision making. Journal of Medical Genetics 33:737-743.
- Dudok de Wit (1997) To know or not know: the psychological implications of presymptomatic testing for autosomal dominant inheritable late-onset disorder. (PhD thesis) Erasmus University Rotterdam, The Netherlands.

Short Term Psychological Impact

- Gene positive and gene negative individuals differed on all psychological outcome measures at 7-10 days post-test but not at 6 months and 12 months of follow-up
- Study based on largest sample size to date measured hopelessness and found differences between gene positive and gene negative individuals may persist long term
- This finding raises some concern as hopelessness has been found to be a predictor of suicide
- Analysis of scores before and after receiving test results indicates that the psychological adjustment of gene negative individuals tend to be unaltered or improved after the receipt of test results

Short Term Psychological Impact

- Baseline levels of depression or hopelessness (rather than test result itself) were found to be the best predictors ort levels of hopelessness and intrusive thoughts after disclosure
- A small proportion of gene negative individuals had serious difficulties coping with their new genetic status
- Contributing factors to this outcome was having made irreversible decisions based on the belief that they would develop HD or overly optimistic expectations of the positive effects of a decreased risk

Short Term Psychological Effects- Partners

- Partners of gene positive individuals tend to have the same course of distress as their spouses
- Compared to the spouses of gene negative individuals, partners of gene positive individuals had significantly higher levels of psychological distress, 1 week, 6 months and 3 years after disclosure.
- Having children was an additional psychological risk factor for spouses

Long Term Effects of Genetic Testing

- Timmen et al. (2004) Adverse effects of predictive testing for Huntington disease underestimated: Long term effects 7-10 years after the test. Health Psychology 23:189-197.
- Gargiulo et al (2009) Long term outcome of presymptomatic testing in Huntington disease. European Journal of Human Genetics 17:165-171.

Dropouts – Timman et al

- Thirty-nine of the 181 individuals who had originally come for testing and for whom pretest measures were available did not return for follow-up or additional counseling
- Individuals who turned out to be carriers and who did not return for follow-up scored at pretest significantly higher on hopelessness, intrusion, avoidance and lower on general well-being than did individuals who turned out to be carriers and did return for follow-up
- No significant difference for non-carriers

Results – Timman et al

- Carriers and their spouses were more distressed immediately after the test results when compared to non-carriers
- Their outlook improved somewhat in 2-3 year post-test period
- However, they became more pessimistic and hopeless thereafter when approaching the age of onset (7-10 years after testing)

Results – Gargiulo et al

- Carriers not more anxious than non-carriers after testing
- However, current depression was significantly more frequent in carriers
- The same percentage of carriers and non-carriers had experienced depression prior to testing
- After testing, the percentage of carriers experiencing depression increased from 42% to 49% whereas the percentage of non-carriers experiencing depression decreased from 45% to 31%

Suicide Attempts – Gargiulo et al

- After testing, there was one suicide attempt and one psychiatric hospitalization for major depression in the carrier group
- After testing, three non-carriers attempted suicide, one was hospitalized for depression and one for a psychotic episode
- Only 31% of carriers and 15% of non-carriers were under psychiatric care ($p<.05$)
- Only 36% of carriers and 15% of non-carriers were under treatment with antidepressants or anxiolytic drugs ($p<.05$)

Factors Related to Depression After Testing

- Having a positive test result
- Having as test motivation “other than for offspring”
- Having a history of depression before the test

Qualitative Research on Testing Outcomes

- Three major positive experiences from testing
  - Relief from uncertainty
  - More appreciation of life
  - Brought the family closer together
- Four major negative experiences from testing
  - Regrets about receiving test results
  - Difficulties in adapting to new genetic status
  - Difficulties with Informing children
  - Difficulties in planning for the future

Hagberg et al. (2011) More appreciation of life or regretting the test? Experiences of living as a mutation carrier of Huntington's disease. Journal of Genetic Counseling 20:70-79

Qualitative Research on Disclosure While Dating

- Whether to tell
- Why tell?
- Why not tell?
- What to tell?
- When to tell?
- Reported reactions to telling
- Alternatives to telling

Whether to Tell

- Privacy vs. obligation to tell
- Most felt clear obligation once one had definitively decided to get married
- But, individuals reported weighing competing pros and cons – balancing ethical obligation to tell with fear of rejection

Why Tell?

- Tell because of ethical obligation
- Tell because of disease in family that could not be hidden
- Tell because children may be affected
- Tell because it is hard to hide symptoms

Why Not Tell?

- Fear of rejection

What to Tell?

- How much detail to provide : “I have this disease in my family” vs. “I am gene positive and will develop this disease in my family.”
- Disclosing symptoms but not diagnosis “My mom is aging a little, forgetful” rather then “”My mom has Huntington disease.”
- Disclose implications for reproduction

How to Tell?

- The content of the information: Framing in as positive a manner as possible
- The form of communication: In person or not (email)

When to Tell?

- Telling when close enough
- Telling only if asked
- Telling only after breaking up

Reported Reaction to Telling

- Rejection vs. acceptance
- Factors affecting reactions
  - Symptoms
  - Treatments

Alternatives to Telling

- No dating as a result
- Third party disclosures

Impact of Testing on Family

- Changes in family membership
- Changes in communication
- Changes in roles surrounding caregiving

Changes in Membership

- Between spouses and partners
  - Some marry some divorce
- Between siblings
  - Survivor guilt
  - Designated patient
- Between parents and offspring
  - Death/desertion
- Between nuclear and extended family
  - Feeling not part of the family

Communication

- Individuals pursuing testing may wish to communicate about HD in ways that other family members are not comfortable with
- Secrets vs open communication
- Sharing test results vs not sharing

Caregiving Issues

- Current role changes with regard to caregiving when symptoms appear
- Anticipated role changes that may increase stress on current marriage and/or family relationships
- Reactivation of dormant conflicts among family members or intensification of existing conflicts

Summary

- Papers published looking at short term outcomes of testing found relatively few negative effects but neglected to examine dropouts.
- At least one study examined dropouts and found that carriers who were lost to follow-up after disclosure of test results reported more pretest distress than did carriers who returned for follow-up
- This suggests that earlier studies may have underestimated negative impact of testing

Summary

- Carriers appear more distressed than non-carriers immediately after receiving test results
- Carriers’ outlook tends to improve 2-3 years after testing but becomes more pessimistic as the age of onset approaches.
- Both carriers and non-carriers exhibit significant levels of depression suggesting that the at risk population is vulnerable and that psychological support should be available regardless of test results.

Summary

- Despite high levels of depression, as a group both carriers and non-carriers alike appear to be undertreated in terms of psychiatric care and/or medication
- Individuals who are single/divorced/widowed have difficulties in figuring out whether and how to disclose information about both the disease in the family and that they are gene positive
- Families are all different and genetic testing can have a severe impact on the family system in a variety of ways

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