Psychological consequences of predictive genetic testing: a systematic review

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Authors' objectives
To describe the psychological consequences of predictive genetic testing.

Searching
EMBASE (from 1990), MEDLINE (from 1990), PsycInfo (from 1984) and the Social Sciences Citation Index (from 1990) were searched for articles, excluding dissertations and book chapters; the search terms were provided in the paper. In addition, the Science Citation Index was searched for citations of authors from two research groups (Tibben et al., The Netherlands and Wiggins et al., Canada) that had produced relevant papers earlier. Experts in the field were also contacted and references were sought from cited papers.

Study selection
Study designs of evaluations included in the review
All studies employed prospective designs with a pre-disclosure baseline assessment and between one and four assessments post-disclosure. Studies were included if they were of standardised outcome measures, but were excluded if they were qualitative (i.e. case studies, no standardised outcome measures and no statistical comparison of groups).

Specific interventions included in the review
The included studies were of predictive genetic testing for Huntington's disease, hereditary breast and ovarian cancer, familial adenomatous polyposis and spinocerebellar ataxia.

Reference standard test against which the new test was compared
The review did not include any diagnostic accuracy studies that compared the performance of the index test with a reference standard of diagnosis.

Participants included in the review
There were no pre-specified inclusion or exclusion criteria relating to the participants. The participants in the included studies were adults (over 18 years) and children aged 6 to 16 years. The proportion of women in the included studies ranged from 44 to 100%. The participants in the included studies were white and Japanese, among other ethnicities (not reported).

Outcomes assessed in the review
All consequences assessed in the review studies were of, or related to, emotion. The emotional outcomes were:

- depression, as measured by the Beck Depression Inventory, Beck Hopelessness Scale, the Centre for Epidemiological studies Depression scale, the social desirability scale, subscale of the Minnesota Multiphasic Personality Inventory for children, the Children's Depression Inventory, and for adolescents the Reynold's Adolescent Depression Scale;

- state anxiety, as measured by the state subscale of the State Trait Anxiety Inventory, and for children the Revised Children's Manifesto Anxiety Scale;

- general distress, as measured by subscales of the revised Symptom Check List 90-R including the General Severity Index,’ the General Health Questionnaire, the Medical Outcomes Study subscales of role functioning and sexual functioning, the General Wellbeing Scale, and the sexual satisfaction scale of the Marital Satisfaction Inventory; and

- situational distress exhibited by intrusive thoughts and attempts to avoid these thoughts, as measured by the Impact of Events subscale: Intrusion and Avoidance.

Studies of mediators of the outcomes of interest, such as information recall, knowledge or attitudes, were excluded.
How were decisions on the relevance of primary studies made?
Two persons independently coded the papers for eligibility using the specified criteria. Any discrepancies were clarified through discussion.

Assessment of study quality
The authors do not report the method used to assess quality, or how the quality assessment was performed.

Data extraction
Two persons extracted the data and resolved any discrepancies by discussion. The categories of data extracted included: authors, condition, country of study, assessment time-points, emotional outcomes, number approached, baseline response, retention at follow-up, retention overall, carriers, non-carriers, decliners, no clear test result, test result not reported, and participant age and gender.

Methods of synthesis
How were the studies combined?
A narrative synthesis of all the studies was undertaken.

How were differences between studies investigated?
The numbers of study analyses reporting a change, decrease or increase in distress amongst either carriers or non-carriers at baseline and at follow-up, were assessed.

Results of the review
Fifteen studies with at least 1,009 participants (not all studies recorded the numbers involved) were included in the review.

None of the 15 studies reported increased distress (general and situational distress, anxiety and depression) in carriers or non-carriers at any point during the 12 months after testing. Both carriers and non-carriers showed decreased distress after testing; this was greater and more rapid amongst non-carriers.

The test result (i.e. being a carrier or non-carrier) was rarely predictive of distress more than one month after testing; it was predictive in only 2 of the 14 analyses. Pre-test emotional state was predictive of subsequent distress in 14 of the 27 analyses.

Authors’ conclusions
There was a lack of informative studies in this field. The studies reviewed suggested that those patients undergoing predictive genetic testing do not experience adverse psychological consequences. However, the studies were of self-selected populations who agreed to participate in psychological studies and were followed up for no more than three years. Most research was on Huntington’s disease and included a follow-up of no more than one year. The results suggested that testing protocols should include a pre-test assessment of emotional state, so that post-test counselling can be targeted at those more distressed before testing. None of the studies experimentally manipulated the amount or type of counselling provided. The relationship between counselling and emotional outcome was, therefore, unclear and awaits empirical study.

CRD commentary
The review commenced with a clear question. However, the exclusion and inclusion criteria were not stated clearly. The literature search was relatively comprehensive, and no language restrictions were noted. There were limited attempts to locate unpublished studies, apart from contact with experts. Therefore, some studies may have been missed. Details of some aspects of the review methodology were given, but there were no details of the quality assessment. A narrative synthesis was appropriate given the nature of the data.
The authors' conclusions appear to follow the results presented in the paper, but should be interpreted with caution given the limitations mentioned.

Implications of the review for practice and research
Practice: The authors did not state any implications for practice.

Research: The authors state that empirical research of the relationship between counselling and emotional outcome is required.

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This is a critical abstract of a systematic review that meets the criteria for inclusion on DARE. Each critical abstract contains a brief summary of the review methods, results and conclusions followed by a detailed critical assessment on the reliability of the review and the conclusions drawn.