



CLINICAL GENETICS SOCIETY
Clinical Governance Subcommittee

**Paper 4 (Version A, 5/7/2001) Protocols For Pre-Symptomatic
(Predictive) Testing For Late Onset Genetic Disorders.**

Replies to: Dr Helen Hughes, Institute of Medical Genetics, Cardiff CF14 4XW

Tel: 01745 534447 Fax: 01745 534431 e-mail: Helen.Hughes@cd-tr.wales.nhs.uk

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2 **Introduction**

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4 The diseases included in this category are single gene disorders such as Huntington disease,
5 or a Mendelian sub-set of common cancer such as familial polyposis coli (FAP), hereditary
6 non-polyposis colorectal cancer (HNPCC) and breast cancer (BRCA 1 and 2).

7

8 Guidelines for the molecular genetics predictive test have been developed for Huntington
9 disease¹. Similarly, guidelines have also been developed for familial breast cancer, BRCA1
10 and BRCA2 gene testing. In the case of bowel cancer, two types of protocols currently exist,
11 an extended one, based on Huntington disease protocol, and a shortened one currently being
12 piloted⁴.

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14 **The rational for such protocols**

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- 16 1. The aim and main purpose is the client-led exploration of the decision concerning testing.
- 17 2. Distinct from diagnostic testing, pre-symptomatic testing for late onset disorders achieves
18 a change in knowledge about self, sometimes in the absence of effective treatments.
19 Therefore, psychological components of the decision, such as reactions and adaptations,
20 are arguably the predominant elements of pre-symptomatic testing¹.
- 21 3. Models were developed to provide an outline structure to assist clinicians in facilitating
22 the decision-making process and preparing candidates for a genetic pre-symptomatic test
23 result. Clinicians need to be clear about their roles and aims while conducting these
24 interviews. Solden et al suggest that this model worked well for Huntington disease, but
25 for other "common" disorders, this model can be used for guidance, but for each disease,
26 the different aspects will need re-prioritising, and new considerations may need to be
27 incorporated. In breast cancer, for example, the different public perceptions of the
28 condition and how individual deals with uncertainty may be an important additional
29 variable to explore with those requesting breast cancer susceptibility testing.

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31 **Aims of pre-symptomatic testing**

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33 Evaluation of the current and developing models is needed and is still mostly awaited, but
34 authors have recognised the need to promote:

- 35 1. Adaptation to a result
- 36 2. Informed decision making
- 37 3. Client-centred focus
- 38 4. The uniqueness of the individuals experience.
- 39 5. The working through of experiences at the candidates' own pace, and in their own
40 way

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42 **The process of pre-symptomatic testing interview has been described as²:**

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44 Clarification → consideration → education → reflection → decision making → drop
45 out/result

46

- 47 1. Clarification of candidate's understanding of the disease and testing
- 48 2. Consideration of the potential impact and available coping strategies
- 49 3. Education using factual information about the disease and testing, impact of the result,
50 stress and coping, coping resources and strategies
- 51 4. Reflection to help individual to assess whether they have the resources to cope with
52 their potential psychological reaction.
- 53 5. Decision-making is a process, typically occurring outside the counselling context.

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55 **Essential elements of a pre-symptomatic testing protocol**

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57 There is a significant deficiency in evidence upon which to base guidelines. Research
58 projects must address this deficit. Our *suggestions* for best clinical practice, to achieve the
59 aims outlined above include:

- 60 a. Pre-test information - either verbal or supplemented by written information. The latter
61 may cut sessions down from 2 to 1 in some disorders.
- 62 b. At least one face to face pre-test counselling session to facilitate clarification,
63 consideration and reflection.
- 64 c. A consideration of time for client reflection in any protocol.
- 65 d. Face to face meeting for result giving, to allow response to questions and support for
66 emotional distress.
- 67 e. Follow-up protocols in place to give support and identify any adverse effects.
- 68 f. Clinician review of each test result.

69

70 **Conclusions**

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- 72 1. Guidelines for Huntington disease have served the patients, their families and the
73 professionals involved in the delivery of the service, very well over several years. They
74 remain very useful guidelines.
- 75 2. Guidelines for breast, ovarian and bowel cancer.
 - 76 2.1. Full adoption of Huntington's guidelines may not be appropriate. Differences exist,
77 as described below.
 - 78 2.2. Unlike Huntington disease, there are preventive surgical &/prophylactic treatment
79 available that may reduce the risk of breast, ovarian and bowel cancer.

- 80 2.3. A period of one month contemplation prior to testing may not be necessary; a shorter
81 period may felt to be sufficient by some workers in some circumstances.
- 82 2.4. While geneticists in some centres give all results, it may not always be practical or
83 feasible in a cancer situation.
- 84 2.5. The requirement of a formal psychological appraisal prior to testing is uncertain.
85 Further research is needed.
- 86 3. Most of the steps required for Huntington disease PST can be viewed as best practice
87 guidelines for all genetic disorders, if adequate resources are available. While curtailing
88 them for Huntington disease may be detrimental, using them in the cancer situation may
89 not be cost-effective. Further study is needed to establish the evidence.
- 90 4. We cannot advise formally on any change of practice, until we have such evidence.
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92 **References**

- 93
- 94 1 International Huntington Association, World Federation of Neurology Research Group on
95 Huntington's Chorea. Guidelines for the molecular genetics predictive test in Huntington's
96 disease. *Neurol* 1994;44:1533-1536.
- 97 2 Psychological model for presymptomatic test interviews: lessons learned from Huntington
98 disease. Soldan J, Street E, Gray J, Binedell J, Harper PS. *J Genet Counseling* 2000;9(1):15-
99 31.
- 100 3 Marteau TM, Croyle RT. Psychological responses to genetic testing. *Br Med J*
101 1998;316:693-696.
- 102 4 Brain K, Soldan J, Gray J, Sampson J. Genetic counselling protocols for hereditary
103 nonpolyposis colorectal cancer: a survey of UK centres. Submitted for publication.

Comparison of predictive testing carried out for 3 main groups of disorders ¹⁻⁴

The disease	Presentation of pre-test information (clarification consideration, reflection & education)	Minimum interval before informed decision making	Proband psychological appraisal e.g. Hospital Auxiliary Depression Scale (HAD)	Result	Post test counselling	Who carries out counselling & where	Post test support	Number of the total counselling sessions	Evidence of effectiveness (cost effective procedure)
Huntington	✓	Usually 1 month, although There is some variation to this principle.	✓	Face-to-face in genetics centres	✓	Geneticist as part of a team including, counsellors, nurses, psychologist, neurologists and social worker. Some centres do not include the psychology component.	✓ counsellor contact within 1 week	Usually 3	✓ Good psychological outcomes may be attributed to the multi-stage model of genetic counselling, but not clear which element ¹ .
Breast cancer	✓	questioned	questioned	There is some debate over other health professionals' involvement	✓	Geneticist/counsellor as part of a team	-Telephone contact within 1 week -Follow up appointment 1 month post result clinic	2-3	?
Bowel cancer	✓	questioned	questioned	There is some debate over other health professionals' involvement	✓	Geneticist/counsellor as part of a team	-Telephone contact within 1 week -Follow up appointment 1 month post result clinic	Median number was 2 (range 1-3) of 16 genetic centres surveyed ¹	?