Attitudes of general practitioners to presymptomatic testing for Huntington's disease

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Abstract

A postal questionnaire was sent to all 797 general practitioners (GPs) in the Lothians, Borders, and Fife (Scotland), enquiring about attitudes to presymptomatic testing for Huntington's disease. The response rate was 74%. Eighty-two percent were in favour of the principle of predictive testing for Huntington's disease. A majority of those not in favour were prepared to refer their patients for testing. However, three-quarters of GPs were unfamiliar with the details of DNA based linkage analysis. Half of the respondents felt that disclosure of the test result and subsequent counselling and support were the responsibility of the genetic clinic. A third of respondents considered that the genetic clinic should disclose the test result while the GP should give post-test counselling and support. These findings suggest that delivering presymptomatic testing to persons at risk of Huntington's disease would be facilitated by a closer involvement of local GPs.

A presymptomatic test for persons at risk of developing Huntington's disease (HD) is now available using DNA markers closely linked to the HD gene.¹⁻³ Numerous studies have been carried out to test the attitude of Huntington's disease families towards predictive testing.⁴⁻⁸ The attitude of general practitioners (GPs) towards this developing service has not previously been considered.

The Human Genetics Unit in Edinburgh serves the geographical area of the Lothians, Borders, and Fife. This is a region with a population of 1.18 million and a prevalence of Huntington's disease estimated to be 6.5 per 100 000.⁹ The Unit has on record 279 persons at a 50% risk of developing HD. Since we initiated

our presymptomatic test programme in August 1987, 86 (32%) of these subjects have enquired about predictive testing and subsequently 33 entered the programme.

There were 25 new referrals to the Unit for genetic counselling for HD from January 1988 to December 1988. Of these, 60% were referred by their GP. There is a strong likelihood that GPs will initiate counselling for presymptomatic testing for HD. Indeed in Edinburgh self referral is not generally accepted and consultands who enquire about the test are encouraged to consult their GP and to ask to be referred to the Unit. There may also be involvement of the GP in the practical aspects of blood sampling for DNA extraction from key family members. The contribution that GPs can make to genetic services specifically within the developing area of disease detection by gene tracking has been outlined.¹⁰

There is an equally strong likelihood that consultands may require GP support or intervention during the period of major adjustment to their HD status. The principle of designing an optimum approach to disclosure of the test result and working closely with relevant medical personnel was advocated before presymptomatic testing for HD became available.¹¹ The protocol for predictive testing used by the Human Genetics Unit involves contact with the GP at various stages (figure).

The opinion of GPs about developing services within the Unit has been sought in the past with a good response. It therefore seemed a useful adjunct to assessing GP's attitudes to presymptomatic testing to ask their opinion about the optimum method of disclosing the test result and subsequent counselling and follow up of the consultand.

Subjects and methods

At the time of the survey there were 797 GPs practising in the Lothians, Borders, and Fife. Permission to circulate the questionnaire was obtained from the Area Medical Committees of the respective Health Boards. Distribution of the questionnaire was undertaken by the Primary Care Departments. GPs were not asked to identify themselves in their responses, to avoid indirectly ascertaining cases of HD.

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Protocol for predictive testing used by the Human Genetics Unit.

The questionnaire was designed in two sections of essentially closed questions, but with space for comment at the end of each section. Section I (seven questions) sought general attitudes to predictive testing. Section II (one question) was concerned with GPs' views on the delivery of test results and responsibility for the after care of consultands. The detailed questions are listed in Results.

Results

SECTION I

GP's with HD patients and patients at risk of HD

The questions were:

- (1) Do you have on your list any patients with HD?
- (2) Do you have on your list any patients at risk of HD?

There were 594 responses as shown in table 1.

Familiarity with type of analysis

The question was:

- (3) Are you familiar with the type of analysis involved in presymptomatic testing for HD?

Of the 594 responses, 24% were yes and 76% were no.

Attitudes towards presymptomatic testing

The three questions asked were:

- (4) Do you as a general principle approve of pre-symptomatic testing for persons at high risk of developing HD?
- (5) Would you be agreeable in principle to referring one of your patients for presymptomatic testing if they requested it?
- (6) Would you feel that it was your duty to inform one of your patients at high risk of developing HD that a presymptomatic test is available.

Table 1 GP's with HD patient or at risk patient on list. Numbers (and percentages).

GP with HD patient	Yes 112 (19)		No 482 (81)	
GP with at risk patient	Yes 81 (72)	No 8 (7)	Don't know 23 (21)	Yes 83 (17)
			No 181 (38)	Don't know 218 (45)

Table 2 GP's attitudes to presymptomatic testing for HD. Numbers (and percentage of respondents).

Attitude to principle	In favour 488 (82)		Not in favour 12 (2)		Unsure 94 (16)	
Agree to refer patient	Yes 488 (82)	No 0	Yes 9 (2)	No 3 (0.5)	Yes 91 (15)	No 3 (0.5)
Duty to refer patient*	Yes 475 (81)	No 9 (2)	Yes 7 (1)	No 4 (0.5)	Yes 74 (13)	No 18 (3)

*Seven GPs did not respond.

Table 3 Optimum approach to disclosure of test result, post-test counselling, and support.

Options	Response
(a) Genetic clinic gives test result, post-test counselling, and support	324 (54%)
(b) Genetic clinic gives result, GP gives post-test counselling and support	178 (30%)
(c) GP gives result, post-test counselling, and support	63 (11%)
(d) No reply	29 (5%)

The 594 responses to questions 4 and 5 and the 587 responses to question 6 are shown in table 2.

Reasons for negative responses

The question asked was:

(7) If the answer to question 4, 5, or 6 is no, would you indicate your objections as:

- (a) the test is likely to do more harm than good (30 respondents);
- (b) it is not my business to provide such information (1 respondent);
- (c) the test might involve me in more support than I can give (5 respondents).

SECTION II

Optimum method for disclosing test result and giving post-test counselling and support

The question asked was:

"If one of your patients decided to undergo pre-symptomatic testing for HD, what do you consider to be the optimum approach to disclosure of the test result, post-test counselling, and support?"

Three options were listed (table 3). There were 565 respondents.

Discussion

Presymptomatic testing for HD began in Edinburgh in August 1987 within a structured research protocol.³ The programme was notified to the Huntington's Disease Association and to local GPs through a newsletter. Although a large number of persons at risk of HD are already known to the Unit, we expected that others would be referred by their GPs. Whatever the method of referral, we felt that informing a GP that his or her patient had decided to enrol in the presymptomatic testing programme was a desirable (though not obligatory) part of our protocol. What has not been clear until now is whether GPs approved of presymptomatic testing and what views they had on its method of delivery.

The response to the postal questionnaire was high (74%), practically all forms were completed in full, and many GPs commented at length on the questions.

Nearly one-fifth of GPs (table 1) stated that they had an HD patient on their list, a surprisingly high figure when the average list size is 1600. In this group, however, 21% did not know whether they had others on their list at risk of developing HD. This confirms the value of a genetic register system as an independent facility for ascertainment, screening, and counselling of persons at risk of a serious genetic disorder. We were encouraged to find that 17% of GPs who did not have an affected patient on their list were aware, nevertheless, that they had persons on their list at risk of HD.

It is clear from our results that the majority of GPs are unfamiliar with the principles of gene tracking through linked DNA markers. Although this result was not unexpected, it does emphasise the need to educate medical personnel about a form of analysis that is used increasingly in prenatal diagnosis and carrier detection in the more common Mendelian disorders. Requests for information from the respondents to the questionnaire prompted us to draw up an explanatory leaflet outlining the principles and problems of presymptomatic and prenatal exclusion testing for HD.

The majority of GPs confirmed that they are in favour of a presymptomatic DNA test for HD (table 2). There were, however, many comments that indicated two major concerns. One was the possible harmful effects of diagnosing presymptotically a serious late onset neurological disorder with no known prophylaxis or treatment. The other related to our ability to distinguish persons who would not be able to cope with the consequences of a positive test result. All our consultands have their psychological status evaluated by a psychiatrist (figure). However, there appear to be few, if any, valid criteria that allow a judgement about suitability for testing to be made prospectively. As part of our programme we are attempting to identify aspects of personality and support systems which lead to a better or worse adjustment to test results. This can ultimately only be achieved by careful long term follow up to those who elect to undergo presymptomatic testing.

Most GPs who were not in favour of presymptomatic testing or were unsure of their views indicated that they would not allow their attitude to deter a patient from pursuing this option. There was clearly some antipathy to the word 'duty' which GPs understood to mean actively seeking out subjects at risk and informing them of this new diagnostic possibility.

They wished their patients to be fully informed but considered that such information was best incorporated into a routine consultation. Although only 6% of the responding GPs gave a negative response to the question on their 'duty to inform', this result indicates the need for alternative methods (for example, genetic register, Huntington's Disease Association, Combat) of reaching persons at risk.

Many GPs acknowledge that resolving uncertainty will be an inducement for many persons at risk to take the test. They envisage that a positive result is likely to create a number of additional stresses and uncertainties. Nevertheless, only five GPs indicated that they were not in favour of presymptomatic testing because it might involve them in more support than they could give. They also acknowledge that a considerable number of subjects at risk will reject the test on the grounds that a positive test result would be too hard to live with. If there is any danger that presymptomatic testing could gain the type of momentum that would exert undue pressure on persons at risk, then counselling by the family doctor in an atmosphere different from that of the genetic clinic would be a valuable corrective.

It is clear from section II of the questionnaire that not all GPs regard the management of tested patients as the sole responsibility of the genetic services. Thirty percent wished to carry out post-test counselling support with a further 11% also wishing to disclose the test result. From their comments, it is evident that many GPs regard the genetic counsellor as better qualified to carry out in depth counselling. They feel, however, that combined care is essential and see their role as complementary to the genetic clinic.

It is our aim to develop a presymptomatic test procedure that is sufficiently flexible to meet the needs of each consultand while adhering to the discrete stages of our protocol. We see no reason why the concerned GP should not play an integral role in the decision making processes before testing and in the years of support that will inevitably follow. Indeed many consultands enrolling in our programme have nominated their GP as their main source of post-test support.

In the short term it is essential that consultands are followed up. This will involve counselling and careful observation for early recognition of psychological problems, evaluating the overall impact of testing, and exploring the self care practices of those who are adjusting to their test result. In the long term we envisage that the need for counselling and support will vary widely. We anticipate that the details of support are likely to be determined by the consultand

rather than the genetic clinic. We are acutely aware that professional intervention should preserve and enhance normal support systems.¹² Inevitably some consultands will sever links with the genetic clinic, particularly if it is the source of bad news. For this reason, and for others outlined above, the GP is likely to play a central role in monitoring and mediating the long term consequences of presymptomatic testing for HD.

We are grateful to all the GPs who responded to the questionnaire and took the time and trouble to add their thoughts and comments on predictive testing for HD. We thank the Primary Care Departments of the respective Health Boards for guidance, statistical information, and distributing the questionnaire. Grateful acknowledgement is made to the Area Medical Committees for authorising this study. This work was supported by a grant from the Ludovici Bequest to the University of Edinburgh.

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