Scottish Parliament Region: Lothian

Case 200800801: Lothian NHS Board

Summary of Investigation

Category

Health: Clinical treatment/diagnosis

Overview

The complainants (Mr and Mrs C) complained that Lothian NHS Board (the Board) did not re-test Mr C for Huntington's disease (HD) when new, more accurate, testing was introduced in 1993. Mr C had previously been diagnosed as a likely sufferer of the condition, but received a negative result when re-tested in October 2007. Mr and Mrs C said that their belief that the condition would affect Mr C, and potentially their daughters, caused a great deal of anxiety and led them to make certain life choices. They complained that, had re-testing been provided routinely upon the introduction of more accurate tests in 1993, much of the stress placed on the family would have been avoided and different decisions made about their daughters' future.

Specific complaint and conclusion

The complaint which has been investigated is that the Board did not act reasonably in failing to re-test Mr C for HD following the introduction of more accurate tests *(upheld*).

Redress and recommendations

The Ombudsman recommends that the Board:

(i) remind clinicians of the importance of open discussions of available new genetic tests with affected patients in order to enable them to make informed choices; and

(ii) remind clinicians of the importance of recording such discussions, including relevant information given to patients.

The Board have accepted the recommendations and will act upon them accordingly.

Main Investigation Report

Introduction

1. The complainant (Mr C)'s mother had Huntington's disease (HD), a neurodegenerative condition which develops in later life. HD carries a 50 percent risk of being passed on to one's children.

2. In 1989, Mr C was tested for HD and received a positive result. This positive result meant that Mr C and his wife (Mrs C)'s two daughters were also considered to be at a 50 percent risk of inheriting the condition, which develops in later life. The linkage test, which was used at that time to determine the likelihood of developing HD, was known to carry an error margin of around four percent.

3. In 1993, a new confirmation exclusion test was introduced, which provided results accurate to more than 99 percent. Lothian NHS Board (the Board) did not automatically offer a re-test with the new test to patients that had previously undergone the linkage test. Mr C told me that he enquired about re-testing but was discouraged by his geneticist (Geneticist 1).

4. Mr C was eventually re-tested in 2007 and received a negative result for HD. Whilst he was very pleased to learn that he and his daughters would not develop the condition later in life, he and Mrs C complained to the Board about the decision not to re-test him in 1993, noting that their family had endured a further 14 years of believing that their lives would be affected by this debilitating condition. Dissatisfied with the reasons given by the Board for not providing an automatic re-test following the introduction of improved testing in 1993, Mr and Mrs C brought their complaint to the Ombudsman in June 2008.

5. The complaint from Mr and Mrs C which I have investigated is that the Board did not act reasonably in failing to re-test Mr C for HD following the introduction of more accurate tests.

Investigation

6. In order to investigate this complaint, I reviewed Mr C's clinical records and all correspondence between him and the Board. I also sought further comments from the Board and the opinion of the Ombudsman's professional medical adviser (the Adviser).

7. I have not included in this report every detail investigated but I am satisfied that no matter of significance has been overlooked. Mr and Mrs C and the Board were given an opportunity to comment on a draft of this report.

Complaint: The Board did not act reasonably in failing to re-test Mr C for HD following the introduction of more accurate tests

8. HD is an incurable, hereditary, neurological disorder of the central nervous system. It causes degeneration of the cells, ultimately affecting muscle coordination and cognitive function. HD's symptoms, which include changes in personality, involuntary movements, and difficulty with speech, normally begin after the age of 35-years-old. Children of individuals who have HD have a 50 percent risk of inheriting the condition.

9. Mr C's mother had HD and, on 3 July 1989, the Board wrote to him offering him the opportunity to undergo a linkage test to establish whether he had inherited the condition. The linkage test was a predictive test carried out prior to any symptoms developing, using DNA samples from other family members to predict the likelihood of an individual having the HD gene. Mr C had the test and was found to have a 96 percent chance of having the gene and, therefore, developing the associated symptoms later in life.

10. By the time of Mr C's linkage test in 1989, Mr and Mrs C had two daughters. Whilst it was recognised that the linkage test had a four percent error margin, it was accepted that they were now at a 50 percent risk of developing HD in later life. Mr and Mrs C described to me the impact that a predictive diagnosis of HD had on their family. They told me that their family had made a number of significant decisions as a result of the HD diagnosis 'hanging over' them. Mr and Mrs C's daughters chose not to undergo testing for HD, as they were 'terrified' of finding out the results. However, awareness of the potential onset of HD led to Mrs C and one of their daughters terminating pregnancies. One daughter was unable to complete her university degree. The family also experienced problems obtaining insurance and were unable to move to a better home.

11. Following his linkage test in 1989, Mr C was reviewed by a locum consultant in September 1990. He was considered to be coping with his HD pre-diagnosis relatively well and showed no signs of the disease. He was aged 32-years-old at that time. The locum consultant suggested keeping in touch

with Mr C by arranging another appointment in a year's time but made no recommendations for further treatment at that stage.

12. Mr C attended review appointments every 12 to 18 months following his positive linkage test results. In 1992 a new consultant clinical geneticist, Geneticist 1, started work at the Western General Hospital in Edinburgh (the Hospital), where Mr C attended. Mr C was first reviewed by Geneticist 1 in February 1995 and was found to be entirely well, again with no HD symptoms.

13. Mr C was an active participant in raising awareness of HD and fundraising and attended various HD related events. I understand that he and Geneticist 1 met occasionally at such events and may have discussed Mr C's treatment and HD issues generally, outside of his normal appointments at the Hospital.

14. HD is caused by the mutation of a single gene. In 1993 this gene was identified. New tests were subsequently introduced that establish whether individuals carry the mutated gene, allowing geneticists to confirm with practically 100 percent accuracy whether an individual will develop the condition in later life. Mr and Mrs C told me that through his involvement with HD support groups, Mr C became aware of the new test, but upon raising the possibility of being re-tested with Geneticist 1, she advised that there would be no benefit to him of undergoing the new test. This discussion is not recorded in Mr C's clinical records.

15. Once Geneticist 1 took over his care in 1995, Mr C continued to attend review appointments every 12 to 18 months. At each of these, he was recorded as showing no signs of the symptoms associated with HD and was generally noted as being 'well'. At his review appointment in June 1996, Geneticist 1 noted that Mr C reported some deterioration in his ability to concentrate. However, this was not impacting on his work and he was generally in good health.

16. In August 1997, Geneticist 1 suggested that Mr C undergo some tests to establish baseline measurements of his speech, language and neuropsychological (brain function) abilities. It was proposed that these tests would be carried out whilst Mr C showed no symptoms and that they would then be repeated annually to monitor changes in his condition.

17. Mr C underwent speech and language tests on three occasions in September and October 1997. Upon completion of the tests, he was found to have some mild reduction in tongue function and language, but his results fell within the normal range. He was noted as showing no obvious signs of reduced abilities.

18. Neuropsychological tests were carried out in November 1997. Again, Mr C performed within the normal range of results, however, some minor deficits were noted in his verbal and visual memory. It was also noted, however, that Mr C was extremely anxious at times during the tests and that this could have affected his performance.

19. Between 1997 and 2007, Mr C continued to undergo annual tests to identify any changes in his condition. He also continued to attend annual review meetings with Geneticist 1. On each occasion his condition remained unchanged and further cognitive (information processing) tests in 2005 found him to be symptom free.

20. On 31 July 2007, Geneticist 1 wrote to Mr C following a recent review appointment. In her letter, she stated 'When the Huntington's gene was originally cloned we talked about whether you would like to have a test to see the number of repeats that you have. At that time you did not think that it would be of any particular benefit to have such a test. Given that you are still symptom free I wonder whether you should be considering a re-test. After all we know that the original test that you had was quoted to have an error risk of 4%'. Mr C agreed to the re-test, as a negative result would have major implications for his children.

21. Mr C's re-test results showed that he did not have the expanded HD gene and that he would, therefore, not develop the condition. This also meant that his daughters were not at risk of inheriting HD from him. Mr C received his test results at the Hospital on 23 October 2007. He and his daughters then attended a meeting with Geneticist 1 on 7 November 2007 to discuss the implications of the reversal of his diagnosis. Geneticist 1 noted after this meeting that Mr C and his family were finding it difficult to absorb the information but they were coping well and further support was available to them from the Board's HD adviser. Geneticist 1 made no further appointments to see Mr C.

22. Mr C wrote to the Board on 9 April 2008. He explained that, whilst he was delighted that he and his family would not be affected by the condition, the predicted diagnosis of HD had had a major impact on the family, leading to certain significant life choices being made. He felt that his family had been put under unnecessary strain which could have been avoided had he been re-tested earlier. Mr C asked the Board three specific questions:

> 'a. In 1993 why were tests not automatically offered to asymptomatic clients who had previously received a positive test result?
>
> b. Why, when I did request to be retested, was I discouraged against this by my geneticist who indicated there was no need?
>
> c. Why have I had the knowledge for the last 18 years of knowing I carried this faulty gene when the reality is that the mistake could have been rectified after 4 years?'

23. In his letter to the Board Mr C further noted that at a recent meeting with Geneticist 1 and his daughters (it is unclear whether this is the 7 November 2007 meeting), Geneticist 1 'apologised profusely and stated that she could not understand why she did not offer a retest'. Furthermore, 'she stated that another geneticist had contacted all their clients by letter offering a retest and this other geneticist questioned why this had not been carried out by [Geneticist 1]'.

24. The Board responded to Mr C's letter on 2 June 2008. They explained that in 1993, when the HD gene was cloned, genetics department staff held weekly meetings to discuss results and policies for genetic testing. The Board had a number of patients in their database who had undergone pre-symptomatic testing for HD using the linkage test. They said that the genetics team discussed whether to re-contact patients to ask if they wanted a re-test, or whether this would be distressing in terms of revisiting the issue and providing false hope. Concerns were reportedly raised as to whether the Board would only contact patients that had received a positive linkage test result, or whether those that had tested negative should also be contacted. The Board said that, as Geneticist 1 was relatively new to her post at the time of these discussions, she did not know the majority of her patients personally and felt unable to make an assessment of the potential impact of re-contacting patients who had previously been tested. A decision was, therefore, made by the department to re-test any patients that requested re-testing, but not to actively contact patients on the database who had been tested prior to 1993.

25. The Board advised Mr C that they were aware that a geneticist in Aberdeen (Geneticist 2) had decided to offer all patients the option of re-testing. They noted, however, that Geneticist 2 had been in her post for several years and knew the patients that had undergone the linkage test. The Board further advised that other health boards (Manchester, Cardiff, Exeter and Oxford) also decided against systematically approaching patients for re-testing.

26. The Board commented on Geneticist 1's recollection of events. They said that she could not recall discussing the possibility of a re-test with Mr C, or actively discouraging him from this. Geneticist 1 did, however, note that she had met with Mr C at a number of Scottish Huntington's Association events and, in hindsight, felt she could have inadvertently given the impression that repeat testing was unnecessary by reinforcing that the linkage test was likely to be accurate during general discussions about testing. The Board noted that it was Geneticist 1's policy to arrange repeat tests when these are requested by patients. Geneticist 1 reportedly noted at the July 2007 review appointment that Mr C continued to show no signs of HD, despite being nearly 50 years of age. This led her to reflect that Mr C had never had his diagnosis formally confirmed under the newer test, prompting her to contact Mr C on 31 July 2007. The Board told Mr C that it was a source of personal regret to Geneticist 1 that she did not suggest a re-test sooner. The Board added their own unreserved apologies to those expressed by Geneticist 1 at the 7 November 2007 meeting for the impact that this situation had on Mr C and his family.

27. Mr and Mrs C complained to the Ombudsman in June 2008, advising that they were dissatisfied with the Board's explanations. I asked the Adviser to review Mr and Mrs C's complaint and to provide comments about HD testing, generally, and about Mr C's specific experiences. The Adviser reiterated the impact that HD can have on families and acknowledged Mr and Mrs C's comments regarding the decisions that their family had made as a result of Mr C's predictive diagnosis. He also noted that whilst the newer, more accurate genetic tests were welcomed in the medical world due to their precision, not all patients wished a certain diagnosis of HD and only a minority of individuals at risk of developing the condition present for testing.

28. The Adviser stressed that the original linkage test that Mr C underwent did not provide an incorrect result. Rather, it predicted the likelihood of Mr C having the mutated HD gene. The 96 percent likelihood was an accurate prediction, however, Mr C fell in the four percent rather than the 96 percent.

29. The Adviser confirmed that there is no recognised, general, approach to utilising new tests or treatments. He considered it likely that most genetic labs would have considered re-testing all patients following the introduction of new HD tests in 1993 but noted that few laboratories would have the ability to perform such tests at that time. He advised that it would be good practice for the Board to align their approach with a 'responsible body of medical opinion' and was satisfied that their awareness of the approaches of other laboratories achieved this. He considered the four laboratories that the Board referred to in their correspondence with Mr and Mrs C represented a responsible body of medical opinion.

30. The Adviser said that he would have expected Geneticist 1 to have discussed the new test with her patients, but not to have encouraged them one way or the other. He acknowledged that Geneticist 1 recalled this being the case, but also that Mr C had a significantly different recollection of events. The Adviser suggested that a record of such discussions could be kept in patients' clinical records.

31. I asked the Board whether their laboratories had the ability to perform the new tests upon their introduction in 1993, and whether this would have been available to Mr C, had he specifically requested a re-test during his early consultations with Geneticist 1. The Board confirmed that they were able to perform the test from as early as May 1993 and that a re-test would have been performed if Mr C had requested it. The Board clarified that, from 1993, the new test was offered to all individuals that requested confirmation of their diagnosis or pre-symptomatic testing for HD. Edinburgh was the testing centre for Scotland. There was no formal procedure for retrospective re-testing of individuals that had previously undergone pre-symptomatic HD tests and no clinical request was made by other Scottish Genetic Centres for systematic re-testing of previously tested individuals.

32. The Board told me that their approach reflected practice across the UK at that time. They explained that, in 1993, the linkage test's 96 percent accuracy matched or exceeded the accuracy of the majority of other genetic tests performed at that time. The new test was not considered to be considerably more accurate than the linkage test and patients had been counselled on the estimated four percent risk of their HD prediction being inaccurate. The new test was still being validated around the world and up until 1997 there remained

some doubt as to the test's ability to predict the onset of HD. Furthermore, the 'potential trauma to patients who had assumed and lived with the low risk of an inaccurate HD prediction was considered to be inappropriate compared to the benefit of reversing a very small number of predictions'. The Board advised that they do not have a blanket policy of not re-testing patients when improved genetic tests become available. Rather, a decision is made regarding the significance of the improvements that will be achieved by re-testing.

33. I asked the Board about their comments in their letter to Mr and Mrs C, dated 2 June 2008, which explained that Geneticist 1 had taken an active decision not to contact previously tested patients as she did not feel that she was familiar enough with each individual to determine their reaction to an invitation to re-test. They told me that no firm decision was made by Geneticist 1 in this regard. Patients were reviewed on an ad-hoc basis in the Genetics Clinic and in some cases a decision was made by the patient to go for a re-test. The Board noted that, whilst Geneticist 2 contacted all of her patients to offer a re-test, a significant proportion of her patients opted not to be re-tested, preferring to hold on to the hope that they fell in the four percent of patients that had been incorrectly predicted as having HD. The Board told me that, in retrospect, Geneticist 1 felt that she placed too much emphasis on Mr C's understanding of HD due to his involvement with the local HD Association. It was a source of great regret to her that she did not actively raise the subject of re-testing sooner.

34. I provided Mr and Mrs C with copies of the comments made by the Adviser and the Board during my investigation. Mr and Mrs C disputed the Board's assertion that they actively decided not to contact patients that had previously undergone the linkage test, and felt that this position was reached by default. They also felt that, as Geneticist 1 became more familiar with her patients over the course of a number of consultations, she should have been in a position to advise patients to be re-tested.

35. Mr C understood that a re-test could have reinforced his HD diagnosis and acknowledged that other patients could have received a positive diagnosis, having previously tested negatively. He complained, however, that he was not given the opportunity to make a decision as to whether or not he wished to be re-tested. He felt strongly that all patients should be given this choice. Mr C confirmed that his involvement in HD was limited to raising awareness of the condition and fundraising for families affected by it. He did not have a detailed

knowledge of the genetics of the condition and relied upon Geneticist 1 to advise him in this regard.

Conclusion

36. I consider there to be two main questions arising from Mr and Mrs C's complaint: was the Board's general policy of not offering re-testing to all previously tested patients reasonable; and were their actions with regard to Mr C's specific case reasonable?

37. With regard to the Board's general approach, I have seen no documented evidence of their having actively considered and rejected the option of contacting all patients that had been previously tested for HD under the linkage test. That said, I do not consider it necessary for this to be specifically documented and I am satisfied that the Board's subsequent explanations indicate an understanding of relevant considerations at that time.

38. I accept the Board's and the Adviser's comments regarding the acceptance of the linkage test as being an accurate method of predicting an individual's likelihood of having HD. I further acknowledge the Board's comments regarding the doubts over the new test for a number of years following its introduction. When considering whether to contact all previously tested patients following the introduction of the new test in 1993, the Board would have to consider the likelihood of previous test results having been inaccurate. I am satisfied that it was reasonable to conclude that very few individuals would fall in the four percent error margin. However, I also recognise the impact on those individuals, such as Mr C, who did fall in the four percent error margin.

39. No guidance exists to advise health boards on when patients should be invited to re-test following the introduction of improved genetic tests. The Adviser felt that the Board's approach to re-testing should be supported by a responsible body of medical opinion and was satisfied that this was the case. I acknowledge Mr and Mrs C highlighted two other laboratories that did decide to contact all patients. In the absence of formal guidance or statutory obligation on health boards to offer re-testing automatically, I consider this decision to be one that requires the professional judgement of genetic laboratory staff. Whilst Mr and Mrs C do not agree with the decision that the Board reached in this regard, I am satisfied that it reflected similar approaches taken by other health boards, and that relevant factors such as the number of patients that would be

affected and the accuracy of the old test were taken into account. I found the Board's general policy of not automatically contacting all patients to suggest a re-test to be reasonable. However, I consider that it is important that geneticists discuss with patients the implications of improved tests as they become available. Only this can allow patients to make informed decisions about testing.

40. Mr C's personal situation was extremely rare, if not unique. He fell in the four percent of inaccurate HD predictions. Whilst I found the Board's general policy on re-testing to be reasonable, I acknowledge the impact that this had on the lives of Mr and Mrs C and their family, and the significant life decisions reportedly taken by family members based on the reasonable belief that Mr C had HD. However, I have seen no evidence of the Board having been involved in these life choices. I can only consider matters for which the Board has responsibility and consider whether the Board carried out these responsibilities in a reasonable way.

41. The Board advised that re-testing was available to patients that requested it during their consultations. Geneticist 1's comments, relayed through the Board's correspondence with the Ombudsman's office, indicate that she made an assumption, based on previous conversations with Mr C, and his active involvement with HD organisations, that he would know about the new test and its relevance to his own situation. Ultimately, Geneticist 1 approached Mr C and suggested a re-test.

42. I do not consider that it was appropriate or necessary for the re-test to be suggested as early as 1993. There was no indication at that time that Mr C should be an exception to the general policy of not offering re-testing. He was 35 years of age, so only just arriving at an age where symptoms of HD could begin to show. It was not surprising that he was asymptomatic and the linkage test was considered to be accurate. As time passed Mr C continued to attend review appointments and undergo neuropsychological and cognitive testing. Whilst he remained within the normal range of test results, some minor deficits were recorded and these may have been interpreted as an indication of HD. It was not until Mr C was 50 years of age and should have been demonstrating symptoms of HD that Geneticist 1 suggested a re-test.

43. It is impossible to say with any conclusiveness when Mr C should have been offered a re-test. On the one hand, he was recorded on an annual basis

as showing no symptoms of HD and was ultimately offered a re-test in the light of this. On the other, there was no cause to single him out for re-testing as early as 1993 and the nature of the condition is such that there is no set age at which the symptoms should develop. As such, one cannot determine a specific point at which Geneticist 1's normal monitoring of symptoms should have changed to proactive testing to confirm the HD diagnosis.

44. Generally, I consider that Mr C could have been offered a re-test earlier than he was. In expressing regret at not having suggested a re-test sooner, Geneticist 1 appears to accept this. I have seen no evidence indicating that Mr C was made aware of the new test and its implications by Geneticist 1. It would not be sufficient for the Board to make no mention of this and wait until individuals raise the subject. The evidence that I have seen suggests that Mr C was aware of the new test, and that discussions could have taken place between him and Geneticist 1 which may or may not have dealt with the specific issue of him being re-tested. There is, however, no formal record of such conversations.

45. Mr C's personal circumstances were very rare and should be considered separate to the wider issue of the Board's general policy. I found that general policy to be reasonable and consider it adequate to offer re-testing as an ad-hoc option during patient reviews. However, there is no clinical record of any discussion between Geneticist 1 and Mr C about the new test. The new test first became available when Mr C was aged 35-years-old. He was not offered the new test until he was 50-years-old. During this period, Mr C showed no symptoms of HD. Exactly when Mr C should have been offered the new test is a matter of clinical judgement, however, my view is that the Board waited too long before doing so and for this reason I uphold the complaint.

Recommendations

46. I acknowledge that the Board have already apologised to Mr and Mrs C for not offering him the new test sooner.

47. The Ombudsman recommends that the Board:

(i) remind clinicians of the importance of open discussions of available new genetic tests with affected patients in order to enable them to make informed choices; and

(ii) remind clinicians of the importance of recording such discussions, including relevant information given to patients.

48. The Board have accepted the recommendations and will act on them accordingly. The Ombudsman asks that the Board notify him when the recommendations have been implemented.

Annex 1

Explanation of abbreviations used

Mr and Mrs C	The complainants
HD	Huntington's disease
The Board	Lothian NHS Board
Geneticist 1	A geneticist at Lothian NHS Board
The Adviser	A professional medical adviser to the Ombudsman
The Hospital	The Western General Hospital, Edinburgh
Geneticist 2	A geneticist at Aberdeen NHS Board