

# The accuracy of diagnoses as reported in families with cancer: a retrospective study

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## Abstract

**Assessment of risk of developing hereditary cancer and subsequent clinical surveillance is largely based on family history. It is considered standard practice to confirm as many diagnoses as possible in cancer families. Our aim was (1) to assess inaccuracies in reporting of cancers by families, (2) to assess the need for confirmation of diagnosis, and (3) to estimate how many families would have been entered unnecessarily or excluded from screening. A retrospective study of 595 case notes was performed in two centres. Methods of confirmation included information from the cancer registries, death certificates, hospital notes, and histopathological records. Accuracy varied by site of cancer and by the closeness of the relationship to the affected person. Reported abdominal malignancies were inaccurate in 20%, whereas 5% of reported breast cancers were inaccurate. In two families the family history of cancer proved fictitious. Management was altered in 23/213 (11%) families following cancer confirmation. The results of the study favour verification of cancer diagnoses particularly if decisions regarding surveillance or prophylactic surgery are based on the family history.**

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There has been a considerable increase in referrals to genetics departments of people with family histories of cancer over the last five years.<sup>1,2</sup> It is important to establish in these families whether they are at increased risk because of genetic susceptibility or whether the cancers in a particular family reflect the fact that cancer is a common disease of which the vast majority occurs sporadically. Estimates of risk are made on the basis of the family history and decisions regarding whether or not to consider both surveillance and prophylactic surgery may subsequently be taken. Inaccuracies in reporting will inappropriately influence these decisions unless verification occurs. As screening remains an area of research, it is important that the information about families entered into trials of screening is accurate, so that risk and the effects of screening on natural history of the cancer in question can be correctly evaluated.

The need or otherwise for confirming diagnoses has financial implications both in

terms of data retrieval and also in terms of clinical screening costs. Assessment of the financial aspects can only be made possible by an evaluation of the need for confirmation and the routes used.

Accuracy in family history taking may also have wider implications in terms of epidemiological studies aiming to elucidate aetiological factors in cancer.<sup>3</sup> Gene mutations that cause cancer predisposition may have a role in oncological therapeutic studies in that particular mutations may confer resistance or sensitivity to particular drugs.

Our aim was therefore to (1) estimate the accuracy of reported family history, (2) assess the need for confirmation, and (3) estimate how many families would have been entered unnecessarily into or excluded from a screening or surveillance programme.

## Materials and methods

### SUBJECTS

Two regional cancer genetics services were used in the study. The Genetics Department based in Newcastle upon Tyne during the period of study had one cancer family clinic per week; other families with a history of cancer were also seen in peripheral clinics. In Newcastle, cancer confirmation was sought after the clinic. The Medical Genetics Department at St Mary's Hospital, Manchester has two clinics per week specifically for referrals regarding a family history of cancer. The Christie Hospital, Manchester also has a specific family cancer clinic. In the Manchester hospitals, information about the family history was collected before the clinic attendance and cancer confirmation is also attempted before the clinic.

The study population consisted of 400 cancer family case notes from the Newcastle department which covered the two year period of 1994 and 1995, 100 consecutive case notes from the Christie Hospital, and 95 case notes from St Mary's in which the referral was for a family history of cancer. There was no selection made on the basis of age or sex of the proband. The overwhelming number of probands were female so no analysis was made regarding different abilities in recall between men and women. Nor was there any selection made regarding whether the proband had cancer or not.

### MEASURES

This was a retrospective study comparing reported diagnoses with confirmed diagnoses. Verification was sought through death certificates, medical notes, histopathology, and cancer registries. If the referring doctor had been

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Table 1 Reasons for failing to confirm diagnoses

|  | Newcastle | Manchester |
|--|-----------|------------|
| No response from medical records or registry or notes unobtainable | 20.4%     | 32%        |
| Insufficient detail  | 78.5%     | 39.4%      |
| Diagnoses too long ago   |           | 16.7%      |
| Relatives overseas   |           | 9.7%       |
| Relative refused permission  | 0.9%      | 2.2%       |

involved in the treatment of an affected proband or relative the referring letter was accepted as confirmation. If confirmation was not obtained the reason was noted.

### Results

The total number of case notes studied was 595. Confirmation was sought in 408 (68.5%) case notes. Of those notes where confirmation was obtained, the diagnosis was accurate in a total of 85%. The probands reported 1811 cancers in relatives. The probands themselves were affected in 74 cases. Confirmation was confirmed in 791/1885 (42%) cancers. Of the total, 688 involved the breast, 872 were abdominal malignancies, and the remainder were at other sites, the majority of those being lung cancer. The number of affected relatives varied between one and 13.

#### INFORMATION TRACING

There was slight variation in technique regarding seeking confirmation. Confirmation was not obtained for the reasons shown in table 1. The two sites are indicated separately as slightly different reasons were assigned. In Manchester, confirmation was sought for all affected family members. In Newcastle, confirmation was sought in 213/400 families. Of the remainder, 96 were at low risk from the family history, 16 failed to attend, and confirmation was not sought in a further 75, there being no specific reason beyond it not being routine policy. In the majority of these 75 families, either the proband or accompanying relative had cancer and was aware of their diagnosis. Confirmation was also not sought if the affected relative or relatives were alive and permission was not given to confirm the diagnoses.

#### DIAGNOSIS CONFIRMATION

The route of confirmation was recorded in one of the centres (Newcastle). Diagnosis was confirmed in 429 subjects (table 2). In 12 subjects it was indicated in the case notes that confirmation was obtained but the relevant confirming document had not been photocopied. Histology was indicated as the route of confirmation if this was photocopied and in the genetics notes; if it was simply the notes that had been photocopied and not including the histology report then it was classed as confirmation from the medical records. This occurred if there were several sets of notes at various hospitals and the original notes with the histology report were unobtainable.

#### ACCURACY OF REPORTED DIAGNOSIS

We divided the cases into cancer by site. Breast cancer was considered as one group, abdominal malignancy as another group, and cancers

Table 2 Routes of confirmation

| Route of confirmation         | Percentage confirmed by that route |
|-------------------------------|------------------------------------|
| Referral letter               | 23.8%                              |
| Cancer registry               | 23.8%                              |
| Histology                     | 18.6%                              |
| Other genetics department     | 10.7%                              |
| Medical records               | 10.5%                              |
| Death certificate             | 9.8%                               |
| Route of confirmation unclear | 2.8%                               |

Table 3 The variation in accuracy of reported diagnoses depending on the degree of relationship

| Degree of relationship | Degree of accuracy |            |
|------------------------|--------------------|------------|
|                        | Newcastle          | Manchester |
| 1st degree             | 86.1%              | 94%        |
| 2nd degree             | 78.4%              | 82%        |
| 3rd degree             | 59.3%              | 81%        |

elsewhere were grouped together under the heading "other". Abdominal malignancy included bowel, ovary, endometrium, pancreas, and prostate. The two centres had close agreement regarding accuracy of breast cancer and abdominal malignancy; in Newcastle 94.9% of breast cancer reporting was accurate and in Manchester 94% was accurate. For abdominal cancer, 81.4% of diagnoses were accurate in Newcastle and 81.8% in Manchester. There was a difference in accuracy of reporting of "other" cancers between the two sites: 88.2% of "other" cancers were accurate in Newcastle compared with 69.2% in Manchester. In this group, further analysis had been undertaken in the Manchester cohort and the results are as follows. For prostate cancer, 100% of those traced were confirmed. For lung cancer, 83% of those traced were confirmed. For throat cancer, 50% of those traced were confirmed. This compares with 90% of bowel cancers that were traced being confirmed and 83% of ovarian cancers being confirmed. Accuracy of reporting was also influenced by closeness of relationship. In Newcastle, we assessed this for all confirmed cases of cancer and in Manchester this question was addressed for the cases of breast, bowel, and ovarian cancer. The results are shown in table 3.

In Newcastle, the errors in reporting were also analysed to elucidate how many cases of cancer were in fact benign disease as opposed to those cases where the error was simply in the type of cancer reported. Four (3%) of the misreported breast cancer cases, six (2.4%) of abdominal malignancies, and two (2.9%) of other cancers were benign disease reported as cancer. This emphasises that most recorded cancer illnesses were malignant and it was the site that was erroneous. In Manchester, it was noted that in 10 cases where one cancer was confirmed another reported cancer in the same person was either secondary spread or a benign disease. However, in 17 cases a second primary was identified at confirmation that was not reported by the family.

In two of the Manchester families, it was found that the proband or a relative had fabricated the family history.<sup>1</sup> In one of the

Newcastle families, a relative had misled the proband, possibly unintentionally.

#### ALTERATION OF MANAGEMENT

This was only assessed in the Newcastle cohort. Management was altered in 23/213 (11%) families. Screening was initiated in eight families and deemed unnecessary in 15 families.

#### Discussion

When considering the clinical management that is recommended in the common cancer syndromes (breast/ovarian and bowel cancer), most genetics departments have guidelines about which families merit screening. In cancer genetics clinics, screening advice is based on the family tree. It is thus important that this is accurate. While the majority of reported cases were accurate, a substantial proportion of reported abdominal malignancies were inaccurate and may have triggered unnecessary screening or inappropriate reassurance without confirmation. Confirmation was not achieved in a large proportion of the notes and information tracing was more difficult on more distant relatives, not surprisingly also the group with the greatest inaccuracy in reported diagnoses. This corroborates other studies regarding accuracy of reported diagnoses and the closeness of the relationship.<sup>4,5</sup> Bondy *et al*<sup>1</sup> reported a difference in accuracy between first and second degree relatives; in fact, in their study there was over-reporting of cancers in the second degree relatives.<sup>4</sup> There was a similar variation in accuracy between first, second, and third degree relatives in another study.<sup>5</sup> One of the main reasons for difficulty in record tracing was the lack of crucial information known by the proband about affected relatives. In order to obtain medical notes or information from cancer registries, it is necessary to know date of birth, full name, date of death, hospital where treated, and address if possible. Even in the tight knit communities of the north east, this information was usually only available for first or second degree relatives. It is also a notable problem that most hospitals now operate destruction policies for medical records and notes are being kept for shorter and shorter periods of time. Pathology records are stored for longer, but in order to access them the date of the operation is usually required, information that is almost always unobtainable. If a relative is still alive, consent forms are needed for release of any information and these are not always easy for the consultand to obtain.

The error in reporting of cancers is also affected by cancer site. The small error in breast cancer reporting, 5.5% when the results from Newcastle and Manchester were combined, is not surprising as there are few other breast diseases which cause such fear and therefore it is likely that relatives will remember the diagnosis easily. Mastectomy also results in a visible change in the shape of a woman, although it could be argued that this is less so now with improvements in prostheses, the increasing use of lumpectomy, and advances in reconstructive surgery. There was a much

greater error in reporting of abdominal malignancies, 19.4% for the two geographical regions combined. This was not unexpected as there are more organs within the abdominal cavity and words that have specific medical meaning may have a more vague meaning for the general public. The word stomach is an example where there is a specific medical meaning and a much vaguer lay meaning.<sup>6</sup> A difference in patients' and doctors' interpretations of medical words was reported by Boyle.<sup>7</sup> It has also been shown that patients have poor knowledge of the site of internal organs,<sup>8</sup> both of which could add to misconceptions about cancer site. These findings are similar to an Australian study which found that 77% of positive family histories relating to colorectal cancer and as reported by subjects undergoing colonoscopy were confirmed as accurate.<sup>9</sup> This gives a similar magnitude of error to our own study.

It is interesting that there is a big variation between the two sites in terms of accuracy of other cancer sites; for Newcastle 11.8% of diagnoses were wrong and for Manchester 30.8% were wrong. If we consider the Newcastle group alone, this may reflect that the majority of cancers in this group were in the lung or brain and patients are better at knowing the sites of these organs than those in the abdominal cavity.<sup>8</sup> In Manchester there was a greater spread of cancer sites, which may account for the variation.

It is worth noting that in the Newcastle series of patients, management was altered in 11% (23/213) of the families as a result of investigating diagnoses. For 15 of these families screening was no longer regarded as necessary. This has implications for the health of these subjects in that screening itself may not be without risk.<sup>10</sup> It also has implications for psychological well being. Finally, there are financial implications regarding the cost of screening.

There were only three cases of fictitious family history, two of which have been the subject of a case report.<sup>1</sup>

As public awareness of the possible genetic nature of some cancers grows, this may become a more common problem and underlines the need for confirmation of diagnoses in enough relatives to justify screening. In practice, it is usually necessary to confirm diagnoses in first and second degree relatives, the group of relatives for whom it is usually possible to obtain sufficient information.

#### Conclusion

(1) Family history can be inaccurate. This probably reflects lack of medical knowledge in the general population and mechanisms of communication within families. Rarely it is because of fictitious reporting.

(2) We recommend that diagnoses are confirmed in first and second degree relatives to justify any clinical screening. In practice, this should be achievable as most patients have access to sufficient information about these relatives, it is unusual to require information

from more distant relatives, and in such situations each family tree should be judged with regard to that particular situation.

Since undertaking this study we have introduced preclinic questionnaires in Newcastle and that has improved the ease with which diagnoses can be confirmed. We also ask on these questionnaires to see any relevant death certificates. We would therefore recommend using a proforma for collection of information concerning affected relatives that will be sufficient to obtain information from the cancer registries or through medical records. In the Northern region, confirmation by use of cancer registries using a standardised form greatly increased efficiency of time spent confirming diagnoses in these families and we would recommend it as a first line in attempting to confirm diagnoses, unless the family or referring doctor provide definite confirmation.

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