Cancer genetics clinics and the surgeon: a valuable role for family history screening

GL WILLIAMS¹, J GRAY², J BEYNON¹

¹Department of Colorectal Surgery, Singleton Hospital, Swansea, UK

²Cancer Genetics Service Wales, Cardiff, UK

ABSTRACT

INTRODUCTION This study examines how a colorectal surgeon can use a regional cancer genetics service to deal safely and efficiently, with community referrals for colorectal cancer screening on the basis of family history.

PATIENTS AND METHODS A retrospective review of consecutive asymptomatic people with a strong family of colorectal cancer referred by the surgeon to the genetics service over a 30-month period.

RESULTS A total of 45 people were referred by the surgeon to the cancer genetics service. Following official verification of family histories, 15 were thought to be in a low-risk category for developing colorectal cancer, 18 were moderate risk, 4 had a high-to-moderate risk and 2 satisfied the criteria for HNPCC. After official authentication, it was discovered that 20% of people had mistakenly informed the surgeon of important inaccuracies in their family history.

CONCLUSIONS The cancer genetics service seeks to identify accurately those at increased risk of developing colorectal cancer due to their family history. It has the time, resources and expertise to verify officially a family history that cannot be properly done in a busy surgical clinic. This study shows that it can provide a valuable role for correctly identifying and counselling people who truly require screening due to their familial predisposition for colorectal cancer.

KEYWORDS

Family history – Screening – Cancer genetics – Colorectal cancer

CORRESPONDENCE TO

GL Williams, Department of Colorectal Surgery, Singleton Hospital, Swansea SA2 8QA, UK

T: +44 (0)1792 285459; F: +44 (0)1792 208647; E: geth_williams@yahoo.co.uk

Colorectal surgeons are frequently being referred asymptomatic people with a family history of colorectal cancer to their out-patient clinics due to their increased risk of developing the disease. From this single surgical consultation, decisions are made regarding advice on screening or surveillance based on the individual's estimated life-time risk calculated from the family history given. It is known, however, that the family history ascertained at the initial out-patient appointment can be inaccurate.¹ It has also been questioned whether all colorectal surgeons are capable of providing a safe screening service based on family history criteria.²

Personal risk of colorectal cancer can be estimated from family histories using empirical risk data. Indirect evidence suggests a small benefit for colonic surveillance for people with: (i) one first-degree relative affected with colorectal cancer < 45 years; or (ii) with two affected first-degree relatives. People satisfying these criteria are considered to be at an increased risk and, therefore, should undergo colonoscopic surveillance.³

The Cancer Genetics Service in Wales (CGSW) is similar to the other regional cancer genetics services throughout the

UK. It provides a service for individuals who meet the criteria of referral on account of their suspected family history of colorectal cancer or other inherited cancers. Its role is to calculate the estimated life-time risk of developing cancer based on detailed and accurate verification of an individual's family history from official sources. The CGSW will then liaise with the colorectal specialist to recommend a particular and appropriate screening strategy for that person.

The aim of this study was to evaluate the outcome of people referred by one consultant colorectal surgeon to the regional Cancer Genetics Service. We sought to compare people's precision of reporting their own family history in the surgeon's clinic to the history officially verified from patient records by CGSW and also whether patient management would have been affected by any inaccuracy.

Patients and Methods

All asymptomatic people with a family history of colorectal cancer referred for advice by their family doctor regarding life-time risk estimation and screening had their family

Table 1 Referral criteria to CGSW for colorectal cancer

One first-degree relative diagnosed at age 40 years or less
Two first-degree relatives at 60 years or less (on same side of family)
Three relatives on same side of family (at least one should be a first-degree relative)
Familial adenomatous polyposis
Hereditary non-polyposis colorectal cancer

history documented by the consultant surgeon in clinic. Over a 30-month period between June 1999 and December 2001, all people with a family history satisfying the criteria set by CGSW were referred on by the surgeon for a more accurate risk assessment (Table 1). Those people suspected by the surgeon of being part of a dominantly inherited familial adenomatous polyposis (FAP) or hereditary non-polyposis colorectal cancer (HNPCC) pedigree were excluded from the study cohort so that the study could focus on the more ambiguous ‘familial’ group more commonly encountered in routine practice.

On receipt of a referral, CGSW send a detailed questionnaire to the patient’s address to identify the full family history and other baseline information. This is then verified against medical records and death certificates to obtain accurate life-time risk estimation. A genetic counsellor will have a telephone consultation with the patient to discuss the findings and to categorise the patient into either high (approximately 1:2 life-time risk of dying from the disease), moderate (~1:6) or low (~1:12) risk groups. The consultant surgeon, GP and patient all receive a detailed letter explaining and providing information on risk grouping and what screening strategy, if any, is needed for the individual (Fig. 1).

Figure 1. Pathway for colorectal family history referrals.

Only the high-risk group will be offered a clinic appointment with a consultant cancer geneticist.

Results

The consultant surgeon referred 45 appropriate people to CGSW over this time period based on the one clinic consultation. Their median age was 49 years (range, 23–61 years) and 60% were male. On two occasions, 6 refused to answer the questionnaire sent by CGSW. Of the remaining 39 whose family histories were accurately and officially collected by CGSW, 15 were found to be in a low-risk category, 18 were of moderate risk, 4 were termed as high-to-moderate risk and 2 unexpectedly satisfied the criteria for HNPCC.

In comparison with information obtained at the surgical clinic, 3 people were downgraded from a moderate- to low-risk group. Another 5 had underestimated their family history: 3 were changed from low- to moderate-risk and 2 were diagnosed with HNPCC from their accurately verified family history. Therefore, 8 (20%) people had unwittingly provided the consultant surgeon with incorrect information regarding their own family history.

Discussion

Heightened public awareness of ‘familial’ colorectal cancer has led to increasing demand for colorectal surgeons to see asymptomatic people in their already overstretched outpatient clinics. In addition, studies have shown that the history provided is often inaccurate and that some form of official verification should be sought.⁴⁻⁶ A given history is often deficient due to various factors such as lack of time, patient nervousness, poor recall, poor interview technique or failure to consult other family members.

This study of non-FAP and non-HNPCC relatives demonstrates that 20% inaccurately reported their family history in the single interview with the surgeon, thus having implications for their further management. Some of these would have been exposed to more intensive screening than they should have received, whilst others would have been under-screened. This may have had far-reaching consequences for these people and the healthcare system including psychological, physical, resource and financial considerations. Incorrect colonoscopic screening strategies would have been adopted for these individuals if they had only seen the surgeon rather than being assessed, in detail, by the Cancer Genetics Service.

The Cancer Genetics Service provides a safe, replicable, accurate and acceptable system of managing asymptomatic people with a family history of colorectal cancer.⁷ Accurate verification of the correct estimated life-time risk by this method allows more efficient use of colonoscopic services

and frees out-patient space for symptomatic colorectal patients. The Cancer Genetics Service enables community referrals solely on the basis of family history to be centralised and audited and any advice given will thus be standardised and protocol driven across a region.⁵ It also has more time, resources and trained personnel available to discuss issues at length with worried individuals.

Conclusions

This study shows that family history obtained from an asymptomatic relative at a surgical clinic may be inaccurate in 20% of cases. Surgeons who aim to screen asymptomatic people with a family history of colorectal cancer should enlist the services of their regional Cancer Genetics Service to prevent inaccurate and inefficient screening.

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