

How can we improve the quality of reporting family history in colorectal cancer care? (And why does it matter?)



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“The overall consequences of widely adopting family history in risk assessment are influenced by the underlying prevalence of colorectal cancer, and the effectiveness of existing policies on prevention, screening and treatment.”

Family history & colorectal cancer

Family history (FH) represents the integration of shared genomic and environmental risk factors [1]. First-degree relatives share half of their genomic information, as well as ways and places of living and working. Even if the molecular etiology of a condition is not fully understood, FH offers insight into disease susceptibility, and may provide a rational basis for personalizing preventive and healthcare interventions [2]. FH is, of course, the primary marker of genetic syndromes such as familial adenomatous polyposis and Lynch syndrome (hereditary nonpolyposis colorectal cancer [CRC]), but these account for no more than 6% of all CRC cases [3]. To this familial clustering should be added thought to be due to (as yet unidentified) genetic variants, which may account for a further 15–25% [4,5].

Setting aside known genetic syndromes, however, there is clear evidence that having just one affected first-degree relative raises a person's lifetime risk of CRC by a factor of approximately two, further

increased with additional affected relatives and earlier age of diagnosis [6,7]. There is also evidence of longer survival in colon (but not rectal) cancer patients with a positive FH [8–12], although this has not been confirmed in all studies [13–15]. It has also been observed that CRC patients with a positive FH may have a higher incidence of secondary cancers [8,12,16]. These findings are thought-provoking, but require further investigation.

Should FH be used routinely in CRC care?

The main area for considering FH in CRC care is in risk assessment and surveillance. Some argue that taking a FH is ‘low-tech’, noninvasive and within the skillset of every health professional, therefore it is an easy approach to personalizing disease risk assessment [17]. However, others argue that its use requires further evidence [18], even if it does not appear, on face value, to cause harm [19]. Enquiring about FH is time consuming and has resource implications if systematically applied in primary

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and general healthcare settings [20]. Although CRC is a relatively frequent cancer, the increase in risk with FH is only poorly predictive of disease development in the individual patient [19]. What are the arguments, therefore, for building FH into CRC risk assessment and clinical management?

For the most compelling rationale, we need to take a population perspective. Worldwide, CRC accounts for approximately 700,000 new cases, and approximately 600,000 deaths, per year [101]. In countries with well-developed healthcare systems, overall 5-year survival is approximately 50%. Prognosis is strongly associated with stage at diagnosis: in the UK, 5-year survival exceeds 90% for those diagnosed at Dukes' stage A, falling to approximately 75, 50 and 6% for stages B, C and D, respectively [102]. CRC therefore distinguishes itself from many other common and serious cancers by being amenable to effective intervention if diagnosed in the early malignant or precursor stages. However, this is generally not achieved, despite the emphasis on patient education to recognize and act on symptoms, and the promotion of population-based screening programmes. The societal costs associated with healthcare interventions and lost productivity are significant. In the UK, no more than half of CRC patients are diagnosed at Duke's stage A or B (<15% at stage A) [20]. With approximately 16,000 deaths per year, even a small 'shift to the left' in stage at diagnosis could represent meaningful individual and societal benefits.

There is no contradiction in suggesting that primary prevention approaches to reduce CRC incidence should be complemented by surveillance efforts that are targeted as accurately as possible on those most likely to benefit. Current screening programs are effective in reducing CRC mortality [21,22], but colonoscopy is an invasive intervention with defined harms, and consumes expert resources, which are in limited supply. Even small reductions in the inevitable false-positive and -negative rates associated with the first stage screening (age and fecal occult blood tests) would lead to better matching of colonoscopy to those at highest risk, and avoidance of unnecessary procedures in the lowest risk groups. Thus, incorporating FH information into surveillance protocols is likely to improve selection of individuals for screening colonoscopy. This is to the benefit of the entire population eligible for screening, and to society as a whole, which ultimately provides the

resources that make surveillance programmes possible, although it may often be impossible to point towards the specific benefits for each individual.

Capturing & using FH information in CRC care

Generally speaking, FH information seems to be under-utilized by clinicians involved in CRC care across all health settings. Multiple studies have documented deficiencies in the recording of cancer FH information in patients' records [23–28], even in situations that should raise a red flag, such as younger patients being treated for CRC [29]. It also appears that recording positive FH information in a patient's chart does not guarantee that it will be acted upon [30]. This situation is compounded by the tendency for patients to under-report CRC in their relatives. Reports of bowel cancer in a relative can generally be believed, but in some studies only a third of relatives actually affected by CRC were reported [19]. However, it seems that under-reporting is more likely for second-degree or more distant relatives (suggesting unawareness or confusion over diagnosis), so the most important preliminary risk assessment FH – parents and siblings – may be reasonably expected to be fairly accurate.

Promoting the more extensive use of FH information in CRC care also needs recognition that changing practice faces many barriers [20]. The simplest suggestion is to raise health professionals' awareness of the value of FH, and to provide them with tools to assist them [17]. There is good evidence to suggest that using a FH tool (of any kind) seems to promote completeness and accuracy of information [31], and informatics infrastructures to support the use of electronic medical records may facilitate this process [32]. However, this does not guarantee its use as intended in clinical decision-making [33]. There is ample evidence that the most widely agreed-upon, effective health interventions are underused [34], and that passive approaches to promoting evidence-based practice do not automatically lead to their adoption [35].

Recognizing this, there are avenues of enquiry and action that may promote the appropriate integration of FH into CRC care. Firstly, while the evidence base on the association of FH and CRC is clear, how this might translate into benefits, harms and costs for any given population is not. The overall consequences of widely adopting FH in risk assessment are influenced

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by the underlying prevalence of CRC, and the effectiveness of existing policies on prevention, screening and treatment. Veenstra *et al.* developed a risk–benefit framework that emphasizes drawing together evidence on shifts in risk classification, clinical interventions, patient behavior, life expectancy and quality of life when a new ‘test’ (like FH) is added to existing protocols [36]. Such an analysis may be very revealing and help clarify the level of emphasis that FH warrants in comparison with other interventions to reduce CRC morbidity and mortality.

If analysis of risks, costs and benefits justifies efforts to use FH, then this should be reflected in the relevant professional guidance. Presently, CRC guidelines have variable recommendations on the use of FH [37]. While the challenges of integrating FH collection into routine practice [20] will not be overcome by simply disseminating a guideline, theory-informed approaches to changing practice are emerging [38].

Finally, we should recognize the role of patients and the public in promoting improvements in

their own clinical care. Since 2004, the US Surgeon General has promoted an annual ‘National Family History Day’. Families gathering for the Thanksgiving celebration are encouraged to talk about their health problems, supported by an online tool (My Family Health Portrait) [103]. Reaching out to the public as agents of change within a healthcare system aligns with the movement towards personalized medicine and patient-driven healthcare, and may be an entirely appropriate way to think about patients, families and shared responsibility for health [39].

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