

REVIEW ARTICLE

Point-of-care genetic counselling Should family physicians counsel patients on genetic testing and screening?

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ABSTRACT

Family medicine has come of age, with family doctors/general practitioners taking on greater roles and responsibilities and health care systems recognizing the important role of primary care. It is in this scenario that the question of pre- and post- testing counselling of genetic tests which are or would be offered directly to the general public through advertising and over-the-counter testing is being raised. This type of counselling would require enough personnel to deal with a large number of people; people who may not have genetic disorders in their families but who are curious about testing such as that for Breast Cancer (BRCA). It is argued that family doctors, albeit needing continuing professional development in this area, already have a solid foundation in genetics and are strategically placed in the community and numerous enough to impart such counselling. This would also liberate the responsibility from specialised geneticists who need to deal with families and individuals who have more serious genetic disorders to be managed.

Key Words: General practitioners, family doctors / physicians, strategically placed, community, genetic counselling, family medicine.

INTRODUCTION

For more than a decade the debate on who should impart pre- and post- test counselling for genetic tests has been discussed. To many, it seems that family doctors (also known as family physicians or general practitioners) are the key to this solution as they are numerous enough, are strategically placed in the community, and easily available to the general public who may have questions regarding what they hear and see about such testing.

It is important in this regard to consider that specialized geneticists cannot be disturbed from the secondary care job they do and should not be dragged into a primary care scenario. They are usually clinic / hospital based and, although family oriented, are not close to the same families in the midst of communities as family doctors are. Moreover, although updates and training in the type of counselling (as opposed to normal psychological counselling) is necessary, doctors have a clear understanding from the nature of their jobs of the underlying science of genetics. Their Colleges and Associations carry a responsibility to impart this continuing professional development for the good of society. It is argued as well that family medicine has 'come of age' and that it is no longer the Cinderella of medicine – family doctors are considered specialists and registered as such after having had formal training post-medical school.

What is special about genetic tests – the real concerns

Genetic information has a tremendous potential to harm as well as to help and stands to affect a broad number of family members (McCanse, 2001). Even welleducated patients may be ill-prepared to understand or deal realistically with the results of genetic tests. The primary care culture is different from the genetics culture but primary care doctors are more community-oriented, asking what specific aspects of a genetic approach to this health problem (or potential problem) are likely to benefit this patient. Howard Brody warns family doctors about the perils of genetic testing for patients and the role the family physician must play (McCanse, 2001, p.1). The ability to genetically screen for diseases far outpaces the ability to treat conditions, such as breast cancer, Alzheimer's disease and prostate cancer. Nonetheless people often consider genetic tests as some sort of cure or prevention of the condition (Lapp, 2002).

Companies may use advertising to entice people into believing that they should have genetic tests carried out (Chandros Hull & Prasad, 2001). They sometimes advise potential patients that there is no need to consult the family doctor or anybody else as their own 'experts' will guide the patients into what tests they should carry out. However genetic tests may not only affect individuals adversely, but also their family members. In this context it is fair for family physicians and their societies and colleges to be wary of the effect these tests can have over patients and their family members. Conversely family doctors, without the proper Continuing Medical Education (CME) imparted specifically to meet the needs of ongoing ethical dilemmas in genetic tests, may find themselves ordering such tests too liberally, once it is the patient who requests them, believing they are respecting the individual's autonomy. Family physicians have been 'urged to warn' patients of the potential pitfalls and dangers of using over-the-counter testing as prices start to fall. Whilst tests may sound enticing to patients, the impact they can have on their personal lives may not be divulged fairly and squarely by someone trying to market the test (Tanday, 2012).

Whilst the definition of genetic counselling continues to evolve, Ciarleglio et al. (2003) argue that the identification of susceptibility genes for common adult genetic diseases is moving the field of counselling into newer more challenging times. Genetic counsellors are also faced with having to translate more and more information which emerge from genetic tests into a way which can aid clients to make decisions, and which will reduce stress and anxiety, to enhance the ability to make life choices (Bennett et al. 2003). Weber and Corban (1996) note that although today geneticists perform most testing and counselling for genetic disorders, in the near future family physicians will increasingly become responsible for this role. Whilst the reasons for testing may be simple, they are likely to ignite fierce issues regarding cost, ethics, insurability, patient expectations and information which family members may wish not to know. How should family doctors consider the role in regard to genetic testing and counselling? In the light of this New Genetics, it may be envisaged that people will first inundate primary care physicians for answers to their questions. GPs accept they have an increasing role to play but may still show some lack of confidence in this area. Emery et al. (1999) say that the experience with counselling on cystic fibrosis in the UK is strong evidence to support the importance of providing genetic services in the primary care setting. Moreover the Association of American Family Physicians states in an editorial of its journal that several studies found that patients would prefer their family physician to facilitate an informed decision-making process on genetic testing and to counsel them about preventive measures. Although family physicians may feel yet quite unprepared due to what the editors call the 'big bang' in the knowledge of genetics, they strongly believe in the 'larger role in genetic counselling' that family physicians should take (Martin & Wilikofsky, 2012)

What are the concerns of genetic tests?

Why should genetic tests cause concern to family doctors more than any other form of test? The prime reason is indeed the novelty of these tests and the aura they are raising. Awareness campaigns sponsored by companies need to be considered for what they may actually be - an impetus for them to promote their product. While such a campaign need not to be bad in itself, if it is to be endorsed by the medical profession, the latter has the responsibility towards society not to be an accomplice in enticing patients to spend more than they should on such tests. Definitely not everyone needs do genetic tests and therefore fears must be quelled. Who is in a better position to quell such fears than family physicians who enjoy the trust of patients and their families? Some may argue that once these tests are available it is not the onus of any physician to try to convince someone not to do them. If one considers a commercially-available breast cancer (BRCA) test without any proper counselling however, there will be those who may not be aware of implications such tests carry to their employment, insurance and family when balanced against what management is available should they test positive - such as a radical mastectomy. This has enticed many states in the USA to have laws protecting against inappropriate access of such tests to the public. But in other countries such laws do not yet exist.

Studies on bilateral prophylactic oophorectomy vs. radical mastectomy (Kauff & et al, 2002; Rebbeck & et al., 2002) show that this is a highly evolving field in which it is wise to seek the advice of a doctor. Haber, analysing the relevance in the statics of such results, showed only that more studies are necessary. Thus by no means is there any certainty about outcomes of BRCA testing other than to recommend it to women past childbearing age and counselling them about an oophorectomy should they test positive (Haber, 2002). Again the operation does not exempt them completely from breast cancer. Notwithstanding the effectiveness of bilateral prophylactic radical mastectomy as has been demonstrated (Meijers et al., 2001), the controversy over such radical treatment remains. Point-of-care Genetic Counselling; the role of the Family Physician

Whereas it is undisputed that the General Practitioner is in an ideal position to counsel patients on genetic testing (BMA, 1998. p. 120) and to know where to refer patients for specialized counselling, Brody argues that a balance has to be stuck between the physicians' hunches, the patient's wishes and the evidence of clinical trials (Lapp, 2002). As mentioned, one concern which is not being addressed adequately, for example, is the implications such tests pose for family members. A possible solution he proposes is that the family doctor is in a position to set up a 'family covenant' before an individual goes through with testing. Such a document would be negotiated among the family members with the help of the physician. Family members who 'opt in' for set conditions are privy to the knowledge that comes out (Lapp, 2002). Yet the concept of covenant is lagging behind advances in genetic testing and it is doubtful how much such a covenant is possible before family doctors establish themselves as the agents of basic counselling.

The British Medical Association (BMA) document argues that primary care physicians should be able to identify patients and families who would need further genetic counselling by specialists, arguing that the rapidity with which genetic technology is developing and the complexity of the decisions to be made in relation to genetic testing mean that specialized pre- and posttest genetic counselling are likely to be required (BMA, 1998. p121). This however only refers to identification of individuals and families who need specialist counselling. It is unlikely that genetic counsellors can reach the public as much as the family physicians because of their smaller numbers and their less easy accessibility for the more general genetic tests being advertised. Moreover the family doctor already knows much about the family and probably its requirements and would be able to identify who would benefit from genetic information. The family doctor is familiar with the background and family dynamics in a way that a specialized counsellor can never be: it is information obtained over time within the context of practicing family medicine. Indeed if it were possible for the counsellor to arrive to such knowledge, it could be argued that this would be a repetition and waste of time for health professionals and patients alike.

Boxes 1 and 2 (BMA, 1998. p. 123-124) show respectively the process of genetic counselling and the framework of exploring decisions laid down by both the BMA and the American Society for Human Genetics. Nothing in this list is in fact beyond the capabilities of the average primary care physician. If people seek the advice of the family physician, it is appropriate that the latter should be able to handle most questions and counselling, leaving to the specialist those who have serious genetic inheritance problems. For those patients seeking to know more about cancer genes, paternity testing and even genetic screening of the unborn, the family physician is in an ideal and maybe better position to impart advice. Family physicians are moreover prescriptive by nature and thus tend to be more directive than the average non-directive genetic counsellor (Ibid. p122).

There are then additional reasons why general genetic counselling should be imparted by family doctors. The strategically placed primary point-of-care position of the family physician favours the role that genetic counselling should play in primary care physician. If people seek the advice of thefamily physician, it is appropriate that the latter should beable to handle most questions and counselling, leaving tothe specialist those who have serious genetic inheritanceproblems. For those patients seeking to know moreabout cancer genes, paternity testing and even geneticscreening of the unborn, the family physician is in anideal and maybe better position to impart advice. Familyphysicians are moreover prescriptive by nature and thustend to be more directive than the average non-directivegenetic counsellor (BMA, 1998.p122).

Of course the family doctor can never replace the role of the specialized genetic counsellor just as he can never replace the specialized radiographer and cardiologist. But the energy of the specialist counsellor is better spent on the hard core cases like Huntington's and Tay Sachs, rather than where the industry is striking hard, namely the cancer genes and such tests as 'cardiovascular panels' and 'thrombosis panels' which are aimed to raise awareness of the public but which may also satisfy a profit motive trumping over a benevolent principle. Specialized counsellors can then continue doing what they have been doing up till now – provide specialised services.

Conversely, if one considers countries where newlyformed companies offer genetic testing to the public, where family physicians provide no such counselling, such fertile ground is the ideal incubator for releasing 'awareness information' onto the public catching doctors off guard. Before there is enough time to prepare for genetic counselling services, people will start believing that there is some inherent cure in carrying out such tests (Lapp, 2002). On the other hand, doctors unaware of the implications of such tests will not counsel the public properly, as has been the subtle warning of the BMA. Specialized services, even if they do exist in theory in the main general hospitals, will not be enough to handle the everyday questions about genetic tests and definitely cannot direct patients into what tests are necessary. An appointment with the service may run into months just to handle the cases that truly need specialized counselling.

This highlights the importance of recognising that general practitioners are strategically placed to train themselves in imparting this counselling, which being a core medical subject is already in their realm. It is the responsibility of colleges, association and academics of family physicians to counsel members to learn more about genetic counselling. There will be no grass-root availability to answer questions about genetic tests of which one has heard about over the media.

The coming of age of Family Practice

A second important reason is the coming of age of family practice. Whilst the history of medicine shows that the family doctor or community doctor was the traditional doctor (Porter, 1996) (p.118)), the last century saw a surge of specialists and sub-specialists. In Britain the Royal College of General Practitioners was founded after the war and incorporated within it almost all general practitioners. It became the strongest political body in Britain to bargain with government over the structure of the National Health Service (Porter, 1996). Conversely, in the United States, the American Academy of Family Physicians brought together Family Doctors raising the status of Family Medicine to that of a speciality. Similar roads were taken in other countries. Family Medicine is now recognised and listed as a Speciality in its own right in the European Union and other continents are adopting vocational training in the field.

Family doctors now provide more and more services which can be offered to people at more reasonable rates making it more acceptable to insurance companies. GPs have always traditionally carried out minor surgery such as removal of sebaceous cysts, cautery of warts and injection of internal haemorrhoids. Nowadays more and more GPs take on more engaging non-invasive surgery such as removal of lipomas, injection of varicose veins, circumcisions and even haemorroidectomies (Brown, 1992) Studies (Siepel et al., 2000) have shown that family doctors who attend a course in ultrasonography can perform ultrasounds as part of an annual physical examination, detecting pathology such as renal tumours, aortic aneurysms and others, before any signs and symptoms are present. Family doctors in the United States train in sigmoidoscopy, gastroscopy, colposcopy and can even have a whole radiological set-up if economically viable. All of this is in the interest of quick diagnostics bypassing long referral lists and delays in a secondary care setting. The UK has been at the forefront experimenting with 'pathways' aimed at reducing costs and waiting times for the NHS and patients respectively, with the GP playing the key role in these reductions. In this setting it is reasonable to assume that family doctors with continued medical education (CME) are taking onto themselves more and more diagnostic techniques which not only increase the scope of general practice but which result in more benefit to patients. With proper CME a genetic counselling service to people and their families is clearly within the scope and definition of family practice.

What is needed with the impact of genetic technologies therefore is a primary care setting that can explain tests to all people, not only to those who have some genetic disorder in their lineage. Someone with a family history of colon cancer may inquire about the relevance and validity of genetic tests; it is reasonable to assume also that any woman may request information about whether she should have a BRCA test done. She may not know she needs counselling (in terms of implications for herself and her relatives and for insurance and employment interests). Therefore besides providing strategic community point-of-care contact, family physicians can bring a broader scope to genetic counselling. They are trained to think of issues such as getting patients to get their houses insured before getting tests done (Lavallee, 1999).

Consequently it is unreasonable to assume or request genetic counsellors to have to deal with this sort of mass population counselling. They would lose time which is valuable to what they are doing at present counselling families, which may indeed be identified by family doctors, in need of further in-depth evaluation. Unless genetic counsellors increase in numbers and become almost as common as the family doctor they may not be able to handle the amount of information which necessarily would need to be imparted to keep up with the media and the rapidly expanding genetic industry. Starfield et al. (2002, p.51) argue that if genetic problems including initiating diagnosis and even management, should be considered, primary-care centred systems offer the greatest resource for improving health.

Training – certification /

re-validation and vocational training

Studies do show that one cannot take for granted that since someone is a doctor, no formal professional development in this regard is necessary. In the first instance the counselling to be imparted is not the type of counselling we usually associate with psychologists, or, for that matter the counselling GPs can usually give to patients with psychological or family problems (Patient UK, 2012). It is conversely an integral part of the genetic testing process to involve both pre-test and post-test counselling. Whilst a genetic test may be available, a family physician would typically ask why the patient is asking about such a test at that point in time; what does the patient seek and what do they intend to do with the results? Making an analysis of whether they are ready can guide family physicians in taking appropriate care about consequences at first-contact point-of-care.

Guidance will certainly include explaining the impact any result will have on relatives and the fact that laws may oblige one to disclose information to other family physicians who are responsible for their relatives (Fulda & Lykens, 2006) and any decisions and legislation taken in this regard at a national level. Where no such legislation and guidelines exist, family physicians can act as patient advocates cautioning against over-the-counter genetic testing, for example.

In a study in New Zealand, Morgan et al (2012) found that General Practitioners have an increasingly important role to play in genetics but that the best way to implement future educational strategies need to be well considered. In their study, most GPs felt that they lacked experience and knowledge of genetic testing and had received very little formal training, even though they recognized the important role they have in this area. As highlighted earlier, Geller et al. (2012) confirmed that family physicians may be more directive in their counselling from conclusions of a study which included obstetricians, pediatricians, internists, family practitioners, and psychiatrists; this involved counselling patients on prenatal diagnosis and abortion. Certainly the change in attitude they advocate for primary care physicians would also have to include viewing genetic counselling from a much broader perspective than merely limiting it to reproductive issues.

The main areas of genetic clinical testing are antenatal screening and cancer genetics testing. More is promised in the future. However, the British Journal of General Practice has recently said that in providing genetic counselling, a family history may still be the most important tool so far, and that it is often neglected as part of a diagnosis (Walter & Emery, 2012). The editorial says that data from people who have taken over-the-counter genetic testing have not really had an impact on their change in life-styles. Perhaps this is a further argument why the pre- and post-genetic counselling should in fact be done by the family doctor, who stands in clinical equipoise (as opposed to someone trying to sell the test) with regard to the person considering the reason they want testing. Perhaps curiosity without a motivation to

BOX 1 (BMA, 1998)

"The British Medical Association states that genetic counseling consists of a series of activities which make a coherent whole. For ease of analysis we separate them in the list given below. In reality, however they are not separate entities, but facets of one process. In general terms, genetic counseling includes:

- Taking a family history and establishing a diagnosis;
- Gaining an understanding of the social and cultural context within which a patient and his or her family live and the values they bring to the counseling process;
- Listening to the questions and anxieties of the patient;
- Providing information about the condition, its inheritance pattern, and its management and raising questions about the potential significance of sharing information with other family members;
- Giving information about reproductive options; and/or
- Giving information about predictive options (if applicable);
- Providing the opportunity to reflect upon the options (implications counseling);
- Providing emotional support; and
- Initiating sustained help, if necessary, to enable individuals to adjust to particular life circumstances (psycho-therapeutic counseling)."

BOX 2 (BMA, 1998)

The description of genetic counseling set out by the American Society of Human Genetics is as follows:

Genetic counseling is a communication process which deals with the human problems associated with the occurrence or risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or family:

- (1) comprehend the medical facts, including the diagnosis, the probable course of the disorder and the available management;
- (2) appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives;
- (3) understand the options for dealing with the risk of recurrence;
- (4) choose the course of action which seems appropriate to them in view of their risk and their family goals and act in accordance with the decision;
- (5) make the best possible adjustment to the disorder in an affected member and/or to the risk of recurrence of that disorder.

change life style may make the patient reconsider testing unless there are more important reasons to do so, such as new forms of treatment. O'Brian says that there is no evidence that information obtained from genetic tests 'will be as valuable as the marketing suggests' (O'Brian, 2012). Moreover, family physician Nancy Stevens stresses the importance of injecting the family practice perspective into genetic medicine (McCanse, 2001). As this perspective is still underrepresented in conversations of genetic medicine, it means that patients of family practitioners are underrepresented. For example, she points out that only someone from high-risk families tends to benefit from BRCA testing.

The role and responsibility of associations and colleges

Certainly the responsibility taken on by family physicians is greater and respective colleges and associations may need to undertake the training of their members both in what we mean by counselling; what counselling should be done by family physicians, and of course when they should refer. Once it is accepted that the family doctor has this role to play in imparting knowledge and genetic counselling to patients, associations and colleges have an obligatory role to see that its members get the CME required in genetic counselling. Family doctors, by their very nature, are already in a position to give evidence-based information, genetics being one speciality they have always had in their curriculum. It would be unreasonable not to accept their role in providing such evidence-based counselling.

Associations and colleges of family doctors, which strive to guarantee excellence of their members to the public, have a special role to play here. But primary-care centred systems may pose a risk of underdetection and undermanagement of genetic problems if information and other educational networks do not actively support practitioners (Starfield et al. 2002, p. 51). Whereas it may be obvious that a family doctor intending to carry out diagnostic ultrasonography would require training, it may not be that obvious that to do genetic counselling one also needs training, because genetics has always formed part of the medical undergraduate curriculum. The focus of counselling is not on Mendelian inheritance explained in layman terms, but is a matter of explaining the social, legal and ethical implications of these tests and also of having a clear understanding of why they are so different than merely having a blood count done. Doctors need to understand and explain that genetic tests are largely non-therapeutic and predictive. The patient therefore needs to be empowered with information by someone who realizes the full potential of these tests and how industry may exploit fear of disease without concern for other family members and implications on employment and insurability.

Associations must guarantee that their members will explain the harm/benefit of genetic testing and screening. They must also guarantee that they will continue to seek the interests of the family and not only of individual people seeking testing. In other words, family doctors need to maintain the trust of the public, that financial gain is not the main motive of the counselling as may be the case for the company providing that test.

CONCLUSION

Whilst more recently a qualitative study published in the British Journal of General Practice has raised concerns about British GPs welcoming an enhanced role in clinical genetics and that the effectiveness on education policy aimed solely on knowledge is questionable (Mathers et al., 2010), it should be acknowledged that generally patients will go to their family doctors for enquiry because they are strategically placed and available. In any case, in many instances they would need a referral by their doctor for genetic services. The family doctor will already have considerable 'genetic' knowledge through the patient's family history (Mathers et al., 2010) and should be in a position not only to act as gatekeeper, given that genetic counsellors are limited, but to recognize his/her role in prevention and intervention – to avoid direct-toconsumer advertisement and over-the-counter analysis, and to counsel patients through the information they would need to know both before and after a test and indeed empower patients to make an informed choice.

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