Should Family Doctors Counsel Patients on Genetic Testing and Screening?

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ABSTRACT

Family Doctors are in an ideal situation to counsel patients on most medical technologies and new developments. In this sense they are in the best position to guide and counsel patients on genetic testing and screening. Indeed most often it is the patient who seeks counsel from the Family Doctor (General Practitioner). The special nature of genetic tests and the potential to exploit people's money with dubious testing puts the doctor in a special situation. Whilst it is argued that the Family Doctor maintains a strategic position to impart information to the patient, it is also argued that the new nature of genetic tests and the way the family may be affected, (including the multitude of ethical dilemmas these tests may pose), favours the position that Family Doctors should be the health professionals who should impart generic genetic counselling. Specialised genetic counsellors may then continue to dedicate their time to special cases. Tests should not be made available over-the-counter. It is the onus of the Family Doctor to refer patients for further counseling should this be necessary. Colleges and Academies of Family Physicians are in the ideal place to outpace industry especially in second and third world countries.

Key Words

Genetic Testing, family doctors, exploitation, counseling, specialist genetic counselors, continued medical eduction (CME).

INTRODUCTION

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 well educated 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 patient-oriented asking what specific aspects of a genetic approach to this health problem (or potential problem) are likely to benefit this patient. Howard Brody warned family doctors about the perils of genetic testing and the role the family physician must play (Ibid., 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).

At the same time Chandros Hull and Prassas (2001) have shown how companies may use advertising to their advantage to entice people into believing that they should have genetic tests. 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 the individual adversely, but also family members of that person carrying out the tests. In this context it is fair for family physicians and their societies and colleges to be wary of the effect these tests can have on 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 request them, believing they are respecting the individual's autonomy.

Weber and Corban (1996) note that although today geneticists perform most testing and counseling 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 their role with regard to genetic testing and counseling?

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 cam-

paigning need not 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 everybody need 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?

Consider the testing for the breast cancer genes BRCA 1 & 2. Those who test positive for the mutations of these genes via a commercially available genetic test are at an increased risk of having breast cancer compared to the general population. Some may argue therefore that once these tests are available it is not the onus of any physician to try to convince someone not to do them. Yet people may not be aware of the implications the result of such a test may have on employment and insurability. Furthermore, it is uncertain whether they know what, if anything, can be done with such knowledge and how this will affect their mental well being and that of their family. In the event such a test is positive, it does not necessarily imply that the person will have cancer; yet it puts them into a high risk category justifying insurers to charge a higher premium or not to insure them at all for breast cancer. This has enticed many States in the USA to introduce laws protecting against inappropriate access of such tests to the public. In other countries such laws do not yet exist.

Recent studies on bilateral prophylactic oophorectomy vs. radical mastectomy (Kauff, 2002; Rebbeck, 2002) show that this is a highly evolving field in which it is wise to seek the advice of a doctor. Haber (2002), analysing the relevance in the statistics of such results, shows 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 counseling them on oophorectomy should they test positive. Again this operation does not protect them completely from breast cancer, especially when there are as yet no studies to show whether the required Hormone Replacement Therapy (to prevent premature side effects of the artificially-induced menopause; namely increased cardiovascular risk and osteoporosis) may itself contribute to an increased incidence of breast cancer which the oophorectomy is being performed to eliminate. Even though the effectiveness of bilateral prophylactic radical mastectomy was demonstrated recently (Meijers, 2001), the controversy over such radical treatment remains.

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; Starfield et al., 2002) and to know when to refer patients for specialized counselling, Brody argues that a balance has to be struck between the physicians' hunches, the patient's wishes and the evidence of clinical trials (Lapp, 2002). 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' 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 genetic counselling.

The 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 genetic counselling, both pre-test and post-test, is likely to be required (BMA, 1998, p. 121). 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 easily as family physicians because of their smaller numbers and their inferior accessibility, especially considering the increasing number of generic genetic tests being advertised. Moreover the family doctor already knows much about the family and its requirements and would be able to identify who would benefit from genetic information. He/she 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 at 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 (Ibid., p. 123-124) show the process of genetic counselling and the framework for 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 or family doctor. When patients seek the advice of the family physician, it is appropriate that the latter should be able to handle most generic questions and counselling, referring on to the specialist only 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 (BMA, 1998, p. 122).

There are additional reasons why generic genetic counselling should be imparted by family doctors. The strategically placed position of the family physician favours the role that genetic counselling should play in primary care. It is the responsibility of family physicians as a group to take on the role of protecting families against commercial interest. This is particularly important because people may not be aware of the implications to other members of the family when doing a genetic test. Who else but the family physician is in the central stage to counsel directly family members? This is all more important because to await the development of genetic services and to wait for specialized counsellors to deal with the true impact of genetic testing is unrealistic even in the United Kingdom and the United States, let alone the rest of the world.

Of course the family doctor can never replace the role of the specialized genetic counsellor just as he can never replace the specialized radiographer or cardiologist. But the energy of the specialist counsellor is better spent on hard core cases like Huntington's chorea and Tay Sachs disease, rather than where the industry is striking hard, namely the cancer genes and such tests as 'cardiovascular panels' and 'thrombosis panels'. The latter are targeted to raise public awareness in order to satisfy a profit motive rather than a genuine social need.

Consider a country like Malta where recently a newly formed company started to offer genetic testing to the public. Presently it uses the services of specialists and family doctors for referrals. No form of counselling is offered to the patient, leaving this onus on the doctor. Indeed there has been little to promote awareness among the medical profession of the special nature of genetic tests and the implications they may have on the life of the person seeking those tests, and on his/her family. This 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, as happens in other countries, that there is some inherent cure in carrying out such tests (Lapp, 2002). Doctors, on the other hand, unaware of the implications of such tests, as has been the BMA's subtle warning will not counsel the public properly. Specialized services, even if they do exist in the main general hospital, are not enough to handle the everyday questions regarding genetic tests and definitely cannot inform patients adequately about what tests are really necessary. Appointments with the service may well run into months, and then just to handle the cases that truly need specialized counselling.

Family doctors are strategically placed to train themselves in imparting this counselling, this being a core medical subject already in their realm. It is the responsibility of colleges, associations and academies of family physicians to stimulate members to learn more about genetic counselling.

The coming of age of Family Practice

Another important perspective 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 specialties and sub-specialities. In Britain the Royal College of General Practitioners was founded after the war and incorporated almost all general practitioners. It became the strongest political body in Britain to bargain with government over the structure of the National Health Service. 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 later in other countries.

Family doctors now provide more and more services, which services that 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, circumcision and even haemorroidectomies (Brown, 1992). Studies have shown (Siepel, 2000) that family doctors who attend a course in ultrasonography can perform ultrasounds as part of the physical examination, detecting pathology such as renal tumours, aortic aneurysms and others, before any signs and symptoms are noticed by doctor and patient respectively. Family doctors in the United States train to perform sigmoidoscopy, gastroscopy, colposcopy and can even have a whole radiological set-up if economically viable. All of this in the interest of quick diagnostics bypassing long referral lists and delays in a secondary care setting. In this setting it is reasonable to assume that the family doctor, with continued medical education (CME) is taking onto himself 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 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. It is reasonable to assume that any woman may request information about whether she should have a BRCA test done. She may not know that she needs counselling (in terms of implications for herself and her relatives, for insurance purposes, etc). Family physicians can bring a broader scope to genetic counselling. They are trained to think of issues such as getting patients to get their house in order vis-à-vis insurance before getting tests done (Lavallee, 1999).

Conversely it is unreasonable to assume or request genetic counsellors to have to deal with this sort of mass population counselling. They would lose time that is valuable for what they are doing at present – counselling to those families, which may indeed be identified by family doctors, who require further in-depth evaluation. Unless genetic counsellors increase in numbers and become almost as common as family doctors they may not be able to handle the demand for 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 when one considers genetic problems, in initiation of diagnosis and even management, primary-care-centred systems offer the greatest potential for improving health.

The responsibility of Associations and Colleges

Family physician Nancy Stevens stresses the importance of injecting the family practice perspective into genetic medicine (McCanse, 2000). As this perspective is still underrepresented in conversations of genetic medicine, it follows that patients of family practitioners are underrepresented. For example, she points out that only one from a high-risk family tends to benefit from BRCA testing. 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 in genetic counselling that is required. Family doctors, by their very nature, are already in a position to give evidence-based information, genetics being one specialty 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, have a special role to play here. But primary-care-centred systems may pose a risk of under-detection and under-management 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 provide 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 from simply having a blood count. 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 understands the full potential of these tests and how industry may exploit fear of disease without concern for implications on employment and insurability and impact on other family members.

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, demonstrating that financial gain is not the main motive of for the counselling as may be the case for the company providing that test.

References

BRITISH MEDICAL ASSOCIATION (1998), Human Genetics, Choice and Responsibility, Oxford University Press, London.

BRODY., H. (1997): "Edmund D. Pellegrino's Philosphy of Family Practice", in: Ed. D. Thomasma, The Influence of Edmund D. Pellegrino's Philosophy of Medicine, Kluwer Academic Publishers, Dordrecht, Boston, London, 7-20.

BROWN, J.S. (1992), Minor Surgery. A text and Atlas, Chapman & Hall Medical, London, New York, Tokyo.

CHANDROS HULL, S. AND PRASAS, K. (2001): 'Reading between the lines: Direct-to-consumer advertising of genetic testing', Hastings Center Report 31(3): 33-35.

F P REPORT, (1999): Special Secion, Human Genetics, July, p.1

Box 1

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 that decision;

5. make the best possible adjustment to the disorder in an affected member and/or to the risk of recurrence of that disorder.

Box 2

The British Medical Association states that genetic counselling 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 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).

FRIEDSON, E., (1988): Profession of Medicine. A Study of the Sociology of Applied Knowledge. University of Chicago Press, Chicago and London.

HABER, D., (2002): "Prophylactic oophorectomy to reduce the risk of ovarian and breast cancer in carriers of BRCA mutations", New England Journal of Medicine, 346(21): 1660-1661.

HOLM, S. (1999): "There is nothing special about genetic information", in Thompson, A.K. and Chadwick, R. (eds.): Genetic information. Kluwer Academic Publishers/ Plenum Publishing, New York: 97-104.

KAUFF, N., et al (2002): "Risk-reducing salpingo-oophorectomy in women with a BRCA1 or BRCA2 mutation", New England Journal of Medicine, 346(21): 1609-1615.

LAPP, T., (2002): "Rise to challenges posed by genetic screening", FP Report, 8(1): 5.

LAVALLEE, M., (1999): "Who better than their family physician to hold their hand and walk them down that scary road?", FP Report, Special Section: 1-2.

McCANSE, C., (2000): "What role will FPs play in the genetics revolution", FP Report, 6(6): 17-20.

McCANSE, C., (2001): "First do no harm...Genetic counseling: To test ot not to Test?", FP Report, 7(9): 1-2

MEIJERS, H., et al (2001): "Breast cancer after prophylactic bilateral mastectomy in women with a BRCA1 or BRCA2 mutation", New England Journal of Medicine, 345: 159-164.

REBBECK, R., et al (2002): "Prophylactic oophorectomy in carriers of BRCA1 or BRCA2 mutations", New England Journal of Medicine, 346(21): 1616-1622.

SIEPEL, T., et al (2000): "The ultrasound-assisted physical examination in the periodic health evaluation of the elderly", in The Journal of Family Practice, 49(7): 614-621.

STARFIELD, B., HOLTZMAN, N.A., ROLAND, M.O., SIBBALD, B., HARRIS, R., HARRIS, H., (2002): "Primary care and genetic services. Health care in evolution", in European Jouranl of Public Health, 12(1): 51-56.

WEBER, R., and CORBAN, G., (1996): "Genetic Testing: Are Family Physicians ready?", FP Report, 53(6): 1-3.