SHOULD FAMILY PHYSICIANS COUNCIL PATIENTS ON GENETIC TESTING AND SCREENING?

LES MÉDECINS DE FAMILLE DOIVENT-ILS CONSEILLER LEURS PATIENTS SUR LES TESTS GÉNÉTIQUES ET LE DÉPISTAGE ?

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SUMMARY

Genetic tests not only affect the individual adversely, but also family members. In this context it is fair for family physicians to be weary of the effect these tests can have over family members. Conversely family doctors, without the proper Continuing Medical Education, may find themselves ordering such tests too liberally, once it is the patient who request them, believing they are respecting the individual's autonomy. 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.

KEYWORDS

Family physicians, genetic testing and screening, counselling.

RÉSUMÉ

Les tests génétiques non seulement concernent les personnes sur lesquelles ils sont réalisés mais aussi leur famille. Dans ce contexte, il est logique que les médecins de famille aient conscience des conséquences de ces tests sur les membres de la famille. Mais, les médecins de famille, sans une formation continue appropriée, pourraient prescrire de façon trop libérale ces tests, demandés par le patient, en croyant ainsi respecter le droit à l'autonomie de celui-ci. En effet, si les tests peuvent sembler attrayants pour les patients, l'impact qu'ils peuvent avoir sur leur vie personnelle ne peut permettre qu'ils soient diffusés par des personnes dont le rôle est de les commercialiser.

MOTS-CLÉS

Médecins de famille, patients, tests et dépistage génétique, conseil.

Genetic information has a tremendous potential to harm as well as to help and stands to affect a broad number of family members. 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 than 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.

At the same time Chandros Hull and Prassad have shown how companies may use advertising to entice

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people into believing that they should do 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 not only may 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 weary of the effect these tests can have over 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. 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.

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 make decisions, in a way which will reduce stress and anxiety and enhance ability to make life choices (Bennett et al. 2003). Weber and Corban 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 the New Genetics the overall workload of the primary care physician is thought to change with patient inundating doctors for advice. GPs accept they have an increasing role to play but may still show some lack of confidence in this area. Emery et al however found that the experience with counselling on cystic fibrosis in the UK is strong evidence of the importance of providing genetic services in the primary care setting. Moreover the Assocation of American Family Physicians states in an editorial of their journal that several studies found that patient would preer their family physician to facilitate an informed decision-making process on genetic testing and to counsel them about preventive measure. 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

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 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 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 via a commercially available genetic test are at an increased risk compared to the general population of having breast cancer. 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 for employment and insurance the result of such a test may have. Also 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 is uncertain. 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 have 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 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, show 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 an oophorectomy should they test positive. Again the operation does not exempt completely them from breast cancer, especially when there are as yet no studies to show whether the required Hormone Replacement Therapy (to prevent premature side effects to the artificially-induced menopause; namely increased cardiovascular risk and osteoporosis) may itself have an increased incidence of breast cancer

which the oophorectomy is trying to eliminate. Even though the effectiveness of bilateral prophylactic radical mastectomy as demonstrated recently, 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 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. 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 (Ibid). 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 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 pretest, is likely to be required (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 easily accessibility as the family doctor for the more 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. 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 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 (Ibid., 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 of family doctor. 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 advise. Family physicians are moreover prescriptive by nature and thus tend to be more directive than the average non-directive genetic counsellor (Ibid. p122).

BOX 1

"The British Medical Association states that genetic counseling consists of a series of activities which make a coherent whole. For ease of analysis w 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 (pschotherapeutic counseling)."

BOX 2

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:
comprehend the medical facts, including the diagnosis, the probable course of the disorder and the available management;
appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives;
understand the options for dealing with the risk of recurrence;
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;
make the best possible adjustment to the disorder in an affected member and/or to the risk of recurrence of that disorder.

There are then additional reasons why general 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 being unrealistic even in 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 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 in order to satisfy a profit motive rather than a benevolent principle. Specialized counsellors can continue doing what they have been doing up till now.
Consider countries where newly formed companies offer genetic testing to the public. In the beginning it uses specialists and family doctors for referral. No form of counselling is offered to the patient, leaving this onus on the doctor. This fertile ground is the ideal incubator for releasing 'awareness information' onto the public catching doctor 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 Doctors, on the other hand, unaware of the implications of such tests, as has been the subtle warning of the BMA, will not counsel the public properly. Specialized services, even if they do exist in theory in the main general hospital are not enough to handle the everyday questions of genetic tests and definitely cannot direct patients into what tests are necessary. Appointment with the service may run into months just to handle the cases that truly need specialized counselling.
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.

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 (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 in 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. 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 as a Speciality in its own right with many EU states and other continents 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 insure 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 lists and even haemorroidectomies Studies have shown 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 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. The UK has been at the forefront experimenting with 'pathways' aimed at reducing costs and waiting times for the NHS and patients respectively; the GP playing the key role in these reductions. In this setting it is reasonable to assume that whilst 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 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 she needs counselling (in terms of implications for herself and her relative and also for insurance etc). The family doctor is therefore not only strategically placed for this role but genetic counselling is within the scope and goals of family practice. Moreover 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 vis-a'-vis insurance before getting tests done.

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 which is valuable to 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 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. op. cit., p.51) argue that if genetic problems should be considered, and initiate diagnosis and even management. Primary-care-centred systems offer the greatest resource for improving health.

Is training necessary?

Certainly the responsibility taken on by family physicians is greater and colleges and associations may need to undertake the training of their members both in what we mean by counselling and what counselling should be done by family physicians, and of course when they should refer. Studies do show that one cannot take for granted that since they are doctors, 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. It is conversely an integral part of the genetic testing process involving both pre-test and post-test counselling. Whilst a genetic test may be available what does the patient seek when inquiring about the test 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.

Consequences 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. This decision is usually one taken at a national level before guidelines and laws are introduced, which family physicians will then be obliged to following from a public health perspective. Certainly ethical issues beyond the scope of the family doctor will have to be taken into account by authorities as one may decide to follow more utilitarian approaches. In this regards issues relating to genetic discrimination certainly will have to be discussed at a national level – such as releasing information to insurance or employers. Family physicians cannot take this responsibility upon their shoulders and work in a consistent way across a country without specific guidelines and what to counsel the patient. For this reason it is also important that the family physician act as a patient advocate in this regard and warn patients where over-the-counter genetic testing is offered and no legislative framework yet not in place. Due to the nature of this problem the question of 'genetic exceptionalism' – that a genetic test is different from testing for other disorder – takes on considerable weight.

Morgan et al admit that General Practitioners in New Zealand 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. Geller et al, on the other hand say that family physicians may be more directive in their counselling from conclusions of a study which included obstetricians, pediatricians, internists, family practitioners, and psychiatrists. However 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 different perspective than merely to do with reproductive issues. It does seem that some of the literature views genetic counselling by primary care physicians as having to do mostly with counselling families whenever reproduction is being considered. This is certainly a limited view.

The main areas of genetic clinical testing are antenatal screening and cancer genetics testing. More is promised in the future. The British Journal of General Practice has recently said that in the latter regard a family history may still be the most important tool so far, complaining that it is often neglected as part of a diagnosis. 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 is in a neutral position (as opposed to someone trying to sell the test) to make the person consider the reason they want testing. Perhaps curiosity without a motivation to 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'.

THE RESPONSIBILITY OF ASSOCIATIONS AND COLLEGES

Family physician Nancy Stevens stresses the importance of injecting the family practice perspective into genetic medicine. 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. 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 that is required. 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. Whilst more recently a qualitative study published in the British Jounral of General Practice has raised concerns about British GP's welcoming an enhanced role in clinical genetics and that the effectiveness on education policy aimed solely on knowledge is questionable (Mathers, J. 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 (Ibid.) and should be in a position not only to act as gatekeeper, given that genetic counsellors are limited, but to recognize her role in prevention and intervention – to avoid direct-to-consumer 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 empowering patients to make and informed choice on whether they really need to do the tests to alleviate whatever concern brought them to inquire about a genetic test. ■