National Consultative Ethics Committee for Health and Life Sciences

Opinion n° 76

Regarding the obligation to disclose genetic information of concern to the family in the event of medical necessity

Reply to the Minister for Health, the Family, and the Disabled, on whether there should be a legal obligation for persons who have been diagnosed for a serious genetic disorder, or for a predisposition thereto, to advise other members of their families who could benefit effectively from treatment and/or preventive measures

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On March 26, 2003, Mr. Jean François MATTEI, Minister for Health, the Family, and the Disabled, referred to CCNE on whether there should be a legal obligation for persons who have been diagnosed for a serious genetic disorder, or for a predisposition thereto, to advise other members of their families who could benefit effectively from treatment and/or preventive measures. This legal obligation would also apply to physicians who had not supplied the information of concern to the family. Both physicians and individuals concerned could incur penal sanction in case of failure to comply or negligence,

This issue is part of the ongoing debate on whether the interests of a third party or of society should take precedence over the interests of the individual concerned. In its Opinion n° 70, CCNE has already pointed out the extreme importance of respect for the proband[1] whose interests must *a priori* be protected.
However, this Opinion did not specifically broach the subject of the interests of third parties in a genetic context. Since in this case, strict observance of the principle of individual autonomy could endanger the lives of several relatives, there is a need to revisit the concept of medical confidentiality in the light of these particular situations.

I - What is at stake?

1- The specificities of genetic testing.
- As a first requirement, the specific characteristics of genetic testing must be understood. This is not ordinary testing for the purpose of diagnosis – although it sometimes is the case (diagnosis of periodic disease, Steinert's disease, hereditary hemochromatosis, etc…).
More often than not, the purpose is to predict, with varying degrees of probability, the appearance of a disease or disorder, depending on the lesser or greater connection between genotype and phenotype, penetrance, early or late onset of the disease, homozygous or heterozygous status, dominant or recessive quality, gender. Genetic mutations may have variable phenotypic expression (cf. for instance hemochromatosis or cystic fibrosis), or even not be expressed at all, as in the case of female fragile X syndrome carriers.

- However indeterminate, the results of a genetic test are not the sole concern of the proband. They also affect the whole family, ascendants, descendants, collaterals, and possibly spouses.
- The test may concern a genetic disease, severe at birth, the expression of which is due to the presence of a recessive gene not expressed in the phenotypes of either parent.

- Secondly, it should be noted that despite these specific traits, a test is not the only way of diagnosing a genetic disease: haemoglobin electrophoresis (for sickle cell disease and thalassemia), renal ultrasound (polycystic kidney disease), coloscopy (polyposis of the colon), or cholesterol assays, can all be as significant to assess a family risk as genetic testing. All of these diagnostic criteria would well **generate** the same legal consequences

[1] We prefer to use « proband » and avoid « index case » because of the stigma which could apply to the latter concept by connotation. The French expression « *mis à l'index* » means « ostracise » or « denounce ».

as a genetic test regarding the disclosure of information, if they could lead to prevention or therapy for other members of the family.

- Thirdly, the predictive or pre-symptomatic and familial nature of these tests, together with implications for the family, gives special significance to the information provided by the doctor: for the individual concerned who must face up to the future with more or less anxiety and disquiet depending on the severity of the possible consequences of the mutation, and for blood relatives since they are involved *ipso facto*. Because of these specific features, legislators have always stipulated that consent must be secured before genetic testing, so that those concerned are aware of the consequences and are able to exercise, if they wish, the right not to know.
Learning that one is a carrier for a genetic mutation or a chromosomal anomaly, is a traumatising experience and even induces feelings of guilt or humiliation: "having bad genes", "having a genetic defect in the family and passing it on to the children", etc… The sometimes dramatic nature of genetic information is sufficient justification for doctors to consider that only the person undergoing a test should be at liberty to decide, according to what conscience dictates, if members of the family[2] are to be informed. The physician should not disobey his patient's wishes in this respect, and speak to the family himself. In any case, he has no knowledge of the family tree of the person concerned except through that person, and therefore cannot inform the family through any other conduit. Respect for privacy is also in this case convergent with medical concern never to undermine a patient's trust in his physician.

2 – Possible preventive care
More often than not, there is no possibility of prevention, as is the case for myopathies, polycystic kidney disease, or Huntington's chorea, but sometimes genetic foresight is an opportunity to initiate effective preventive measures, as is the case for phenylcetonuria, congenital myxedema, hereditary hemochromatosis, familial polyposis of the colon, familial glaucoma, and quite a number of familial cancers, or juvenile forms of ornithine transcarbamylase deficiency, etc…

- Prevention can be achieved through dietary measures (late-onset ornithine transcarbamylase deficiency, phenylcetonuria), or therapy (preventive surgical or endoscopic excision of organs, fitting a pace maker), exposure to medication (glucose 6-phosphate deshydrogenase, acute intermittent porphyria, genetic halothane sensitivity etc.) or surgery.
  or diagnostic measures (prenatal genetic testing, etc.). It is because preventive action can be more or less effective that it may be crucial to inform the family. Awareness by families and doctors of the possibility of hereditary disease may speed up alertness for certain specific symptoms, may help to arrive at a difficult diagnosis, enable early preventive action, encourage targeted screening for certain cancers (breast or colon for example), or even specialised prenatal screening which may lead to termination of pregnancy for medical reasons.

Ways of coping with these situations depend of course on the specifics.

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[2] Cf. CCNE Opinion n°25, dated June 21, 1992 : Family members should be contacted personally by the relative requesting the test, if necessary with the assistance of a doctor as it might be technically difficult for him to explain his request.

- Extremely critical familial disorders in the pre-natal phase are, in principle, recessive, with each heterozygotous parent transmitting to the child a modified allele of the same gene. In such cases, the possibility of remedial action in the immediate postnatal period is fairly rare: liver transplant in the habitually infantile form of ornithine transcarbamylase deficiency, exchange transfusion in various pathological situations…
  However, these situations are so severe that there is clearly justification, at the parents' request, for pre-natal diagnosis in high-risk families, which means that information must be circulated. In this case, however, both father and mother are involved, hence the need to inform their respective families.

- It is also with a view to pre-natal diagnosis that there can be good reason for communicating information to the family concerning later onset genetic diseases, of the kind for which there is neither preventive nor therapeutic remedy, be they transmitted by dominant or recessive genes. Huntington's chorea is the archetype of this sort of disease. CCNE dealt specifically, in its Opinion n° 72 dated July 4, 2002, with some of the problems arising out of a pre-natal diagnosis of this description. It is worth noting that, in such situations, simply collecting information concerning the family history is often sufficient to identify the genetic risk.

There can be, therefore, a strong conflict between the strict observance of confidentiality, which may be desired by the person who has been diagnosed for a mutation, and the possible interests, sometimes of major proportions, of other people who would need to have this information for their own benefit.

Stakes may be high, and numerous.

1. Are there limits to medical confidentiality? Is it possible to overrule confidentiality because others would benefit? Does medical confidentiality exist solely in the interest of the person concerned, or for the benefit of all?
2. If a family were to prosecute for lack of information, could medical liability become both medical and legal?
3. Is voluntary retention of information by a person who is a carrier for an identified genetic disorder, whose family could have benefited from preventive and/or early treatment for that disorder, equivalent to failure to assist a person in danger, or to endangering the life of others?
4. In other words, could genetic information be included in the list of "mandatory notifiable information", on the grounds that the family would benefit?

II. Secrecy

Medical secrecy, the strict confidentiality that must be observed as regards the relationship between a doctor and his patient, is the foundation of the healthcare relationship. Any disease can put a patient at risk of social, and sometimes familial ostracism, and may generate feelings of fundamental guilt, of self-reproach, even for quite common illnesses. Genetic disorders follow the same rule.

• If there is no medical gain attached to the information, any announcement of impending future anguish, sometimes of an indeterminate or indecisive nature for oneself and one's children, will be a difficult burden to bear when there is no prospect of effective therapeutic remedy. Is there any point in forecasting if prevention is out of the question? This dramatic situation is particularly evident in cases such as Huntington's chorea. The binary nature of the dilemma (carrier/non carrier) contains the frightening threat of a lethal neurological disease later in life. The tragic nature of this situation justifies psychological assistance at the time of taking the test and when results are communicated. In effect, advance notice is given of a condemnation as a result of a genetic curse. In such cases, the disease is generally known to the family, and the issue is whether relations wish, or do not wish, to know. There are cases, however, where diagnosis is tardy, during the psychiatric phase of Huntington's chorea. The question then arises of whether to inform the family, because although the disease has no known treatment, the physician may suffer pangs of conscience. Should one announce what is known? How then can the right not to know be observed? It is singularly difficult for a doctor to warn (or even recommend warning) relations that they may be carriers of the gene and transmit the disease. CCNE is its Opinion n°72 wrote on the difficult issues connected to the right not to know whether the gene is present in a given individual, whilst protecting that person's wish to be the parent of an unscathed child.

Thus, even in the case of a genetic mutation causing a disease for which there is neither prevention nor cure, keeping secrets is a problem. Furthermore, the matter does not only have an impact on blood relations, since it may also affect a spouse. Should this information be divulged to a partner who also has rights regarding his/her offspring? Many high-risk families do request pre-natal diagnosis, but this of course, is only possible if the genetic risk is known.
It would seem that maintaining secrecy should solve the doctor's moral dilemma in some dramatically incurable diseases. Indeed, insofar as any patient has a right to the observance of medical confidentiality whatever the disease, insofar also as the disease offers no hope of a remedy to relations, and that they might well be justified in blaming their doctor for having pointlessly turned their lives into relentless anguish, it would seem unacceptable to impose a duty of communication to the family in such cases. Nonetheless, it does not seem possible either to deny a request from the family for diagnosis of a late-onset genetic disease, even if the information cannot lead to preventive action.

The ethical conflict is even more acute in situations where information could lead to effective action:

- When potential medical benefit can be derived from the information

Should the principle of medical confidentiality be systematically waived if preventive or therapeutic measures could reduce or correct the effects of a genetic anomaly? This is not the first time the issue of disclosure or breach of medical confidentiality arises. Information regarding the risk incurred by carriers of a severe but preventable genetic disease, is generally communicated by the persons concerned, if they are well advised and aware of the risks, and wish to assist their families or avoid the birth of a child suffering from a particularly severe and incurable disorder.
It can happen that people are negligent, that a man (or his wife) have doubts regarding the identity of the biological father, that there is some major family conflict, or a psychiatric situation, or a very ambiguous feeling of "not wanting to go under alone".

Such situations are however sufficiently few and far between, so that there would not be an overwhelming need to associate genetic testing and disclosure. Were that to be the case, the climate of trust generated by medical confidentiality could well be destroyed, and those who were uncertain about their genetic status might be deterred from consulting a specialist.

On the contrary, safeguarding a privileged trustful relationship should give the doctor the best chance of convincing his patient that collaterals should be informed of the genetic risk they may be incurring. It is far from clear that any legal obligation of "mandatory notification" of at risk genes, would necessarily achieve its aim, i.e. improving the prevention of genetic risk in collateral relations.

III – The concept of mandatory medical notification

Withholding information and thus endangering the life of others has been discussed time and again in the context of the risk of HIV contamination of a spouse. Should society demand that a doctor breach the rules of medical confidentiality in favour of the sexual partner of the infected person, because to do otherwise would be endangering the life of a third party or failing to give assistance to a person in danger? Even in such a situation, CCNE has always been in favour of keeping the principle of medical confidentiality absolutely intact; to break it is to destroy the trust of a patient. In this case, the utilitarian and high moral grounds converge to declare that principles should remain inviolate. Patients inform their doctors about their sexual affairs and their partners. It is only because patients are convinced that their secrets will be safe with their doctors that they are willing to prolong the dialogue and that the opportunity will arise for firm and reiterated persuasion regarding the need to inform partners. A doctor must do everything in his power to convince a patient that a partner or partners should be informed, but he must not step in personally and breach medical confidentiality. No one would disagree that this represents a major conflict of values, but it would seem illusory to believe that authoritarian measures are likely to solve that conflict (fears that medical screening and legal coercion are part of the same process).

Obviously, the analogy between danger of contamination by an infectious agent and transmitting a genetic "defect" cannot be taken too far. Only the partners of the contacts are concerned in the first example, whereas the risk extends to all or part of a family line, or even to an unborn child, in the second. Eradication of an infectious agent is conceivable, whereas "eradicating" a detrimental gene is not as yet a technical possibility. However, as we have just observed, the two situations are not dissimilar as regards an analysis of what course to take. In both cases, a strict application of the principle of medical confidentiality, as well as utilitarian attitudes, argue against systematic breaching of confidentiality. An attitude of trust and persuasion would seem more likely to lead to partners in the first case, and collateral relations in the second, being warned of the dangers to which they are exposed and the best chances of avoiding them.

IV. Rights and duties of the proband, moral and legal obligations.

Strict observance of medical confidentiality can therefore be justified simply because it is likely to be the most efficient way of gathering, and later – only later – transmitting information. However, for a person to withhold information that would have been required

to avoid or treat in a timely way a genetic disease which threatens collateral relatives, appears in principle to be morally condemnable. In the same way, the attitude of a physician who did not do his utmost, in the course of person-to-person dialogue with his patient, to persuade him to inform his family, would be reprehensible, particularly when such information was needed to avoid dramatic outcomes. However, we have alluded to some of the mindsets that could make a person reluctant to pass on such information, or even oppose its disclosure. If one excepts cases where the proband is not in fact biologically related to the rest of the family, how can the chances of necessary transmission of genetic information be increased?

In all cases where that information turns out to be crucial for preserving the life or the health of collaterals, the proband should be informed with precision of the situation and of the risks involved in leaving the family in ignorance. Those undergoing genetic tests should always, even before these are performed, be asked to give free, specific, and informed consent. Furthermore, the information given on that occasion, and the signed form would state specifically that those concerned are aware of the consequences of the anomaly which could be detected, for themselves, for their next-of-kin, and their relatives, and would emphasise how important it is to circulate that information in the event that grave injury, possibly mortal injury, to collateral relations also affected by the genetic anomaly if it was not detected early enough, could be avoided.
The doctor should then give his patient a document outlining the risks in connection with the genetic anomaly concerned, and the possible steps that could be taken to minimise them. It would then be up to the person in question, or the parents in the event of serious genetic disorders during childhood, to hand this document over to collaterals who might be concerned. The latter would then contact their own family doctor. This document would have to be very specific about the nature of the genetic disorder. Furthermore, the document would present the proband as the essential link in the family chain, through which the family would have the chance of protecting itself, if only partially, and therefore would underline the moral responsibility of the proband for the circulation of information.

Whatever solution is adopted to deal with this problem, it does not appear that one could take matters any further than the above, even though it is not possible at this time to evaluate what follow-up might be given to complaints from those who have been deprived of information, for instance the parents of a child who might have been saved, or whose birth could have been avoided, if information had been available. Lawsuits can always claim medical liability, and special legislation is not necessarily essential to penalize some kind of negligence. Professional associations should undertake the task of drafting more precise guides than are now available of good practices in this field, entailing the moral responsibility of the party involved as regards other parties.

CONCLUSION

1/ Observing medical confidentiality is an essential principle for constructing a trustful relationship between doctors and patients, and, as has been clearly analysed in the case of HIV contamination, any transgression of this principle might well lead to less screening. It would be counter productive if the very notion of genetic screening were to create *a*

priori reluctance for those who might feel threatened by compulsory disclosure of their biological privacy.

2/ The quality of their relationship is the starting point for an in-depth sincere dialogue between a specialist and his patient. On that occasion, the latter must be fully informed of his personal responsibilities and obligations to next-of-kin. The procedure to secure free, specific, and informed consent, as a necessary prelude to genetic testing should be conducted in such a way that the person concerned fully understands the meaning for himself and possibly for his family, of the results that will be made known to him. If it turns out that the person concerned could have transmitted or be a carrier for a genetic defect which demands remedial action as regards affected collaterals, it would then be the physician's task, as a continuation of information already supplied, to secure the consent of the person concerned to pass on or to allow such information to be passed on to potentially concerned relations. In any event, the doctor should give the proband (the person tested or the parents of a child who has been diagnosed as suffering from a severe genetic disorder) a letter for the attention of the relations concerned. Such a document would explain, in plain language, the nature of the genetic observations, the disorders they could be exposed to, and the steps to be taken with the assistance of their attending physician, to whom the document would then be communicated.
A good quality dialogue between the physician and persons who have been diagnosed as the carriers of a genetic anomaly, should make it easier for him to make sure on further occasions that the information has indeed been passed on, or if not, to try and convince them that it should. Apart from very exceptional situations when the transmission of the information is of particular urgency (high risk pregnancy, collateral relation under immediate and otherwise avoidable threat of complications because of the undiagnosed genetic disease) the proband must be allowed sufficient time to overcome possible initial disinclination. It should be possible to set aside the time needed to take in and reflect upon the gravity of the consequences for himself, and for his family. The complex psychological effects when such information is revealed should be reason for obtaining the help of specialised professionals, geneticians, psychologists, etc. In fact, there is a more complex gap than is immediately perceptible between information and the communication of that same information. In the circumstances, cases of obstinate refusal to transmit potentially helpful information to collateral relations should be truly exceptional and would certainly not justify the adoption of new legislation, thus adding exceptions to the rule of medical confidentiality.

3/ Genetic diagnosis does not of course exclude clinical action which is as important as it ever was. For example, a patient with polyposis of the colon will be warned of the need to inform his family so that they can all undergo screening coloscopy and treatment if required. This highly significant clinical information should not raise any protest. However, the doctor will never take the initiative of warning family members unless the proband requests him to do so.

4/ To the strict observance of the principle of medical confidentiality that CCNE recommends as regards the transmission of genetic information, there can be no objection based on the well known exception to that same confidentiality i.e. mandatory notification

of infectious diseases[3], or the steps taken to avoid or slow down the extension of epidemics. In the first case, the urgency of the situation is unmistakable justification for regulations designed for the protection of the public and its well-being, which supersede individual rights. As regards the circulation of genetic information, it would only be very exceptionally of an urgent nature, so that procedures for information and persuasion as recommended above would surely be better adapted to allow for maximum protection of those under threat while maintaining respect for individual rights.

At the close of this analysis, in the context of the factual situations in which the issue of the need to communicate genetic information to a person's family, arises out of the result of a test, CCNE considers that the most effective way of arriving at the desired result, i.e. protecting the family whilst strictly preserving personal privacy, is to implement adequate procedures within the bounds of strict observance of medical confidentiality.
The best interests of the community must not be defended in courts of law through penalties inflicted on the person concerned or on the doctor. The rare situations in which information is not communicated for some reason should not be seen by law as representing "failure to assist a person in danger" or "endangering others".
There is always a temptation to legislate on the basis of situations which are highly charged in emotion, or using the more recent acquisitions of science as justification, but there is a risk that fashioning law in this way leads to inequity by giving too much weight to a few exceptional, singular, or sensitive situations, to the detriment of respect for personal rights.

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[3] Mandatory notification does not always breach confidentiality. For instance, notification of AIDS or of HIV contamination is anonymised and does not lead to identification of the person concerned.