

Occasional Survey

Disclosing Genetic Information to Relatives or Carriers?

S FROEHLICH, HM SASS, Y WANG

Abstract

Genetic information and counseling of relatives of carriers can be in conflict with traditional medical, legal and moral rules of confidentiality. A case study was mailed to 200 physicians, nurses, and students in medicine, philosophy and business in Asia (Hong Kong, Beijing) and Europe (Vienna, Frankfurt, Bochum). We asked about moral institutions in professional disclosure and moral obligations of carriers towards family members. A majority, even stronger in Asia, supported the 'right to know' versus a 'right not to know', also called for changes in health policy to make genetic testing and counselling affordable and more widely available.

Key words

Confidentiality; Family ethics; Genetic information; Genotyping; Health care policy

Confidentiality is one of the major bioethical principles in medicine and supported by age-old traditions in Eastern and Western physician's ethics. In modern days, data protection has become one of the most important features in the respect of privacy and the protection against exploitation and discrimination. Confidentiality is particularly important in regard to medical records. Patients will not trust their physicians and their medical institutions, if they feel that confidentiality has or might be broken. On the other hand, patients and their families might not trust their doctors if they do not everything possible to prevent, to ease or to heal diseases, pain, and disorders.

On the other hand, there are quite a few situations where the protection of the public warrants to balance individual rights and professional or public obligations, e.g. the right

of the individual to confidentiality with the duty of the physician or the medical or social community to warn or to protect others from harm and with the right of fellow citizens to be protected from unwarranted harm. Ethical, medical, and political conflicts between confidentiality on the one side and the disclosure of information on the other in the case of HIV carriers are well documented and may serve as a paradigm for these complex ethical and medical situations.¹

While HIV infection may be a threat to everyone, hereditary genetic disorders affect member of the wider family. Severe genetic diseases such as Cystic Fibrosis, Chorea Huntington, and Polycystic Kidney Disease [ADPKD] are a burden and a risk to carriers and will one way or the other influence their life, lifestyle, and quality of life. For some of these disorders it does not make a difference today, whether or not carriers know about their status as no remedies or modifications in lifestyle are yet known to postpone, to reduce or otherwise to counterbalance the effects of these hereditary diseases. But other disorders, such as ADPKD, the most frequent among the severe genetic diseases, can be influenced in regard to onset by good hypertension control and the avoidance of stress to the lower abdomen; therefore it would be beneficial, if carriers would know early enough about their status and get appropriate advice. Access to genetic screening and the right to disclosure of genetic information

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in these and very soon in an increasing number of their genetic disorders definitely may prevent harm and provide help for presymptomatic carriers.

The Case and the Questionnaire

We have asked participants of the Second Hong Kong Medical Genetics Conference in Hong Kong and of the Second Sino-German Interdisciplinary Symposium on Medical Ethics in Shanghai, both held in October 1999, to respond to the following case:

Mrs. Han, a hypertension patient, suffered from adverse reaction when her medication was changed from calcium antagonist to a beta blocker as the correct doses could not be established. "Now she has pain in her lower abdomen and renal hematuria, symptoms associated with ADPKD, an autosomal dominant genetic endstage renal disease. In retrospect, her father might have died of the same disorder 30 years ago without a diagnosis but with similar symptoms. Mrs. Han fortunately was accepted as a hemodialysis patient; she faces death, if dialysis will not be continued three times a week or if she does not get kidney transplant. Both substitution therapies have side effects, are costly and not readily available except in economically rich countries. Most likely members of Mrs. Han's family are presymptomatic carriers of ADPKD, the onset of which can be postponed by hypertension therapy. Genetic screening can be done prenatally and postnatally."

We got 43 responses (21 from Hong Kong, 22 from Shanghai, roughly a 50% return rate). Two months later we gave the same case and questionnaire to physicians, nurses, and students in Germany and Austria with 71 responses (25 postgraduate students of the University of Vienna, Austria, 25 business students of Frankfurt, 12 physicians and nurses of a Dortmund hospital and 10 postgraduate students of Bochum University). While the number of participants [n=43 from Asian conferences, n=73 from European groups] was quite small, there are nevertheless preliminary hints how the conflict between confidentiality and the disclosure was perceived and might be handled.

Responses and Results

Firstly, we discussed the physician's ethical obligations. We asked "Must the physician ask Mrs. Han to inform all members of their family about her disorder and their

prospective carrier status?" The 'strong yes' and 'yes' (on a scale of 1 to 5) from the Asian conferences were 51% and 18% [median 2.00 (Hong Kong 2.33; Shanghai 1.68), standard deviation 1.24], from the European groups 50% and 30% [median 1.86, dev. 1.12]. – Then we asked whether there was a moral right or duty of the physician to contact members of Mrs. Han's family. The answers 'strong yes' and 'yes' were 26% and 26% from Asia [median 2.62 (Hong Kong 3.35; Shanghai 1.95), dev. 1.40] and 30% and 27% from Europe [median 2.60, dev. 1.46]. – When we asked about a legal obligation of the physician to inform family members against Mrs. Han's will, responses were less favourable: we got a 'strong no' and 'no' from 36% and 19% of Asian responses [median 3.67 (Hong Kong 3.85; Shanghai 3.50), dev. 1.24], and 44% and 19% of Europeans [median 3.75, dev. 1.41].

Secondly, we discussed ethics and obligations of carriers. We asked whether Mrs. Han had a right to disclosure and to encourage her family members to seek diagnosis. 'Strong yes' and 'yes' from Asian participants were 44% and 26% [median 1.63 (Hong Kong 1.81; Shanghai 1.45), dev. 0.82], from Europeans 79% and 16% [median 1.28, dev. 0.61]. Then we asked whether there was an ethical obligation of Mrs. Han to inform: 'strong yes' and 'yes' of Asian responses were 42% and 33% [median 1.98 (Hong Kong 2.48, Shanghai 1.50), dev. 1.10], or European 44% and 31% [median 2.01, dev. 1.20]. – We also asked whether 'minor genetic disorders' would warrant disclosure. 'Strong no' and 'no' from the Asian conferences were 26% and 14% [median 3.23 (Hong Kong 3.43, Shanghai 3.05, dev. 1.34], while 'strong yes' and 'yes' were favoured by Europeans 23% and 22% [median 2.68, dev. 1.26].

Finally we addressed questions of health care policy. We wanted to know whether prospective carriers of severe disorders such as ADPKD should get access to free testing. Overwhelmingly 'strong yes' and 'yes' from Asian conferences was 63% and 16% [median 1.72 (Hong Kong 2.19, Shanghai 1.27), dev. 1.14], and from Europeans 55% and 21% [median 1.77, dev. 1.09]. – When asked whether there would be a 'moral duty' of potential carriers to seeking testing, the 'strong yes' and 'yes' answers from the Asian conferences were 30% and 33% [median 2.40 (Hong Kong 2.33, Shanghai 2.45), dev. 1.35], from the Europeans less favourable 23% and 20% [median 2.82, dev. 1.50; 'strong no' 25%, 'no' 21%]. – The question whether health care professionals should limit prenatal testing services to a number of severe disorders. Again, was answered differently by Asians with 'strong no' and 'no' 33% and 14% [median 3.42 (Hong Kong 3.10, Shanghai 3.73), dev. 1.38], and by

Europeans with 24% and 17% [median 2.97, dev. 1.54; 'strong yes' and 'yes' was 27% and 14%].

We used the opportunity (and the case of Mrs. Han) to evaluate attitudes towards mass screening and genotyping for drug metabolism of the P450 cytochrome complex² should physician genotype for drug metabolism so that patients get better medication and have less side effects? Responses were mostly favourable for routine genotyping. 'Strong yes' and 'yes' from the Asian conferences were 44% and 26% [median 1.98 (Hong Kong 2.48, Shanghai 1.50), dev. 1.08], from Europeans 29% and 26% [median 2.52, dev. 1.38].

Discussion

In light of these responses, it appears that lay people and health care professionals see a strong obligation in physicians ethics and genetic ethics to not only counsel carriers who seek testing and advice for themselves, but to strongly advise patients to inform and motivate potential carriers in their family to seek testing, medical help, and genetic counseling. The traditionally highly individualised and confidential interaction between physician and patient and its traditional set of physician's ethics does not seem to appropriately describe the moral rights and obligations of physicians and patients in human genetics. Confidentiality still is one of the most important bioethical principles but its exclusive priority needs to be balanced by considerations of preventing medical harm from and providing medical and educational service to potentially vulnerable fellow-humans, in the case of hereditary disorders to family members. The most preferred and ethically accepted way to inform family members is through those who know about their own status. Making it a legal duty of physicians to contact members of the family against that patient's will got a less favourable response.

As far as carrier ethics is concerned, all answers favoured the moral right – and to a lesser extent the moral obligation – of carriers for disclosure and for informing their relatives. In regard to disclosure of 'minor genetic disorders' we found no significant tendency to either disclose or not disclose; a third of all participants did not prefer either one of the opposites, probably wanting to decide case by case and disorder by disorder in specific scenarios.

As to health care policy guaranteeing and providing free genetic testing, Asian responses were even more in favour than Europeans; this is remarkable as genetic services are much more widely provided free of charge in Europe than

in Asia. Participants from Asia also voiced a somewhat stronger support of a moral duty of potential carriers to seek genetic testing, thus being less favourable towards a 'right, not to know'. Asians also were more in favour of not limiting prenatal genetic testing to a number of severe disorders, thus asking for best possible information in parental choice.

It was surprising, given widely reported public scare of genetic screening and potential discrimination as a result of disclosure of individual medical characteristics, that both, the participants of the two Asian conferences as well as the various European groups, responded favourably to genotyping for drug metabolism. Health care professional and lay people seem to recognise the benefits of individualised efficacious drug delivery, comparable to the efficacy and risk reduction of blood transfusion after blood typing.

Conclusion

The recent controversy discussing the dangers of new paternalistic eugenic movements favouring disclosure and genetic testing, highlighted by the discussion following Mao's³ view on recent screening programs and results⁴ and ongoing studies on the possibility of eugenic attitudes among genetic professionals⁵ are indicative of the still unresolved issue of balancing confidentiality with disclosure, and the duties to know with the rights not to know.

Proposed International Guidelines on Ethical Issues in Medical Genetics by WHO require that 'the genetic service provider should encourage the individual to ask the relatives to seek genetic counselling'. But in case the individual refuses, recommend that 'the genetic counselor may ethically make direct contact with relatives'.⁶ Sass² recommends to consider the patient, whether a carrier or not, to be the prime moral agent responsible for disclosing risks associated with carrier status to relatives as the golden rule, while the German association of human geneticists propose to handle cases individually.⁷ Given the probability of influencing the time of onset and severity of ADPKD, Kielstein and Sass value the 'duty to know' higher than a questionable 'right, not to know'⁸ and have developed a genetic-ethical action guide for carriers as well their counselors to deal with disorders such as ADPKD.⁹

As Wertz notes, there are strong temptations among professionals to violate confidentiality in favour of disclosure and of avoiding harm; on the other hand,

professionals in human genetics, as expressed by the proposed WHO guidelines, understand that they have responsibilities and obligations to those who have not asked for their service yet and who do not know that they are carriers of genetic disorders. A German gynecologist recently compared the ethical challenges in balancing confidentiality, disclosure, harm to patients, harm to others, interests of society, eugenics, and discrimination to the risks associated with 'hiking along the ridges of a mountain'.¹⁰

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References

1. Wang Y. A strategy of clinical tolerance for the prevention of HIV and AIDS in China. *J Med Philos* 2000;25:48-61.
2. Sass HM. Ethical Issues in Genotyping for Pharmacogenetics and Genetic Disorders. *HK J Paediatr (new series)* 2006;11:297-305.
3. Mao X. Chinese geneticists' views of ethical issues in genetic testing and screening: evidence for eugenics in China. *Am J Hum Genet* 1998;63:688-95.
4. Chen Z, Chen RB, Qiu RZ, Du RF, Lo WH. Chinese geneticists are far from eugenics movement. *Am J Hum Genet* 1999;65:1199-201.
5. Wertz D. Eugenics is alive and well: a survey of genetic professionals around the world. *Sci Context*, 1998;11:493-510.
6. World Health Organization. Proposed International Guidelines on Ethical Issues in Medical Genetics and Medical Services Geneva: WHO [doc.ref. who/hgn/gl/eth/98.1], 1998.
7. Gesellschaft fuer Humangenetik. Positionspapier. *Med Genetik* 1996;1:125-32.
8. Kielstein R, Sass HM. Right not to know or duty to know? Prenatal screening for polycystic renal disease. *J Med Philos* 1992;17:395-405.
9. Kielstein R, Sass HM. Ethical Issues in Kidney Disease Ethics and the Kidney, cd Levinski NO. Oxford: Oxford University Press, 2000, Chapter 10.
10. Schmid-Tannwald I. Prenatal diagnosis or prenatal selection. A grave step of grade for the physician. *Internist (Berl)* 2000; 41:M30-3.

Literature

McGleenan T. The jurisprudence of genetic privacy. *Med Health Care Philos* 1998;1:225-33.

Peoples Republic of China. Maternal and Infant Health Care Law, 1994.

Sass HM. Some Cultural and Ethical Reflections on Molecular Genetic Risk Assessment Proceedings of the International Bioethics Committee 1994, Vol. II, Paris: UNESCO, 1995.