Gason AA, Aitken MA, Metcalfe SA, Allen KJ, Delatycki MB
Genetic Susceptibility Screening in Schools: Attitudes of the
School Community towards Hereditary Haemochromatosis
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Genetic Susceptibility Screening in the School Setting?

While school-based genetic reproductive screening programmes, for instance the Tay-Sachs disease carrier screening programme, are offered and acceptable to students and the community in Australia, genetic susceptibility screening has never been offered in the school setting. Experience has shown that the success of a population-based programme relies on the acceptance of the community. Gason et al. sought to determine whether or not a school-based genetic susceptibility screening programme for haemochromatosis (HH) is acceptable to a selection of nineteen secondary schools in the state of Victoria. HH is a common, treatable and preventable late-onset condition (1 in 300 Australians are homozygous for C282Y). The penetrance of HH remains an issue for debate. The results of this study indicate that offering genetic susceptibility testing in schools could be a public health success with the support of the community; 68% of students following education, 61.4% of parents and 61.8% of staff indicated that the likelihood was either definite or probable that they would have a test for their genetic susceptibility to HH. In the discussion, Gason et al. try to explain this positive attitude of the school community. Other studies have shown that a personal experience or having an affected family member can cause a more positive attitude. Gason et al. show that the school community has some familiarity with the condition, which is a possible explanation for the acceptance of screening for HH. The results of Gason et al., as do other studies, subscribe the importance of education for attitudes and knowledge. They found a linear correlation between students' knowledge and attitudes. However, another study reported that high knowledge about genetic testing is not always related to approval. According to Gason et al., this difference may be a reflection of the nature of educational material presented. The educational material in the study of Gason et al. focused on genetic susceptibility screening for HH in particular, rather than all genetic screening. Furthermore, the emphasis is on the availability of a prevention of HH. Gason et al. also found that participants who thought that they knew a sufficient amount about HH would be more likely to decide to be tested. The

uncertainty of C282Y penetrance was described in terms of level of risk rather than numerically and may contribute to the belief of having inadequate knowledge. Gason et al. note that from a public health perspective, the issue of clinical penetrance of susceptibility genes needs to be resolved, whereas comprehensive programmes with effective educational strategies, counselling, support and thorough consent procedures will need to be developed. Effectiveness of the programmes will be realized, if they are developed with a collaborative effort of health and education professionals.

Comments

The study of Gason et al. showed that the majority of the Victorian secondary school community was positive about genetic screening for HH susceptibility. However, apparently the authors did not worry about the autonomy of decision-making. Because of the school setting, it is likely that there might be pressure on the target group to decide to undergo a genetic test.

Knowledge is a major contributor in influencing attitudes towards genetic testing. According to Gason et al., familiarity with HH, receiving educational material, thinking that you know a sufficient amount and knowing that a suitable prevention method is available all play a role in getting a positive attitude. The authors indicated in their discussion that also the content of the information influences the attitude of people towards genetic testing. In this study, the emphasis of the information was on the availability of a prevention of HH. Had the material emphasized the probably low penetrance of HH, then, in my opinion, it is plausible that the percentage of positive participants would have decreased. Thus, I want to underline that providing adequate educational material and giving full information to the target group is a crucial aspect in the field of genetic screening. The information must describe all advantages and disadvantages realistically and balanced. Only then will people be able to make a good and well-considered decision. In this era of genomics, the field of genetic screening asks for a transdisciplinary approach. I subscribe to the message of Gason et al. that the effectiveness of screening programmes will only be realized if they are developed with a collaborative effort of health and education professionals.

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Newson AJ, Humphries SE

Cascade Testing in Familial Hypercholesterolaemia:
How Should Family Members Be Contacted?

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How Should Family Members in a Cascade Screening Be Contacted?

Familial hypercholesterolaemia (FH) is an autosomal dominant condition. If left untreated, it can lead to elevated cholesterol concentrations and a more than 50% risk of coronary heart disease (CHD) by the age of 50 in men and at least 30% in women by the age of 60. Treatment with statins in combination with a healthy lifestyle can be effective in preventing CHD, but requires early diagnosis. Cascade testing based on lipid or genetic testing in the relatives of known FH probands is likely to be cost effective in terms of life-years gained.

Newson and Humphries examine the ethical implications of alternative methods by which relatives can be contacted and describe criteria that will allow for a balance between efficiency and respecting interest of probands and relatives. Standard practice in clinical genetics involves providing information and clinic contact details to the proband, who will then pass this on to relatives. This method protects the confidentiality of the proband. However, probands may sometimes refuse to mediate access to relatives, may not feel ready for discussing the risk or fail to provide their relatives with accurate information. Response rates using direct contact, by which probands provide the clinic with relatives' names and contact details in order that the clinic can contact them directly, are likely to be higher. Concerns about breaching the relatives' right 'not to know' may not hold for a serious but treatable condition such as FH. In order to protect probands' confidentiality, it is preferable to contact relatives after initial family contact has been established by the proband, if they indicate their wish to do so. Only if a proband fails to respond to the clinic, a breach of confidentiality should be considered.

Comments

Newson and Humphries argue that direct contact is an ethically justifiable method of contact tracing in cascade screening for FH, provided certain provisions are adhered to. Their paper indeed addresses most of the ethical issues of contact tracing. However, some aspects of cascade testing may warrant further considerations.

Although the authors acknowledge the potential for reactions of alarm and concern, they do not take into account that people may experience a sense of fatalism when confronted with the knowledge that a genetic condition is present within the family. In a screening program, effort should not only be put in preventing feelings of anxiety but also in preventing feelings of fatalism.

The authors also assert that using direct contact instead of family contact will not further raise the potential for insurance discrimination. For example, people with a family history of high cholesterol or CHD are already subjected to higher premium levels.

This may be true for relatives who are aware of their family history, but this sort of information is not always spread throughout families. Taking into account the family history, receiving a letter with warning of increased risk of FH can pose unexpected difficulties with insurances, despite the potential benefit of knowing one's family history. Independent of the method of contact tracing, limited access to insurances is a potential problem that should be addressed. In a study from the Netherlands, FH screenees reported unanticipated insurance problems, such as the requirement to pay a higher premium, to undergo additional medical tests, to let the insurance company scrutinize medical records and complete rejection of the application for insurance [1].

L. Claassen, Amsterdam

Reference

- 1 Marang-van de Mheen PJ, Van Maarle MC, Stouthard ME: Getting insurance after genetic screening on familial hypercholesterolaemia; the need to educate both insurers and the public to increase adherence to national guidelines in the Netherlands. *J Epidemiol Community Health* 2002;56:145–147.

Harris J, Sulston J

Genetic Equity

Nature Rev Genet 2004;5:796–800

Knoppers BM, Chadwick R

Human Genetics Research: Emerging Trends in Ethics

Nature Rev Genet 2005;6:75–79

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The New Bioethics: Turning towards the Community

Does biomedical ethics turn away from the individual towards the community, and is this important to scientists? Two very well-written articles on current trends in biomedical ethics were published in *Nature Review Genetics* in October 2004 and January 2005, respectively.

In the first article, Harris and Sulston propose a principle of 'genetic equity' alongside a new concept of human dignity. Both are grounded in the classical principle of equality, which is basic to the hypothetical 'social contract' that modern states derive their legitimacy from. According to the authors, 'genetic equity' should be made operational, as a further and more effective line of protection for the rights of the individual. This is needed, because with the rapidly increasing ability of science to analyse genetic information, the risk of misuse increases as well. Harris and Sulston state that they do not agree with the current tendency to reject 'genetic exceptionalism' (the view that genetic information differs signifi-

cantly from other information). They hold that 'there are good reasons to focus more general principles specifically on genetics'.

The second article, by Knoppers and Chadwick, looks back at 10 years of developments in genetic research and in the accompanying ethics. Ten years ago, they identified autonomy, privacy, justice, quality and equity as the leading ethical principles in genetic research [1]. Today, they note a shift in emphasis, away from the paramount position of the individual towards family, community and population-defined interests. The protection of this broad range of interests requires a normative framework featuring reciprocity, mutuality, solidarity, citizenry and universality. Reciprocity is connected with notions such as transparency, informed choice and respect for cultural values, and is a relevant principle in the context of studies among specific communities and populations. Mutuality implies risk sharing, and because of the familial nature of genetic information, it is relevant to the sharing of both risk and information within families. An ethical duty to warn family members at risk may be inferred from the principle of mutuality. Citizenry refers to issues of collective identity that are particularly important for minority groups. Universality refers to the human genome as the common heritage of humanity, to obligations to future generations, benefit sharing and genome databases as global public goods.

The authors point out the fact that the ethical principles they present are not a replacement of the various ethical frameworks that have been in use now for several decades. They focus on the current shift in emphasis that is tied to the development of genetic research: 'ethical thinking inevitably continues to evolve as the science does'.

Comments

The fact that *Nature Review Genetics* published two sophisticated papers on ethical theory within 4 months can only be interpreted as a clue to an important development in ethics with direct relevance to the scientific community of genetic research. The mes-

sage that both articles contain is clear: the shifting focus of genetic research from individual genetics and the problems of hereditary disease in families to community genetics and the related problems of epidemiology and public health requires new principles and a new ethical framework.

However, the argument of Harris and Sulston is not fully clear. They call for a more effective protection of the rights of the individual, but their proposed principle of genetic equity is grounded in the position of the individual as a member of the community. Their reference to the concept of equality as used by Rousseau, and in particular their reference to Plato's *Republic*, may fit a communitarian model rather than a liberal concept that strengthens the position of the individual.

Their stance on genetic exceptionalism is confusing: 'although we do not believe that genetics raises unprecedented or qualitatively different issues, we do think that there are good reasons to focus more general principles specifically on genetics'.

The ethical principles proposed by Knoppers and Chadwick document the shift in attention from the rights of the individual towards the interests of the community. They try to show that ethics follows developments in science. Their claim that 'there might not, and cannot, be universal norms in bioethics, as emerging ethical norms are as 'epigenetic' as the science they circumscribe' touches upon a deep controversy in ethical theory.

However, for scientists involved in genetic research, it may be very useful to be at least informed about the emergence of new norms and principles in bioethics. Reference to a normative framework is a standard requirement in most research proposals. Knowledge about new ELSI/ELSA trends will be important.

J. Lunshof, Amsterdam

Reference

- 1 Knoppers BM, Chadwick R: The Human Genome Project: Under an international ethical microscope. *Science* 1994;265:2035-2036.