



Ethical Challenges in Teaching Genetics for Medical Students

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Although inclusion of ethics as a study course in medical students' curricula is a common practice, special approaches in teaching ethics in the context of genetics should be considered. In the realm of genomics, there are several ethically sensitive topics such as diagnosis of genetic diseases, *in vitro* fertilization, and identification of genetic susceptibility to common diseases. In addition, in communication with the general public, genetic terms should be used with caution. Demonstration of the phenotypes of affected individuals should be regarded as a particular aspect of teaching genetics. In a description of a patient's phenotype, not only is it necessary to provide scientifically precise characteristics of a patient; voice timbre, facial expression, and body language should also be carefully controlled. Furthermore, in medicine, the theory–practice gap is a problematic aspect, and students often find it difficult to apply knowledge on ethical issues to real situations in clinics. For this purpose, clinical cases are presented during classes and their analysis requires a very respectful attitude on the part of both students and lecturers. For many genetic diseases, evaluation of minor anomalies such as a curved fifth finger, low situated ears, or missing of some teeth is required. Some minor anomalies are found in healthy individuals too, and interpretation of such features must therefore be considered carefully. This article describes our experiences in teaching genetics at Riga Stradiņš University, ethical problems faced while teaching genetics, and their solutions.

INTRODUCTION

Ethics education is often based on knowledge and application of traditional ethical theories such as utilitarianism and deontology (6). Genetics classes for medical students are mostly organized around discussions about genetic phenomena on the basis of individuals affected with the genetic disorder. A special approach in teaching ethics in the context of genetics should therefore be applied. Topics taught to medical students should be carefully evaluated in terms of both content and moral aspect. Ethically-sensitive topics require a particular attitude and moral maturity on the part of the lecturer. It is vital that educators make conscious ethical decisions to ensure that their professional practice meets the highest possible ethical standards of conduct and responsibility (4). Students might feel uncomfortable or embarrassed due to the terminology used in genetics, especially shy students, students from different cultural environments, or those whose native language is

not the language of instruction. A balance between the use of scientific terms and colloquial speech must be found to make the topic accessible to all students, and students should learn how to convey complex information to patients. Many human genetic studies are aimed at identifying genes that determine a predisposition to genetic disease. A diagnosis of the disease or a finding of inherited predisposition in a family member has implications for other family members (8). The lecturer should indicate that a medical doctor has to feel confident about the disclosure of such information to other family members, especially non-biological family members. It is also critical to note that not all ethical dilemmas that arise during genetics teaching will be solved. In many cases, unpredictable situations and questions might arise, and the lecturer must be confident about being able to prevent embarrassing situations. This article discusses some ethical issues arising while teaching genetics for medical students.

ETHICALLY-SENSITIVE TOPICS

There are concerns about ethical issues associated with several topics included in the genetics study programs for medical students at the Riga Stradiņš University. Examples of such topics will be discussed in the following sections of this article.

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Diagnosis of genetic diseases

From a diagnostic perspective, the following aspects should be pointed out: what kind of genetic information can be disclosed, to whom and how this information should be disclosed, who has the right to be informed, and how genetic information might be used, perceived, or missed (3). Genetic diseases could be diagnosed at the postnatal or prenatal period of an individual's development. Prenatal diagnosis of genetic diseases has been a focus of ethical debates for years. For medical students, this topic should be discussed in detail and several sensitive points should be considered. Prenatal diagnosis can result in the identification of genetic pathology, and these diagnoses present specific ethical dimensions. Some ethical aspects of prenatal diagnosis are associated with the interpretation of test results. It is not helpful for a counselee to be informed only about the diagnosed genetic disease, as the counselee's main concern might be how severely the child will be affected just after birth or as an adult, and what particular possibilities exist to treat the condition.

Excluding the option of pregnancy termination, accurate diagnosis of fetal health status allows the delivery of a child with special needs to be planned, with special neonatal therapy. The task of a genetic counselor is not only to convey information on risk in an understandable way, but also to do it in a manner that enables those counseled to make their own informed choices (13).

The notion of disorder, that is, what counts as a disorder, is an important issue that deserves to be discussed. For instance, in the case of late onset diseases (e.g., Huntington's disease) individuals who carry the mutant gene will develop the disease, but should the individual be considered affected before the onset of the disease (2)? Carrier status is also very sensitive in the case of autosomal recessive diseases, especially in case these diseases are widespread within a population. The possibility for two carriers to mate could be rather high, and carriers in turn are at high risk (25%) of producing an affected child. All these confusing problems should be discussed with students, who should be encouraged to give their own opinion. At the end of the topic, the lecturer should clearly identify the problem, clarify solutions to the problem, and demonstrate ways of developing future treatment strategies.

In vitro fertilization (IVF). There are millions of people worldwide whose conception was achieved by *in vitro* fertilization. *In vitro* fertilization offers new promises for couples who are infertile due to genetic problems. In this case, however, a solution requires careful evaluation of the benefits and possible psychological harm that infertile couples might face. Nevertheless, the fact that many infertile couples are willing to spend thousands of dollars and face the physical and mental challenges of IVF rather than adopt a child suggests a strong emotional need for biological offspring that is not influenced by social

pressures (7). Medical students encounter several ethical problems when studying different aspects of IVF. Firstly, the couple's attitude to the embryo and, consequently, to the expected offspring should be considered. If one or both parents are not able to produce sex cells, the embryo is not fully genetically related to the parents. Secondly, the fate and state of the embryos should be discussed. A sufficient number of embryos are needed to secure pregnancy. The remaining embryos are frozen and may be used for research purposes or destroyed. The question is whether the destroyed embryos are considered to be harmed in this process. The answer depends on another question, namely, whether an eight-cell-stage embryo should be regarded as a human being, and whether arresting its development should be considered a wrong. Students should be encouraged to give their opinion about the stage at which human life begins. If an embryo is considered as a group of totipotent cells, than destroying these cells could not be regarded as destroying a human being. For many individuals, the generally accepted view is that a human organism becomes a person at the moment of birth; the early embryo does not at this stage of its development have the moral status that is later acquired, as it develops capacities such as sentience, consciousness, and agency (1). However, the pre-embryo will eventually develop into an individual who is a human being. Attitudes around these ethical dilemmas should be concordant with the personality of future parents. The medical doctor/counselor must be able to customize and to explain these issues from the standpoint of those being counseled.

The possibility of multiple pregnancies from IVF is another very sensitive question. The chance of survival of a single embryo is rather small; therefore, multiple embryos are often implanted to increase the chance of pregnancy. However, this also significantly increases the likelihood of multiple pregnancies, especially in comparison with traditional conceptions (7). Multiple pregnancies could influence the physical and mental health of the mother, and also present a threat to the well-being of the expected offspring. Medical students must be introduced to these problems and should be encouraged to work out how to find answers to these ethical issues. A medical doctor must be able to explain to future parents all the dilemmas based on scientific facts and should be able to help future parents realize that despite the disadvantages of this method, without it, these children would not be born at all.

Identification of genetic susceptibility to common diseases

Completion of the Human Genome Project has enabled the study of the genetics of common complex disorders such as cardiovascular diseases, cancers, and diabetes (9). Both genetic and environmental factors play a role in the etiology of these diseases.

Several human genetic studies are aimed at discovering genetic susceptibility to common diseases that can provide early diagnosis and better treatment strategies for the diseases, and even their prevention (1, 9, 10). Genetic tests are increasingly accessible to physicians, companies, and the general public (17). However, individuals often perceive susceptibility genes as being more fateful than other risk factors such as excessive use of medication, smoking, alcohol abuse, or diet abnormalities. In these cases, the role of the medical doctor is important. Before a doctor gives genetic advice, ethical considerations are required.

Medical students must realize that physicians should explain in a delicate manner that genes are not the only factors to predict the development of a disease, and that it is very important to follow a healthy lifestyle. For example, if an individual is determined to have highly-penetrant breast cancer gene BRCA 1, the physician should not convey this information without context. The physician must explain the concept of “lifetime risk,” present the possibilities of preventive surgery used in cases of high risk, and discuss efficient treatment methods. The physician’s task is to assist in developing a plan for a healthy lifestyle in a friendly atmosphere (e.g., by suggesting restricting the use of hormonal contraceptives, quitting smoking, or having a physically active lifestyle). In such cases, evidence about families with individuals who have the gene and did not develop cancer, even late in life, could be very reassuring. The ethical issues described above highlight the importance of “genetic literacy” for both the counselor and counselee (5). Another aspect is the degree to which the counselee can apply the information given by the counselor. The more complex the inheritance of the disease, the greater the ethical responsibility that lies with the medical doctor who gives genetic advice.

Interpretation of minor physical anomalies (MPAs)

MPAs are mild, clinically and cosmetically not significant errors of morphogenesis. They may provide information on neurodevelopmental genetic disorders for diagnostic purposes. Investigation of the prevalence of minor physical anomalies in mentally healthy individuals using the Waldrop Physical Anomaly Scale shows a low mean score of minor physical anomalies (12). Several studies (14, 16) have shown higher frequencies of MPAs in patients with neurodevelopmental disorders. This enables MPAs to be used as indicators of some neurodegenerative developmental genetic disorders (e.g., autism, Tourette syndrome). It is also well known that some MPAs, such as the epicanthal fold and curved fifth finger, are rather common in the general population (11). Therefore if MPAs are used to demonstrate the phenotype of an affected person, the lecturer should stress that MPAs are detected in healthy individuals too, and only a specific set of MPAs along with other phenotypic markers could be considered a phenotypic marker for genetic disease. A medical doctor should know and be able to explain in which cases a minor anomaly is a sign of a major health problem,

and together with the patient or patient’s relatives decide about further investigation.

Presentation of clinical cases

Case reports are an essential element in teaching medical students. Only previously published cases are used in lectures. Consent to use a patient’s story should be obtained prior to publishing the report whenever possible (15). Clinical cases serve to advance students’ understanding of genetic diseases, as well as to bridge the gap between theoretical and clinical subjects. Key elements for successful use of clinical cases in teaching genetics include demonstration of a respectful attitude toward the patient and his/her family on the part of the medical doctor. Usually, clinical cases involve pictures of affected individuals. The lecturer must remind the students that the picture is not simply an illustration of the disease, but that the person with a particular condition portrayed in the picture is real and deserves respect. Frequently integrated case reports in the lecture may help prepare students to face a real clinical environment, and their use expands students’ knowledge in genetics. For example, the lecturer presents a case about a 40-year-old female, Mrs. X, who tells the physician that her sister has been diagnosed with breast cancer at the age of 38 due to a mutation in the BRCA 1 gene. Family history reveals that her mother, aged 65, is healthy and never had a cancer diagnosis, whereas the father’s sister died from ovarian cancer at the age of 52, and the father’s mother died at the age 54 from breast cancer. If Mrs. X would like to know whether she should be concerned about the sister’s diagnosis, medical students must be able to give information on this mutation. The lecturer has to point out that a genetic counselor should inform the patient in a delicate manner about lifetime risk for breast cancer, which ranges from 40 to 85 percent, while for ovarian cancer it is lower, namely, 16 to 60 percent (18). The patient should be informed about the possibility of molecular diagnosis to learn whether she has this mutation, and the medical doctor should explain that the gene comes from her father’s side of the family. It should also be made clear that even if the mutation is detected, it does not mean that cancer will develop. In such situations, the medical doctor has to give detailed information on promoting a healthy lifestyle to avoid the development of cancer to the maximum extent possible. In such delicate situations, the medical doctor faces some ethical dilemmas, such as whether it is ethical, if Mrs. X has female cousins on her father’s side, to inform them about the possible increased risk of having mutation of this gene. Another dilemma is whether Mrs. X should be encouraged to share this information with her husband and her two daughters.

Contextual use of terminology

Due to increasing information in the field of genetics, scientific terminology is essential for proper presentation of

information during lectures in genetics. The lecturer must take into account that some students might not have come across genetic terminology during their previous studies. Therefore, the lecturer should convey information in a way that is accessible to all students. In these situations the lecturer can use scientific terminology in specific contexts. Some examples are given below.

- i. “Advanced maternal age” is defined as over 34 (age of 35 at delivery). The majority of textbooks contain information that there is an increased risk for nondisjunction of chromosomes at this age, which leads to trisomy in the fetus. Such information should not be provided without explanations on various testing methods during pregnancy, and possible solutions in case of detection of trisomy.
- ii. “Lifetime risk” is defined as the risk of developing a disease over the course of a lifetime. The lecturer should stress that risk is almost never 100% and even a person with increased susceptibility can prevent development of the disease.
- iii. “Carrier” is a person who has a mutant allele of a gene in their genotype. Several variants of carrier status should be discussed. In the case of a recessive mutation, the carrier is not at higher risk of developing the disease. On the other hand, if a female is a carrier of a mutant gene located on the X chromosome, she will most likely not develop symptoms of the disease, but she is at high risk (50%) of passing on this mutant gene to male offspring, who will be affected. If an individual has a dominant mutant gene, then different possibilities should be considered. If a gene with incomplete penetrance is detected, then only those with a gene expressed in the phenotype should be considered as affected. If a gene shows late onset, the probability of being affected depends on the age of the person.

There are a many examples, and in each case the lecturer should consider the use of terms with caution and create a safe emotional environment for students.

As lecturers, we should remind our students that a medical doctor’s task is to educate the general public and, with our attitude, demonstrate that the affected person is not a burden, but an object of love, affection, and, most importantly, respect.

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