Ethical problems associated with prenatal diagnosis

Problemy etyczne wiążące się z diagnostyką prenatalną

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Abstract

Prenatal diagnostic methods and principles as well as broadly understood moral values constitute a subject of considerations concerning issues of ethical nature. The main aim of prenatal examinations is the desire to broaden the knowledge regarding the health of the fetus. The procedures implemented during prevention and treatment examinations result in the possibility of excluding genetic defects and conducting treatment. The legal aspects and the ethical and moral values are frequently of different nature. Prenatal diagnosis is conditioned by cultural, religious, moral, and social factors. The awareness of parents and medical personnel regarding the possibilities of reducing doubts pertaining pregnancy increases together with the advancement of knowledge on examinations of children in the prenatal period. The contemporary eugenics enable us to undertake broader health-related measures regarding the unborn child. It should be remembered that prenatal examinations have many advantages, such as identification of high-risk pregnancies and fetuses with anatomical or functional defects. These examinations may lead to the application of adequate measures thank to which the child will have the best possible chance of receiving medical assistance.

Key words: ethics, prenatal examination, morality, medical ethics

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Streszczenie

Metody diagnostyki prenatalnej oraz zasady i szeroko pojęte wartości moralne są obiektem rozważań problematyki o charakterze etycznym. Głównym celem przeprowadzania badań prenatalnych jest chęć poszerzenia wiedzy o stanie zdrowia płodu. Postępowanie wdrażane podczas badań prewencyjnych lub leczniczych skutkuje możliwością wykluczenia wad genetycznych i leczenia. Aspekty prawne oraz wartości etyczno-moralne nierzadko mają odmienny charakter. Diagnostyka prenatalna warunkowana jest przez czynniki kulturowe, religijne, moralne oraz społeczne. Wraz ze stopniem zaawansowania wiedzy na temat badań dzieci w okresie prenatalnym wzrasta świadomość rodziców oraz personelu medycznego na temat możliwości zmniejszenia wątpliwości dotyczących ciąży. Współczesna eugenika pozwala nam na podjęcie szerszych działań zdrowotnych wobec nienarodzonego dziecka. Należy pamiętać, iż badania prenatalne mają wiele zalet, takich jak identyfikacja ciąż podwyższonego ryzyka, płodów z wadami anatomicznymi lub funkcjonalnymi. Badania te mogą prowadzić do zastosowania odpowiednich kroków, aby dziecko miało możliwie jak największe szanse na otrzymanie pomocy.

Słowa kluczowe: etyka, badanie prenatalne, moralność, etyka lekarska

Introduction

Welcoming a child to the world is an important moment in the life of parents. They experience distinctive emotions and feelings, dominated by concern regarding the health of the unborn child, while waiting for the child's arrival. Thanks to the progress in the field of genetics, in particular concerning diagnostic methods, the contemporary medicine makes it possible to conduct prenatal examinations and thus obtain information regarding the developing fetus. Already at an early stage of the pregnancy, the doctor can gain knowledge regarding the possible risks to the fetus and undertake preventive treatment measures; future mothers, on the other hand, are able to rule out serious genetic and developmental defects of the fetus.

Prenatal diagnosis comprises all examinations which can be performed before the birth of the child, both invasive and non-invasive. Invasive techniques include amniocentesis, fetoscopy, cordocentesis, and biopsy of trophoblast villi, while non-invasive – ultrasound and biochemical tests of the markers produced by the fetal/placental unit in mother's serum as well as analysis of fetal cells circulating in the mother's bloodstream. The main subject of discussions and dilemmas are the invasive methods, which bear a small risk of pregnancy loss[1] as they interfere with the interior of the fetal/placental unit. However, they are always performed under ultrasound surveillance. It should be emphasized that the contemporary methods of prenatal diagnosis are characterized with increasing safety rate.[2]

The ethical problems associated with prenatal diagnostic examinations pertain mainly to their purposefulness, the dangers associated with the examinations performed with the use of the invasive methods as well as possible termination of pregnancies due to inauspicious results of these examinations. The indicated problems entail a number of moral doubts and controversies both among doctors and parents. Currently, the most serious ethical dilemma is the very issue of the purpose of the examinations and the circumstances in which they are conducted. This paper touches upon only a few ethical problems appearing in the discussion on prenatal diagnosis, as each of the indicated dilemmas brings new questions, and the subject of these considerations can be discussed on many levels, referring to the dilemmas from the perspective of parents or physicians.

The basic principle of medical ethics, that is acting for the benefit of the patient, also applies to prenatal diagnosis. Genetic examinations, in accordance with the main principle of physicians' conduct, should be performed in accordance with the current medical knowledge. *The Code of Medical Ethics* states that the physician is obliged to inform the patient about the possibilities of contemporary medical genetics, diagnosis and prenatal treatment, as well as risks associated with the performance of prenatal examinations.[3] The genetic examination should be conducted only with patient's informed consent. This is where the first ethical dilemma appears. To what extent is the consent of the mother informed and should the fetus also be treated as a patient? In fact – yes, as the *Code* indicates that the fetus is, indeed, a patient, and the doctor, in accordance with the ethics, is to take care of both the health of the parent – by informing them about the health status of the fetus – and of the yet unborn child – through the diagnosis and possible treatment.[4]

Another issue is the ethical problem of respecting the dignity of the child and its right to life. In accordance with Art. 39 of the *Code of Medical Ethics*, in the case of a pregnant woman, the physician is responsible both for her health and life and her child's health and life, also before its birth. How can we, therefore, refer to the right to life from the perspective of the physician, when the diagnosis itself can lead to pregnancy termination, depending on its results? The Catechism of the Catholic Church presents a clear position on the subject, indicating that while prenatal diagnosis itself is morally acceptable – when the life of the child is respected and the diagnostic examinations are performed in order to protect it or for the purposes of potential treatment – the examinations should not entail a death sentence.[5] In

the case of physicians, it should be indicated that their highest precept is the good of the patient – *salus aegroti suprema lex esto*, also with regard to the unborn child, which is also treated as a patient. How should we, therefore, refer to the life of the child in accordance with the principle of respect for human life and the principle of justice? Does pregnancy termination performed by a physician as a result of prenatal examinations violate the principle of justice, which is guided by equal treatment? In this situation, both the child and the mother should have an equal right to life.

In accordance with the Act of January 7, 1993 on Family Planning, Protection of Human Fetus and Pregnancy Termination Conditions, pregnancy can be terminated only by a physician in the case that prenatal examinations or other medical rationales indicate that there is a high risk of severe and irreversible impairment of the fetus or an untreatable disease threatening its life (up to 22nd week).[6] In reality, a woman entitled to pregnancy termination may experience difficulties executing her right. In accordance with the decision of the Constitutional Court of October 7, 2015, a physician may invoke to so-called conscience clause, under which they are – among other things – not obliged to terminate pregnancy. The moral law always pertains to the human being as a whole person. Even if a given act is not regulated by law, it is subject to moral evaluation.

The roots of abortions stemming from inauspicious prenatal diagnosis reach the eugenic postulates. Ethical discussion related to prenatal diagnosis often refers to the problem of the association of such diagnosis with the aforementioned ideology. The term "eugenics" was coined in the 1880s and means selection based on specific genetic characteristics. It is a set of views that assumes the possibility of perfecting the hereditary characteristics of the human being, the goal of which is to establish and consolidate the conditions enabling the development of positive hereditary characteristics and reduction of the negative ones. We can basically distinguish 2 types of eugenics – positive and negative. The positive is based on birthing children with characteristics generally perceived as desirable, while the negative is based on the prevention of birthing ill children.[7,8]

The term "eugenics" used to refer to larger groups or populations, not individuals, and meant planned and forced policy aimed at the creation of a pure race. Genetic defects and impairments were seen as "bad genes".[9] New eugenics is characterized by the emphasis on the individual aspect of prenatal examinations, without taking into account the social issues. However, the ethical problem of prenatal research associated with the new eugenics remains. Should we treat the contemporary diagnostic and genetic examinations as elimination of fetuses? Should we decide to conduct the examinations knowing that it may bring new dilemmas? Or, perhaps, we should remain in ignorance until the moment of birth?

For example, in Germany, more than 90% of cases of prenatally diagnosed genetic diseases end in abortion on medical grounds. In France, half of Down syndrome cases in the years 1988–1992 were diagnosed prenatally, and the majority of those pregnancies – with small exceptions – were terminated.[10] However, the Jérôme Lejeune Foundation, which supports mothers after the birth of a child with a Down syndrome and during the process of raising the child, operates in that country.[11] In many cases, upon receiving the inauspicious diagnosis, parents decide to have the child and prepare for its arrival. In such situations, prenatal diagnosis of a genetic disease is of great importance, since in the case of genetic syndromes with non-characteristic phenotype post-natal diagnosis is sometimes delayed, which results in the lack of early rehabilitation. If the genetic syndrome is diagnosed prenatally, specialist stimulation of child's development can be conducted and thus its chances for life in the future can be increased.[12] The decision on conducting prenatal diagnostic examinations is very frequently understood unambiguously as a decision to terminate the pregnancy in the case the results are adverse. In reality, early diagnosis of congenital malformations and genetic syndromes in the prenatal period makes it possible for parents to prepare to make an autonomous decision. The question regarding prenatal eugenics is also the question regarding social awareness of the definition of disease or the condition of the society concerning genetic counselling and impartiality while informing the patient. Sociological studies involving recording of actual genetic counselling in the form of films showed that geneticists to a significant extent suggest the course of action to the patients.[13] The level of paternalism in the advice was higher in cases of patients of lower socio-economic status. It should be indicated that some of the parents who decide to terminate the pregnancy feel unable to accept the child, as they see no possibility of making a living and are afraid of rejection and social isolation, as well as of the fact that the child's life will be filled with suffering. In such cases, genetic counselling in conjunction with psychological and material is extremely important and may have a significant impact on the decision of the parents, which should still remain independent.[14]

Another ethical problem concerning prenatal examinations is that of human rights to happiness and health in the context of severe impairment, when no effective treatment exists. Supporters of abortion believe that it is the only way not to cause misery to the child. In medical-legal language, such cases are referred to as eugenic indication.[15] In this matter, Christian ethics postulates that the problem be viewed from the perspective of the unborn child and urges to consider to what extent the disease of the fetus constitutes a violation to its right to be healthy and happy.[16]

Prenatal examinations, however, should not be viewed solely as problems and threats, as early detection of con-

genital malfunctions and genetic diseases in the fetus also results in the possibility of saving the child, or reduces the risk of perinatal complications in the case of medical intervention during the period of intrauterine life. There are many situations in which prenatal detection of disorders and implementation of therapeutic action during the pregnancy is the only chance for the child. Prenatal diagnosis is therefore a way for the society in terms of future-oriented approach to medicine and possible ways to seek methods to overcome health problems.

In a certain way, the contemporary world imposes the responsibility for health on each of us, but not only with regard to an individual, as each one of us bears the responsibility towards the future generation. A woman is required to do everything in her power to give birth to a healthy child, which might be to a certain extent considered as cultural pressure. The access to prenatal examinations imposes the decision to undergo them, as omission of prenatal diagnostic tests and giving birth to a disabled child is currently viewed as an irrational and immoral act. It is assumed that the desire to obtain information and actions aimed at reducing the risk are rational. It is the accessibility of prenatal examinations that should induce women to at least consider performance of diagnostic tests. However, the question should be raised whether prenatal examinations are an independent choice, or, perhaps, are to a certain extent imposed by the contemporary expectations of the society? The awareness of the fact that a disabled child can encounter lack of acceptance of its peers, together with cultural pressure, the pressure exerted by media on health and healthy lifestyle, and with knowledge concerning the cost of medical care regarding genetic diseases or impairments jointly lead to a conclusion that it is difficult to determine whether these choices are independent. Health, however, is not a value superior to freedom. Each one of us should be entitled to make their own choice. A woman who has prenatal examinations conducted and subsequently finds out that her child is suffering from an incurable disease faces a dilemma; however, in accordance with patient's rights, she also has a free choice as a patient.

Should abnormalities be diagnosed in the nasciturus, the physician is obliged to provide the parents with as much information as possible regarding the given defect and possible forms of help. They cannot, however, suggest abortion. In the situation in which the genetic defect of the child results in a legal possibility of pregnancy termination, but financial support and the support of patient's loved ones are not provided, the choice becomes social and parents' independence is limited. There are, however, many situations when parents decide to continue the pregnancy despite the lack of effective treatment. Perinatal hospice, offering medical, psychological and spiritual support for parents as well as medical care for the child, could be a solution.[9] How should we address the choice of parents who decide to have a child with diagnosed genetic defect? Not much can be said about human dignity, which is the source of all rights of the child from the moment of conception. It is an ungradable category. The act being in itself is sufficient to be bestowed with dignity. Human dignity is closely associated with the fact that the human being cannot be treated as a means to an end.[13] The decision of the parents is not only an obligation to provide care, but also a consequence for the child itself. According to the teleological theory, moral evaluation of choice is negative due to the criterion of wrongdoing.[18] According to the Polish law, protection of human life is guaranteed by the Constitution. Article 38 of the Constitution provides that the Republic of Poland guarantees legal protection of life to every human being,[19] whilst John Paul II mentioned that "[t]he human being should be respected and treated as a person from the moment of conception and, therefore, it should be granted the rights of a person, including primarily the right to life of every innocent human being, that very moment".[20]

Prenatal examinations entail many moral dilemmas not only for parents-to-be, but also the medical staff involved in the diagnostic process. Should the results be inauspicious, the future parents may experience dilemmas concerning further steps regarding continuation or termination of pregnancy. We must remember, however, that a lot of good stems from prenatal examinations as they enable identification of high-risk pregnancies and fetuses with anatomical or congenital defects, thanks to which appropriate medical measures can be taken and children can be born at such place and time when they can be provided with medical assistance.

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