

Ethical Dilemmas and Challenges in the Application of Prenatal Diagnostics

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Abstract

Prenatal diagnostics is one of the most dynamic areas of modern medicine. Its application provides invaluable benefits through early detection of fetal malformations, timely planning of delivery, and the possibility of intrauterine therapeutic interventions. At the same time, it opens a series of ethical challenges related to maternal autonomy, the dignity of the unborn child, the responsibility of physicians, and the role of society.

The article presents the fundamental principles of medical ethics – autonomy, beneficence, non-maleficence, and justice – as key guidelines in decision-making. Special attention is devoted to the dangers of test commercialization, the risk of false-positive results, and the possibility of discrimination based on sex or disability. Emphasis is placed on the importance of trust and quality communication between the physician and the family, as well as the need for equal access to modern methods regardless of socioeconomic status.

Future developments in prenatal diagnostics, including the application of artificial intelligence and genomic analysis, raise new questions about the boundary between medically justified procedures and covert eugenics. Therefore, prenatal diagnostics cannot be viewed merely as a medical procedure but also as a socio-ethical process in which personal decisions, legal frameworks, and cultural values intertwine.

In conclusion, prenatal diagnostics justifies its place in modern medicine only if it remains focused on the protection of life and respect for human dignity. This requires ongoing dialogue between medicine, ethics, and society, as well as strong social support for families with children with disabilities, since their integration and acceptance represent a measure of the humanity and maturity of the community.

Keywords: prenatal diagnostics, medical ethics, autonomy, beneficence, non-maleficence, justice, fetal anomalies

Introduction

Prenatal diagnostics encompasses a set of procedures and methods used to assess the health, development, and genetic status of the fetus before birth. The primary goals are the detection of structural anomalies, chromosomal abnormalities, and genetic and metabolic disorders in the early stages of pregnancy, thereby enabling timely counseling, perinatal planning, and, in certain cases, therapeutic interventions (1,2).

The development of prenatal diagnostics began in the second half of the 20th century with the introduction of ultrasound into everyday clinical practice and later expanded with the introduction of invasive methods (amniocentesis, chorionic villus sampling) as well as sophisticated molecular and genetic tests (3–6). These methods have greatly contributed to a better understanding of intrauterine development and have enabled advances in safeguarding life, both through the early detection and treatment of certain fetal diseases and through the planning of safe delivery in specialized centers (7).

Despite continuous improvements in these methods, it is important to note that approximately 2–3% of children are born with congenital malformations, while the vast majority (about 97–98%) are born healthy (8). It is precisely in this minority of pregnancies that the key ethical dilemmas of prenatal diagnostics arise. While the main purpose of prenatal diagnostics is to improve the health of both mother and child, in practice, diagnostic findings often provoke fear, uncertainty, and pressure in decision-making, even when the pregnancy outcome ultimately proves favorable. Thus, an atmosphere is frequently created in which pregnant women are preoccupied in advance with the possibility of an adverse outcome, although such cases are relatively rare.

Although the fundamental medical purpose of prenatal diagnostics is to improve pregnancy outcomes and maternal and child health, ethical dilemmas arise when diagnostic findings become the decisive factor in parental decisions regarding the continuation or termination of pregnancy. At

the heart of these dilemmas lies the balance between the autonomy of the pregnant woman, the rights and dignity of the unborn child, and the broader social context of selective reproductive decision-making (9).

Methods Used in Prenatal Diagnostics

The development of prenatal diagnostics has brought a wide range of methods currently used in clinical practice. These are most often divided into non-invasive and invasive procedures, while molecular and genomic methods have become increasingly important in recent years. Preimplantation genetic diagnostics, performed exclusively within the framework of in vitro fertilization procedures, also occupies a special place.

Among non-invasive methods, ultrasound diagnostics is the foundation of prenatal screening and has been in routine use for decades. Standard ultrasound examinations in the first and second trimesters allow assessment of fetal anatomy, detection of most major malformations, and evaluation of growth and development (1,10).

Of particular importance is the so-called mini anomaly scan, performed at the end of the first trimester, most often between 11 and 13+6 weeks of gestation. In addition to the basic measurement of crown-rump length (CRL) for precise determination of gestational age, the scan includes assessment of nuchal translucency, presence of the nasal bone, blood flow in the ductus venosus, flow across the tricuspid valve, as well as early morphological indicators of brain, facial, abdominal wall, spine, and limb development (7,10). This examination, in combination with biochemical markers, significantly increases the sensitivity of screening for chromosomal anomalies and enables early detection of a large number of structural malformations.

First-trimester combined screening includes nuchal translucency measurement by ultrasound and biochemical markers (PAPP-A and free β -hCG) in maternal blood. This method, performed between 11 and 13+6 weeks of pregnancy, has high sensitivity for trisomy 21 and other common chromosomal abnormalities (11). When combined with

maternal age and other risk factors, it achieves detection of about 85–90% of Down syndrome cases, with an acceptable false-positive rate (12).

Non-invasive prenatal testing (NIPT) of cell-free fetal DNA in maternal blood represents the most important advancement in modern screening for chromosomal abnormalities. This method is based on the analysis of fragmented fetal DNA circulating in maternal plasma outside fetal cells and provides very high sensitivity and specificity for the most common aneuploidies (trisomy 21, 18, and 13) (13). Although NIPT has great clinical value, it is a screening rather than a diagnostic method, since the analyzed fraction of fetal DNA originates from the placenta; therefore, abnormal results require confirmation by invasive procedures (14).

Historically, in the second trimester, chromosomal abnormalities were screened using the double test (AFP and hCG), later the triple test (AFP, hCG, and estriol), and eventually the quadruple test (with the addition of inhibin A). These tests today have very limited clinical value, having been replaced by combined screening and NIPT, given their significantly lower sensitivity and specificity (15).

Invasive methods include chorionic villus sampling (CVS) and amniocentesis (AC). These represent standard invasive procedures for obtaining fetal genetic material. CVS is most often performed in the first trimester, while AC is performed after the 15th week of pregnancy. Both procedures are associated with a small but non-negligible risk of miscarriage, estimated at 0.1–0.3% in experienced centers (16). Cordocentesis, or fetal blood sampling from the umbilical cord, is less frequently used today due to its technical difficulty and higher risk, but still plays an important role in specific indications, such as diagnosis and treatment of fetal anemia or infections (17).

The introduction of chromosomal microarray analysis (CMA) enabled the detection of submicroscopic deletions and duplications, which significantly increased diagnostic yield compared to standard karyotyping (18). In addition, next-generation sequencing

(NGS), including whole exome sequencing (WES) and whole genome sequencing (WGS), is becoming increasingly available in prenatal diagnostics, particularly in cases of complex malformations when standard methods fail to provide a clear answer (19). However, the application of these methods also raises numerous ethical dilemmas, including the interpretation of variants of uncertain significance, secondary findings, and the possibility of predicting diseases that manifest later in life.

A special category is preimplantation genetic testing (PGT/PGD), performed exclusively within in vitro fertilization (IVF) procedures. This method enables genetic analysis of embryos before uterine transfer, detecting chromosomal and certain monogenic diseases (20). The primary purpose of PGT is to prevent the transmission of severe genetic diseases in families with known risk, while in practice, it is also often used in the context of PGT-A (preimplantation genetic testing for aneuploidy) to increase the probability of implantation and reduce the risk of miscarriage. Although this is a highly sophisticated method, its application raises numerous ethical issues, including embryo selection, the possibility of “positive eugenics,” and the boundaries of reproductive medicine.

Information, Communication, and Trust in Prenatal Diagnostics

It is important to emphasize that the findings of prenatal diagnostics are not always final or unambiguous. Ultrasound markers, biochemical screenings, and even the most advanced genetic tests have certain rates of false-positive and false-negative results (9,21). For example, increased nuchal translucency may indicate a chromosomal abnormality, but it can also be a transient variant of normal development; NIPT is extremely reliable for trisomy 21, but it is not a diagnostic test and requires confirmation by invasive methods; even microarray and sequencing can yield findings of uncertain clinical significance. For this reason, decisions regarding further management must not be based on a single result alone. From both an

ethical and medical standpoint, it is justified to recommend that the pregnant woman and her partner seek additional consultation, whether with an experienced perinatologist, geneticist, or multidisciplinary team. Such an approach reduces the risk of wrong decisions and provides parents with a sense of security and trust. This represents an important ethical moment: recognizing the limits of one's own knowledge or methodological limitations and referring the patient for additional expert evaluation.

In all dilemmas that accompany prenatal diagnostics, the fundamental orientation must always be the pursuit of good. This applies not only to the individual, but also on several levels: the good of the pregnant woman, the good of the unborn child, the good of the family, and the good of the community as a whole. In practice, this means that the physician must evaluate not only medical data, but also the broader psychological, social, and moral context. Of utmost importance is high-quality, complete, and timely information. Only a well-informed pregnant woman, together with her partner, can make a decision that is truly autonomous and responsible. In this process, the responsibility of the physician is enormous, as the way findings and recommendations are presented can decisively shape the decision (22,23). If prenatal diagnostics leads to an early therapeutic intervention that saves the child's life, it is a clear example of achieving good. However, if a procedure carries a risk of greater fear, suffering, or the loss of a healthy child, then it is not consistent with the principle of good. The same applies to decisions that affect the entire family, since pregnancy outcomes shape not only the lives of the mother and child but also the lives of the partner, siblings, and even the wider community.

If only the negative aspect of a finding is emphasized, the pregnant woman may experience unnecessary fear and pressure, while on the other hand, simplified or overly reassuring interpretations may underestimate the real risk. Therefore, the physician finds themselves in a sensitive role as mediator between medical facts and the

personal values of the family, where expertise must be united with compassion and ethical responsibility (24).

This dimension of communication represents a bridge between ethics and clinical practice. The way in which the physician conveys information and builds trust with the pregnant woman and her partner directly influences the reduction of ethical dilemmas and unnecessary fear.

Prenatal diagnostics does not end with making a diagnosis; it only then acquires its full meaning through communication between the physician and the pregnant woman, and her family. The manner in which information is transmitted decisively affects decision-making and shapes the overall pregnancy experience (9).

The physician is not only a technical executor of diagnostic procedures, but also a mediator between medical facts and the life decisions of parents. The decision of the pregnant woman and her family is primarily based on the information they receive from the physician, which makes their responsibility extremely significant. Information must be accurate, clear, and comprehensive, while it is also important to avoid suggestive or alarming formulations that could compromise the pregnant woman's autonomy in decision-making. It is equally necessary to emphasize the limitations of specific methods, and not only highlight their diagnostic power, so that parents gain a realistic picture of the value and reliability of the findings (25). The physician must always speak the truth: it is equally wrong to downplay the seriousness of the condition to "sweeten the situation" as it is to exaggerate the problem and present it as more serious than it actually is. Both approaches undermine trust and can lead to decisions that are not based on actual facts (26).

The physician must convey only those pieces of information of which they are completely certain. When there is doubt or uncertainty about the accuracy of findings or the diagnosis, this should be clearly emphasized and presented to the pregnant woman and her partner. In such cases, it is responsible

to recommend further diagnostic workup and the inclusion of other specialists, such as a clinical geneticist, fetal cardiologist, or neonatologist (27). This approach reduces the risk of wrong decisions and provides parents with confidence in the medical process.

Clinical experience shows that when confronted with an unfavorable prenatal finding, the pregnant woman is often unable to remember or transmit all the information provided by the physician. For this reason, it is advisable that the partner, and when necessary, other family members, participate in the communication. Their presence strengthens the sense of togetherness and shared responsibility, reduces the risk of misunderstandings or incomplete information, and facilitates decision-making that is crucial for the family's future (28, 29). Such an approach increases the woman's sense of security, since the burden of difficult information and decision-making is shared with a trusted person and close family.

An unfavorable prenatal finding does not affect only the medical dimension of pregnancy but also deeply impacts the emotional balance of the woman and her family. Fear, uncertainty, sadness, and feelings of guilt are common parental reactions to such information. Therefore, the physician must show empathy and understanding in communication, but also recognize when the emotional burden is so strong that it exceeds their professional competence. In such situations, psychological support specialists should be involved to provide comprehensive care and to enable parents to make decisions that are not only informed but also emotionally balanced (30).

Complex clinical situations, such as severe fetal malformations or suspected rare genetic syndromes, require collaboration among multiple specialists. In such circumstances, the process should involve a gynecologist, clinical geneticist, neonatologist, pediatric cardiologist, psychologist, and, if necessary, an ethicist. A multidisciplinary approach ensures greater professional safety, reduces the risk of wrong decisions, and provides parents with the feeling that their child and family are being cared for comprehensively

and responsibly (31). Beyond raising the quality of medical decision-making, this model reduces the sense of isolation of the pregnant woman and her partner, since the decision is not left only to them and one physician, but is made within a team framework.

Ultimately, all ethical dilemmas and clinical uncertainties in prenatal diagnostics boil down to the relationship of trust between the physician and the patient. Trust is built on truthful and clearly conveyed information, the physician's willingness to admit the limits of their certainty, the inclusion of the partner and multidisciplinary team, and recognition of the family's emotional needs. When trust exists, prenatal diagnostics becomes a process that brings not only medical benefits but also human security and a sense of togetherness. Without trust, even the most sophisticated diagnostic procedures lose their value and may become a source of insecurity, fear, and irreversible wrong decisions (32).

Ethical, Legal, Cultural, and Social Framework of Prenatal Diagnostics

Prenatal diagnostics does not take place in a vacuum, but within a broader social, legal, and cultural framework. Attitudes toward its purpose and limits differ significantly among countries and communities. While some emphasize the autonomy of the pregnant woman and her right to choose, others begin from the dignity of unborn life and highlight the obligation to protect the fetus. Understanding these differences is essential for a comprehensive assessment of ethical dilemmas.

In Catholic bioethical tradition, prenatal diagnostics is acceptable and justified when aimed at protecting the health and life of the mother and child, or when it helps prepare for therapeutic or palliative approaches (33). However, it is unacceptable if used as a tool for pregnancy selection and discrimination of the unborn based on sex, disability, or genetic traits. Catholic ethics emphasizes that the value of life does not depend on its "quality," but on the inherent dignity of the human person.

Secular bioethics primarily rests on the principle of the pregnant woman's autonomy and her right to decide on the course of pregnancy. In this context, fetal rights are not recognized to the same extent as maternal rights, with the emphasis placed on freedom of choice and informed decision making (9). In many health systems, prenatal diagnostics is also considered from a public health perspective to reduce the incidence of severe malformations and genetic diseases. However, this approach may raise the question of the boundary between legitimate care and covert eugenics.

Furthermore, the legal regulation of prenatal diagnostics and pregnancy termination varies greatly: from liberal systems (e.g., the Netherlands, Sweden), where termination is permitted until late gestational weeks, especially in cases of severe fetal malformations (34); to restrictive systems (e.g., Poland, Malta), where termination is allowed only in exceptional circumstances, such as threats to the mother's life (35).

These differences directly shape everyday clinical practice, since the availability of prenatal methods becomes meaningless if the legal framework does not allow decisions resulting from diagnostic findings.

Cultural factors strongly influence perceptions of prenatal diagnostics. For example, in some Asian countries, there is the problem of sex-selective abortions, while in Western societies, concern is growing about "quality-of-life screening" and the possible stigmatization of people with disabilities (36). In cultures that emphasize family values, pregnancy decisions are more often made collectively, involving the partner and extended family, whereas in individualistic cultures, the emphasis remains on the personal autonomy of the pregnant woman (37).

Ethical questions and dilemmas during the prenatal period cannot be limited only to the moment of diagnosis and the decision on the course of pregnancy. Equally important is the question of what kind of support society provides to families who decide to accept a child with disabilities. Parental decisions

are often influenced by perceptions of the quality of life of the child and family, which are in turn shaped by the availability of social resources and the level of solidarity within the community (38).

Examples of good practice include prenatal hospices, which offer palliative care and psychological support to parents facing life-limiting diagnoses (39). These models allow pregnancy to be completed with dignity, without the feeling that termination is the only option. Furthermore, society can significantly contribute to the quality of life of children with disabilities and their families by ensuring parental rights and benefits, access to specialized kindergartens and schools, adapted public transportation and infrastructure, and systematic psychological and social support (40).

When society provides such forms of support, parents are more likely to continue the pregnancy and accept a child with disabilities, as they feel they will not be left alone to face the challenges. In this way, prenatal diagnostics retains its original purpose—to help in life planning and care, rather than becoming a mechanism of selection (41).

Conclusion

Prenatal diagnostics is one of the most dynamic areas of modern medicine. Its development brings invaluable benefits for maternal and child health, enables early recognition of malformations, timely planning of delivery in tertiary centers, and even intrauterine therapeutic procedures that significantly improve outcomes. However, these same diagnostic tools also open complex ethical and social issues: from confronting the possibility of pregnancy termination to the dangers of covert eugenics, commercialization of testing, and the deepening of social inequalities.

All ethical dilemmas can be viewed through the four fundamental principles of medical ethics – autonomy, beneficence, non-maleficence, and justice. Autonomy requires that the pregnant woman and her family receive truthful, complete, and clearly

explained information in order for decisions to be free and responsible. Beneficence obliges the physician not only to technical expertise but also to providing support, empathy, and acceptance. Non-maleficence reminds us that no method should bring greater psychological or medical harm than benefit, especially in the context of false-positive results or commercialized testing. Justice demands equal access to modern methods for all pregnant women, regardless of their socioeconomic status, as well as respect for the equal dignity of every unborn child, without discrimination based on sex, disability, or genetic traits.

One of the central questions remains the boundary between legitimate care and covert eugenics. When prenatal diagnostics serves to improve health outcomes and plan therapy, it is ethically justified. But when it results in selective terminations due to disability or sex, it becomes a tool of discrimination and social pressure. The same applies to the rapid technological progress in genome sequencing and artificial intelligence: while it promises more precise diagnostics, it also raises new questions about algorithm reliability, predictive genetics, and the very notion of human identity.

The future of prenatal diagnostics will therefore depend not only on technological progress but also on the responsibility of society. If the community ensures quality living conditions, adequate health care, social support, and inclusive education for families with children with special needs, the perception that termination is the only option will diminish. In this way, society becomes not just a passive observer of ethical positions, but their active shaper.

The physician's role in this context remains crucial. They are not only a diagnostician but also a companion to the family, a guardian of trust, and the one whose manner of communication can empower parents to make decisions in accordance with their values.

In conclusion, we face questions to which medicine and society must yet provide clear answers: how to prevent the misuse of prenatal diagnostics for discrimination,

how to ensure equal access to modern methods, how to set boundaries between medically justified procedures and positive eugenics, and how to reconcile technological progress with the dignity of human life. The answers to these questions concern not only the individual but also shape the future of families and society as a whole.

Only if prenatal diagnostics remains in the service of life and dignity will its development be fully justified. And this requires ongoing dialogue between medicine, ethics, and society, directed toward the well-being of families with children with disabilities, since their integration and support represent a measure of the humanity and maturity of every community.

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