Bioethical aspects of prenatal diagnosis

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Abstract. Prenatal diagnosis has become both a requirement and right for women, regardless of its medical practicalities or reasons. It is frequently observed that women choose to undergo prenatal checks, actively participating in the reproduction of their society's culture and thus often considering themselves feminists i.e., women who have taken their lives into their own hands. According to some observers, this process has now been taken to excess and that from its use as a valuable cognitive procedure, prenatal diagnosis has rapidly developed into a tool for prenatal selection. A qualitative judgement is always given, starting from the 'vision' of a phantom created using technical instruments. It is as if the outcome of an examination assigns a higher or lower value to the life of an unborn child, with consequent psychological repercussions given the symbolic power of reproductive technologies used on women. The opportunities offered by diagnosis may generate new desires but also lead to new fears and a drive for measures aimed to avert them, resulting in physical pathologies and social imbalance.

Key words: bioethics, prenatal diagnostics, pregnancy

Introduction

The bioethical aspects of prenatal diagnosis, a set of techniques and methods that make it possible to detect anomalies and malformations in the embryo and fetus, can be divided into three different areas (1). The first is the private sphere, which considers the ethical issues concerning the couple and the fetus as a patient (2). The second is the professional sphere, which deals with the moral issues faced by those who perform prenatal diagnostic tests, usually doctors or laboratory technicians (3). The third and final sphere is the social one, dealing with public ethical issues related to social and health policy measures. However, these three areas should not be considered as separate entities since they are closely linked and often interact, though occasionally in conflict. Indeed, bioethics derives from (and flourishes as a result of) precisely this form of continuous confrontation (4).

As far as the private sphere is concerned, prenatal diagnosis has a profound existential significance for the woman and the couple themselves, as the technique impacts directly upon the experience of pregnancy (5, 6). In some cases, it has been found that seeing the image of one's child on an ultrasound machine screen consolidates a couple's relationship. In others, seeing the normality of one's child reassures the anxious minds of even the most apprehensive parents (7). On the contrary, it may also be the case that the ultrasound scan is perceived negatively by the couple as an intrusion into their most intimate private sphere (8). Indeed, a ghostly image of the unborn child and its visualization on a screen may provoke ambivalent feelings. On the one hand, there is the risk that the mother may see the fetus as something foreign to her; on the other hand, she may feel a pleasant sensation at seeing its movements for the first time, as if this has allowed her to become acquainted with it beforehand.

As previously noted, an ultrasound scan conditions the image which the mother-to-be and father have of their unborn child (9). Hence, one of the principal criticisms levelled against prenatal diagnostic methods is that of fostering expectations of a perfect child (10). A degeneracy which is perhaps more

realistic is the so-called 'medicine of wishful thinking'. Prenatal diagnosis has made it increasingly possible to control facets of pregnancy but can unfortunately also be misused (11); opening a window on a world that remained inaccessible for millennia has contributed to a fulling of desires that were once unthinkable. It is one thing to want a healthy child, which is an entirely legitimate aspiration, but quite another to desire a tailor-made baby which panders to the parents' preferences.

While prenatal diagnosis in its clinical form is fundamentally aimed at assessing the health and development of the fetus, there exists a distinct and ethically contentious domain often referred to as the pursuit of 'tailor-made' babies. This latter concept extends beyond the medical imperative of identifying and managing health issues and ventures into the realm of selecting specific non-medical traits.

Clinical prenatal diagnosis encompasses procedures like ultrasound scans, blood tests, amniocentesis, and chorionic villus sampling, which are primarily intended to screen for congenital anomalies, genetic disorders, and other health-related issues in the fetus. The primary objective of these procedures is to inform expectant parents about the health of their unborn child and to prepare them for any necessary medical interventions or decisions.

In contrast, the concept of a 'tailor-made' baby emerges from a different desire: the selection of specific traits such as gender, physical attributes, or even certain genetic characteristics. This drift towards a form of selective reproduction is facilitated by advanced reproductive technologies and genetic testing but is mired in ethical debates. Unlike clinical prenatal diagnosis, which is driven by the need to ensure fetal health, the selection of traits is often guided by personal or societal preferences and raises profound questions about the nature of parental choice, the definition of a 'desirable' child, and potential long-term societal impacts.

The ethical distinction between these two approaches is significant. Clinical prenatal diagnosis, while not without its own ethical complexities, is generally accepted as a part of responsible prenatal care. However, the idea of designing a baby to fit specific parental or societal preferences ventures into a controversial ethical territory. It poses risks of eugenics, inequality, and the commodification of human life. Therefore, it is crucial to maintain a clear boundary between using prenatal diagnosis for medical purposes and the speculative realm of creating 'tailor-made' babies.

This distinction necessitates a thoughtful exploration, as it reflects broader issues about the role of technology in reproduction, the scope of parental autonomy, and the societal values placed on human life and diversity.

Thanks to technology, there has been a revolution since the early years when technological control over pregnancy began to be adopted (12). Today, through prenatal diagnosis it is possible to act directly *in utero*. While in some ways this is positive, there is always the risk of lapsing into a selection of non-pathological characteristics.

Initially, the search for chromosomal aberrations in the fetus was limited to women who became pregnant after the age of thirty-five (13). However, there has been a change from fears of a malformed child to the demand for a quality child; from the right to counselling and consideration came the pregnancy-to-order experience (14). The application of classic prenatal diagnosis, which has become routine in non-technologically induced pregnancies, has long worked in the direction of particular forms of eugenics. Every woman expecting a child today is now faced with changes in contexts and their related images using prenatal diagnostic techniques. For this generation of women, a new obligation has emerged: making a decision on the birth of their child on the basis of prenatal examinations results.

Questions also arise as to whether these examinations are necessary from a clinical point of view (15). Can practices such as amniocentesis be considered medical procedures? (16). They are performed as a precautionary measure, to be on the safe side and to act quickly. However, their most significant side effect is the possibility that should malformations or hereditary diseases be found, these outcomes will lead to the woman's decision to terminate the pregnancy (17, 18). In order to read the effects of prenatal diagnosis correctly, it is necessary to distinguish between the two practices, the diagnostic act itself and

the potential act of abortion that may result from it. Indeed, it is incorrect to equate prenatal diagnosis with selective abortion (19-22).

The assertion that prenatal diagnosis should not be simplistically equated with selective abortion warrants a more nuanced exploration, as it touches upon complex ethical, medical, and societal dimensions. Prenatal diagnosis, at its core, is a medical practice aimed at assessing the health of the fetus. It provides critical information about potential genetic disorders, congenital anomalies, or developmental issues. This knowledge enables healthcare providers and expectant parents to prepare for the medical needs of the child, including interventions that might be required immediately after birth or even in utero.

Selective abortion, on the other hand, is a decision made following prenatal diagnosis, often in response to the detection of anomalies or genetic conditions. While it is one potential outcome of prenatal diagnosis, equating the two oversimplifies and misrepresents the purpose and the range of outcomes associated with prenatal screening. Many parents proceed with prenatal diagnosis with no intention of considering abortion but rather to prepare themselves for the birth of a child with special needs, or to inform themselves about the health of their developing baby.

Moreover, the decision to terminate a pregnancy following a prenatal diagnosis is a deeply personal and often a complex one, influenced by a myriad of factors including medical advice, personal beliefs, the severity of the detected condition, and the family's capacity to care for a child with special needs. To reduce this decision to a direct consequence of prenatal diagnosis is to overlook the emotional and ethical deliberations involved.

In addition, advancements in prenatal care and treatment options mean that a diagnosis of a fetal anomaly or genetic disorder does not necessarily equate to a poor quality of life for the child. There are many instances where prenatal diagnosis has led to early interventions that significantly improve the health outcomes for the child.

Thus, while selective abortion can be an outcome of prenatal diagnosis, it is just one aspect of a complex set of decisions and outcomes. Prenatal diagnosis serves a broader purpose of enhancing prenatal care and preparing parents and healthcare providers for a range of possibilities. To equate it solely with selective abortion is to ignore its multifaceted role in modern obstetrics and the diverse outcomes it can lead to.

We must not forget its numerous merits: it allows healthy children to be born to sick parents who would not have given birth without the necessary reassurance and it also makes it possible to already treat certain illnesses *in utero* or to prepare in advance pediatric treatment that the diagnosis has shown will be necessary. However, why are these two procedures so often equated? (21, 23-25). It appears to stem from the fact that in the early days, when prenatal diagnosis first took hold, machinery was not as advanced as today and therefore the techniques used only made it possible to recognize severe pathologies for which no intervention was possible. For this reason, it was perhaps easier previously to opt for abortion compared to today. Nowadays, a request for abortion may be made because of the lack of therapeutic alternatives, not because of the discovery of abnormalities in the fetus.

In contemporary prenatal care, decisions regarding abortion are increasingly complex and are not merely based on the detection of fetal abnormalities. Often, these decisions are influenced by the availability, or lack thereof, of therapeutic options for diagnosed conditions. For instance, certain genetic disorders detected via prenatal diagnosis may have no known cure or effective treatment post-birth, placing parents in a challenging position where abortion might seem like the only feasible option (26).

Take, for example, the case of Tay-Sachs disease, a genetic disorder for which there is currently no cure. Prenatal diagnosis can identify this condition in the fetus, but the lack of treatment options for Tay-Sachs often leaves parents facing the heart-wrenching decision of whether to continue the pregnancy (27). Another example is Trisomy 18, also known as Edwards syndrome. While some babies with Trisomy 18 are born alive, the severity of medical problems is significant, and the life expectancy is generally very short. This reality may influence the decision-making process of parents following a prenatal diagnosis (28). Furthermore, advancements in prenatal screening techniques, such as non-invasive prenatal testing (NIPT), have made it easier to detect chromosomal abnormalities

with high accuracy. However, the ease of obtaining this information does not necessarily translate to an increase in therapeutic options, leaving some parents with decisions that are more ethical than medical (29).

In all these scenarios, the decision to request an abortion is not driven solely by the discovery of an abnormality but is heavily influenced by the broader context of available medical interventions and the expected quality of life for the child. These decisions are multifaceted and deeply personal, reflecting a nuanced interplay between medical information, ethical considerations, and personal values.

In the context of prenatal diagnosis, it is crucial to distinguish between the use of technology as a necessary medical tool and its potential misuse as a means of genetic selection. On one hand, prenatal diagnostic techniques, such as amniocentesis or ultrasound, are vital tools for assessing fetal health and identifying potential pathologies that may require early intervention or specific postnatal care. This usage falls within the realm of preventive medicine, where the goal is to safeguard both the mother's and the child's health (30).

On the other hand, with the advancement of technology and increasing understanding of genetics, there is a risk that such practices may extend beyond disease prevention and treatment to become tools for selecting non-pathological characteristics, such as sex or other genetic traits. This shift towards selectionism raises significant ethical concerns, as it moves away from the primary goal of medicine, which is to treat and prevent diseases, and ventures into a territory where choices may be driven by personal and social preferences rather than medical necessity (31).

The history of prenatal diagnosis is a journey of medical and technological evolution, beginning with rudimentary methods and evolving into the sophisticated techniques we see today. The earliest forms of prenatal assessment were primarily observational, relying on a mother's description of fetal movement and basic palpation techniques by the physician. The development of the stethoscope in the early 19th century allowed for the first auditory monitoring of the fetal heartbeat, providing a rudimentary but significant insight into fetal health.

However, it was not until the mid-20th century that prenatal diagnosis began to take its modern form. The introduction of ultrasound in the 1950s revolutionized prenatal care, offering a non-invasive window into the womb. This technology allowed for the visual assessment of fetal development, identification of multiple pregnancies, and detection of certain complications.

The 1960s and 1970s saw further advancements with the development of amniocentesis and chorionic villus sampling (CVS), providing means for genetic testing and the diagnosis of chromosomal abnormalities. These techniques marked a significant shift from mere observation to active diagnosis, enabling early detection of conditions like Down syndrome and other genetic disorders.

In the late 20th and early 21st centuries, the field of prenatal diagnosis continued to expand with the advent of more sophisticated imaging technologies and the introduction of molecular genetic testing. Techniques such as detailed ultrasound scans, fetal echocardiography (32), and DNA-based tests have greatly enhanced our ability to detect a wide range of fetal anomalies and genetic conditions, sometimes as early as the first trimester (33).

This historical progression from basic observation to advanced diagnostic capability reflects a broader narrative of medical advancement. However, it also underscores the need for continuous ethical vigilance as we navigate the complex implications of these technologies for expectant parents, healthcare professionals, and society at large.

Conclusion

The history of prenatal diagnosis reveals how these practices have evolved from simple detection methods to sophisticated procedures that can profoundly influence reproductive decisions. From early ultrasounds and screening tests to modern genetic diagnosis techniques, the history of prenatal diagnosis is marked by continual technological advancement (34). However, it is essential that the evolution of these techniques be accompanied by ongoing ethical reflection, to ensure they remain tools in service of health and well-being, rather than means to pursue ideals of perfection or personal preferences (35).

Prenatal diagnosis has contributed to establishing the concept of the fetus as a patient (22, 36-38), which has led to a redefinition of the roles of all those who have to deal with it: mother, doctor and society. Since health is the fetus' right, as for all patients, any therapeutic intervention can be justified provided that it guarantees a fair balance between risks and benefits.

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