

Ethical Paradox of Down Syndrome Screening: Navigating Reproductive Autonomy and Inclusive Ideals

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ABSTRACT

In an era increasingly defined by the pursuit of equality and “inclusive design,” a significant ethical tension has emerged between the societal integration of the differently abled and the technological advancement of prenatal screening. Down Syndrome, primarily caused by Trisomy 21, remains a focal point of this debate due to its high prevalence and the significant socioeconomic impact on families and public health systems. While modern screening methods ranging from traditional serum markers and ultrasound to high-precision Non-Invasive Prenatal Testing offer pregnant women expanded reproductive options, they simultaneously raise concerns regarding contemporary eugenics. Approximately 92% of women who receive a definitive prenatal diagnosis of DS choose selective termination, a trend that has led to a marked decrease in DS birth rates despite advancing maternal age. This study examines the ethical complexities of PS, highlighting the discrepancy between the theoretical goal of reproductive autonomy and the practical reality of inadequate clinical counselling, biased information delivery, and the lack of prenatal therapeutic interventions. Ultimately, the study suggests that while the world moves toward the linguistic and social acceptance of “differently abled” individuals, the systematic use of screening to eliminate these same individuals necessitates a critical re-evaluation of public health priorities and the provision of non-directive, expert-led counselling.

Keywords: Down Syndrome (Trisomy 21); Prenatal Screening (PS); Non-Invasive Prenatal Testing (NIPT); Bioethics; Reproductive Autonomy; Disability Rights/Differently Abled

Abbreviations: WHO: World Health Organisation; PS: Prenatal Screening; DS: Down Syndrome; hCG: Total Human Chorionic Gonadotrophin; AFP: Alpha-Fetoprotein; uE3: Unconjugated Oestriol; CVS: Chorionic Villus Sampling; NIPT: Non-Invasive Prenatal Testing

Introduction

Diversity and Inclusion, from Disabled to the Differently Abled

Today's world is very different from the one we lived in less than a century ago. While numerous technological breakthroughs have been made, significant advances have also been made in the pursuit of equality, diversity, and inclusion. The global environment values

equality and encourages inclusion in all fields. Inclusion and diversity have different definitions but share some similarities. Inclusion promotes participation and appreciation of diversity by integrating and leveraging it into daily work processes. In contrast, diversity is defined as the heterogeneous makeup of organizational groups [1] Inclusive design, design for all, or universal design emerged as an essential component of a more recent international trend toward mainstreaming the integration of older and disabled people into society.

The concept ensures that the products and services meet the needs of the broadest possible audience, regardless of age or ability [2]. Language has long been regarded as more than just a means of conveying ideas between people. In some cultures, language has been used to shape people's behaviours.

Naming as a component of language has also been the subject of heated debate, particularly in the context of identity formation. The process of naming produces a subject whose sense of self is linked to society's definition. Individuals are thus recruited into identifying with labels and identities created by society rather than by themselves [3]. The UK government has published on inclusive communication, emphasizing words to use and words to avoid when writing about disability [3,4]. Concluded that the terms disability and disabled are divisive, stigmatizing, and anti-transformational. It represents flaws, incapacity, and inferiority. Therefore, the term differently abled appears to have more empowering connotations than disability [3]. The term "disabled" has been replaced by "differently-abled," and efforts are being made in every field to be more inclusive of the differently abled. However, do the same acceptance and standards prevail in welcoming the differently abled to the world/ society?

Screening, Prenatal Screening, and Ethical Concerns in Screening

In 1951, the United States Commission on Chronic Illness defined screening as "the presumed diagnosis of an unrecognized disease or defect by the use of tests, examinations, or other fast administered procedures." Screening tests separate healthy individuals who are likely to have an illness from those who are not, and the screening tests are not diagnostic [5]. In the latter half of the 20th century, screening tests for chronic medical conditions began to be widely available [6]. The era of modern screening began in 1968, with a pioneering publication by Wilson and Jungner for the World Health Organisation (WHO), [7,8]. Even though screening marks an advancement of medical technology, ethical issues that need consideration also accompany it. Before any screening test, it should be assumed that an abnormality will be found and that individuals with the disease will benefit from further care or treatment. Wilson and Jungner's report to the WHO, which listed 10 screening principles and circumstances in which screening can be judged morally appropriate, further clarified this. Since then, these principles have been widely applied to evaluate and conduct screening program [9].

There are a number of significant characteristics to consider while evaluating screening tests. First, the test must be safe, well described, and acceptable to patients. Then test characteristics, such as specificity and sensitivity, must be considered [10]. The best screening tests should be sufficiently accurate to rule out false-positive results while being sufficiently sensitive to detect the disease. Patients undergoing screenings should be able and willing to undergo additional diagnostic procedures as well as a possible treatment for the diseases they are being examined for [10]. A screening test should be affordable in

terms of disease detection and treatment [10]. In terms of ethics, testing, treatment, and medical care should be equally available to the screened population. In the hope that early intervention may improve results, screening tests are performed to identify the disease before the onset of symptoms. Additional illnesses suitable for screening should be clinically important, clearly defined, and have a major influence on public health [11].

For an illness to be worthy of screening, it must not only have an adverse effect on life expectancy or quality but also have a latent stage during which discovery and treatment are still possible. Contemporary prenatal screening primarily targets aneuploidy (chromosomal rearrangements), structural abnormalities, and disease-causing genetic anomalies. Ultrasonography or ultrasound in conjunction with maternal serum testing is used to check for structural abnormalities [11]. Risk assessment based on family history, maternal serum testing, and molecular genotyping are used to screen for disease-causing genetic disorders. A combination of age-based risk evaluations, maternal blood testing, and ultrasound scans is used to identify aneuploidy or, the presence of extra or missing chromosomes [10-12]. To examine various facets of prenatal screening (PS) tests 50 years after Wilson and Jungner initially published their principles of screening, a modern set of principles are employed as a model [6].

Down Syndrome and Screening

Down syndrome (DS) is the most common chromosomal rearrangement in humans, and it is caused by an extra copy of chromosome 21 or trisomy 21 95% of the time, it is a global health issue because of its high prevalence and poor quality of life. The reported prevalence ranges from 2-3 per 1000 live births in low- and middle-income countries to 1.2 per 1000 live births in high-income countries [13]. The most critical risk factor for trisomy 21 is maternal age. A woman's likelihood of having a child with DS is 1/1,300 at the age 25; at age 35, the risk rises to 1/365. The probability of having a child with DS climbs to 1 in 30 at age of 45 [14]. DS can cause spontaneous abortion or perinatal mortality. Surviving infants frequently have mild to moderate intellectual disability, delayed growth, and poor quality of life. Although they have a higher mortality rate than the general population, their survival rate has recently increased dramatically [15].

The life expectancy of people with DS has steadily increased over the last 60 years, rising from 12 in 1947 to 55 in 2016 due to timely surgical interventions for significant anomalies [16-17]. The lifetime cost of having a child with DS ranges from \$400,000 to \$800,000 [18]. A preliminary study on the economic burden of DS in China concludes that the economic impact of DS is significant for both the family and society. Appropriate management and prevention of DS are required to alleviate the severity of the burden carried by individuals with DS and their families [19]. According to the Family System Theory, every problem within one family subunit affects all other subunits. The presence of a child with DS influences the lives of other family members, their relationships, and all family functions. A qualitative study

conducted in Turkey of mothers having a child with DS demonstrates that these families face considerable social, physical, economic, and emotional challenges, as in other countries. Children encounter difficulties in their daily lives and the future [20].

Although DS can significantly impact both length and quality of life, people with DS syndrome can be academically successful, accomplish different academic achievements, live independently, engage in decent employment, and be active members of society [21]. In 1989, a triple test for second-trimester serum screening for Down's syndrome was introduced in the UK (total human chorionic gonadotrophin [hCG] with maternal age, alpha-fetoprotein [AFP], and unconjugated Estriol [uE3]) [22]. First-trimester screening with nuchal translucency measurement and maternal age was implemented in 1993. Since then, serum and ultrasound screening for DS has become more common, both in the UK and elsewhere [22]. Pregnant women worldwide already have a wide selection of prenatal tests for DS. Triple screen, quadruple screen, first-trimester combined screen, step-wise sequential screens, and fully integrative screens are used to determine the statistical odds that their foetuses have DS, with varying degrees of detection [23].

Maternal serum marker screening and ultrasound imaging (ultrasonography) to detect chromosome rearrangements and other deformities are routine in the first and/ or third trimester of pregnancy in many countries. However, both methods have high false-positive rates, ranging from 2% to 7% [24]. If these tests indicate that a foetus is at an increased risk of aneuploidy (chromosomal rearrangement), invasive diagnostic procedures such as chorionic villus sampling (CVS) or amniocentesis are advised. CVS is typically performed between 10 and 13 weeks of pregnancy, whereas amniocentesis is performed after 15 weeks of pregnancy. Due to the physical discomfort and the associated 1-2% risk of procedure-induced miscarriage, many women are uncomfortable with invasive testing [25]. Recently, the introduction of a new prenatal test has rapidly altered the prenatal testing approach. Non-invasive prenatal testing (NIPT) provides a step between serum screening and invasive diagnostic testing. NIPT entails analysing the cell-free foetal DNA present in a maternal blood sample to determine the likelihood of a foetal chromosomal rearrangement [26]. NIPT is more precise than serum screening and generates fewer false-positive results, but it is not diagnostic [27].

When considering PS, the question of how the purpose of such screening should be defined arises. Why would PS be available for disorders such as DS given that a positive diagnosis would result in just two options: abortion or continuation of the pregnancy? The typical objective of population screening (health benefits gained through timely treatment or prevention) is not readily applicable to this sort of PS (although it does apply to PS for conditions such as infectious diseases or Rhesus status) [28]. Prenatal screening for fetal abnormalities is typically understood to provide reproductive options to pregnant women (and their partners) [28]. Currently, there are no

prenatal therapeutic interventions for DS; therefore, pregnant women seek prenatal identification for one of three primary reasons. The first reason is that they want to terminate their pregnancy if the fetus has DS [29]. The second reason is that they want to learn more about DS before the birth of a child they intend to raise, or they want to start looking into adoption options. According to an international meta-analysis of data from the United States, the UK, France, New Zealand, and Singapore, roughly 92% of women who receive a definitive prenatal diagnosis of DS choose to terminate their pregnancies [29].

Women worldwide delay childbirth. The birth incidence of DS was expected to rise because advanced maternal age is linked to DS. Worldwide, DS births have decreased by 2%–18% per year [30]. Without prenatal testing, the number of babies born with DS in the United States would have increased by 34% between 1989 and 2005 [31,32]. There were 15% fewer births, a difference of 49% between the predicted and observed rates. Without prenatal testing, there would have been a 58% increase in babies born with DS in the United Kingdom between 1989 and 2006. Instead, there was only a 4% increase, which was 54% less than anticipated. These trends are mainly attributable to the availability of prenatal testing and the maternal preference for selective terminations in the United States, the United Kingdom, and internationally, [30]. In China, 55.6% - 100% of pregnancies in which DS was identified during pregnancy were subsequently terminated [13]. Will DS gradually disappear with the advent of prenatal testing?

Is Prenatal Screening a form of Contemporary Eugenics?

Eugenics, popularized in the early 20th century, the scientifically inaccurate and immoral theory of “racial improvement” and “planned breeding.” Eugenists believed that humans could be perfected through genetics and inheritance and eliminate so-called societal issues. They believed that involuntary sterilization, segregation, and social exclusion would eliminate unfit individuals from society [33]. Despite the common perception that eugenic practices are applications of Darwinism, they may be better described as meta-Darwinism. In a strict sense, natural selection implies that selection is natural and that those who survive are fit. In stark contrast, eugenics requires human control to be substituted for natural selection. Survival in an environment based on eugenics is artificially manipulated based on a judgement or consensus regarding what constitutes fitness [34]. Some French philosophers, biologists and health professionals have condemned DS screening in particular, attributing it to a new type of eugenic, sparking the ethical discussion. Once it is determined that a foetus has DS, the only treatment available is abortion, which most women choose [34].

It has been argued that society as a whole is attempting to eradicate malformed foetuses to improve the quality of the human race [35,36]. Thomas and Rothman argued that prenatal screening for DS (specifically NIPT) is a form of modern eugenics because it eliminates, devalues, and possibly prevents the birth of individual with the

condition [36]. Considering the complex links between eugenics and public health, PS appears to be predicated on the implicit idea that people with impairments cannot have a fulfilling life, are not wanted in society, and that their parents will desire to prevent their birth. The notion that there are significant public health benefits if people with disabilities are never born is strengthened by tightening the relationship, and persistent incorrect beliefs regarding the social and monetary “costs” of disability maintain pronatalist eugenics in the twenty-first century [37]. The delivery and developments of healthy children and promotion of reproductive freedom and choice for women should be fundamental objectives of public health initiatives. However, a decreased birth predominance of an illness for which PS links public health programmes to eugenics is not an appropriate evaluation of health improvement or choice [37].

What choices do the mothers have regarding, reproductive autonomy, and what is the basis for that?

Public health is subject to ongoing debates on limited versus broad conceptions of health, public versus individual goods, and efficiency versus equity. This question (or a similar formulation) is arguably the most important ethical question surrounding proposed public health interventions. It is a facet of the considerable tension between collective and individual goals. By their very nature, public health interventions require the “coordination” of, or some degree of “intrusion” into social life and social practices. This raises concerns regarding individual liberty. Some interventions are straightforward and immediately forceful [38,39], argued that the individual is constantly under intense pressure to submit to the examination because the institution of health care or society took the initiative to do so rather than the individual. It is extremely questionable to organize prenatal diagnosis as screening programmes based on the claim that reproductive autonomy is a key goal of the practice. This is due to the same factor that makes screening programmes difficult from an autonomy standpoint [39].

Mothers from the United States, Spain, and the Netherlands who received a prenatal diagnosis of DS and chose to continue their pregnancies reported that their physicians frequently provided incomplete, inaccurate, and occasionally offensive information about the disorder (Skotko [30]). Mothers in the Netherlands who terminated their pregnancies after receiving a prenatal diagnosis of DS primarily based their decisions on the belief that DS was “a severe abnormality” and a “burden” that was “too heavy” for the child [40]. Consequently, some have questioned whether mothers make informed clinical decisions regarding their pregnancies [41]. Medical schools do not prioritize delivering clinical training regarding intellectual disabilities. Postgraduate trainees in Obstetrics and Gynecology in the United States largely accepted that their training was inadequate to counsel a pregnant mother who’s prenatal screening indicates a high risk for DS. The vast majority of studies imply that physicians of today and tomorrow are not appropriately equipped [42].

Therefore, the question arises, “Are the physicians competent and trained enough to give accurate information on DS for the parents to make a decision?” [42,43]. The next question of debate is whether physicians or genetic counsellors infuse their own opinions in counselling. Academic societies around the world adhere to non-directive counselling providing expectant parents with unbiased information so they can make educated decisions within the context of their own views and values [30]. However, is this true in, actual practice? A study conducted on 499 physicians and 1084 genetic professionals from the United States who presented a prenatal diagnosis of DS to expecting couples revealed that overemphasis on positive and negative elements indicates that not all pregnant women receive impartial information from their healthcare providers [30].

An essential ethical circumstance for the introduction of prenatal testing is that women should be able to accept and make decisions based on their own values [44]. As no treatment is available for the defects detected by the test, women who are offered the test are believed to be confronted with ethical questions regarding the value of a disabled life and parental responsibilities regarding a fetus with a defect. Consequently, the ethical beliefs of women likely play a significant role in the decision regarding prenatal testing [45]. DS is a varied condition, and the prognosis for individuals with the disorder is still uncertain even though some symptoms are common. This means that if a pregnant woman obtains a prenatal diagnosis of DS, she is unlikely to receive information regarding the child’s physical and cognitive impairment severity. Even the most accurate diagnostic tests for disorders such as DS can only inform pregnant women whether the fetus has a specific chromosomal marker. Women are not informed of the severity or breadth of possible abnormalities; a fetus diagnosed with DS may not survive pregnancy, although a child born with the disease may graduate from college [36].

Conclusion

Changing the term “disabled” to “differently abled” and altering designs to be more inclusive of people with varying abilities indicate that the world is moving toward acceptance of diversity and inclusion in every aspect. Ironically, however, the same technologically advanced world is attempting to eliminate the differently abled. Screening for diseases for early diagnosis and intervention has been a practice since the 19th century. Ethical screening principles are vital, and the first set of ethical screening principles appeared in the early 20th century. Initially, these principles focused primarily on non-communicable diseases that posed a threat to public health and were gradually adapted to fit the overarching prenatal Screening (PS). Children born with disabilities pose a significant public health risk and an economic burden. DS poses a significant economic impact on society and the family, as well as social, physical, and emotional challenges to families with a child with DS.

The concept of PS was established in the 1950s. PS tests can detect preterm birth, congenital anomalies, and chromosomal rearrangements in fetuses. With the development of technology, less invasive and more precise PS techniques have emerged. Owing to ethical concerns and the expense of screening with advanced and more accurate methods, PS is only accepted and offered in development countries and some developing nations. In screening for chromosomal rearrangements such as trisomy 21 or DS, no therapeutic outcome is offered in the event of a positive result, contrary to the principle that a disease or condition can only be screened if a therapeutic outcome is available and offered. The effects of DS can include severe congenital malformations and a shorter life expectancy, but technological advances in surgery for children with DS have increased life expectancy. In addition to advancements in education for children with special needs and the availability of opportunities, children can now live into adulthood and beyond and lead a nearly normal life.

Maternal autonomy in decision-making, the availability of non-biased information leading to decision-making, and respect and courtesy for any decision made by the mother or the parents, are, given less consideration but are crucial to the discussion. Every screening test is not 100% accurate and has a small margin of error. Therefore, the potential for false-positive results and the subsequent termination of relatively healthy fetuses poses a significant ethical concern. Behind the concept of PS lies the question of eugenics, which raises a severe ethical consideration regarding the potential elimination of individual with disabilities from society. Countries that have accepted prenatal screening as routine in antenatal care do not openly and extensively discuss this topic. Due to the serious ethical issues involved in DS screening, screening should only be offered to populations at high risk. It is also essential to share unbiased, non-directive information at the point of decision-making and ensure that information providers are equipped with the necessary knowledge, training, and mannerism of delivery.

Consent for Publication

We certify that this manuscript has not been published elsewhere and submitted to another journal.

Competing Interests

The authors declare that they have no competing interests.

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