

Genetic Screenings as Public Policy: Benefits, Limitations, Risks, & Public Perception

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Abstract

Genetic screenings mark a quantum leap forward in diagnostic capability and could be a powerful boon to public-health informatics. However, the integration of genetic screenings into public-policy frameworks requires a careful consideration of benefits and risks. A growing body of literature concerning the ethical implications of genetic screenings describes their potential to cause psychological sequelae, encourage unnecessary prophylaxes, and foster a climate that is implicitly intolerant of genetic diversity. The public's perception of, and responses to these risks are shaped by historical instances of genetic discrimination, personal experience, and socio-cultural values. As genetic screenings grow increasingly powerful and affordable—and thus more alluring as a facet of public-health policy—understanding why the public is wary of genetic screenings will help bioethicists at the public-policy vanguard hedge against harm and discrimination while protecting genetic diversity.

Key words: bioethics, genetic screenings, genetic sequencing, biotechnology

1. Introduction

Genetic screenings are an extraordinary achievement of medical technology. Today, healthcare providers wield the ability to rapidly scan a patient's genome in search of single-gene Mendelian disorders such as Duchenne Muscular Dystrophy [1]. Providers can also use carrier tests to determine whether parents might pass a genetic disorder to their progenies and predictive tests to assess patients' likelihood of developing late-onset disorders like breast cancer [2]. Genetic screenings preemptively detect thousands of disorders [1], allowing patients to begin treatment or make lifestyle changes while asymptomatic. Furthermore, genetic screenings bolster neonatal and family care by informing parents of their children's healthcare needs [3, 4]. As technology advances, these capabilities will flourish, and genetic sequencing will become a staple of healthcare [5].

Nevertheless, the ethics of genetic screenings demand scrutiny. Genetic screenings are linked to psychological sequelae [6], discrimination [7-9], and reduced genetic diversity [7]. As genetic screenings grow increasingly powerful and ubiquitous, healthcare providers must ensure sequencing respects patients' psychological well-being, sociocultural values, and genetic diversity.

2. Overview of Genetic Screenings

2.1. Definition

According to the Task Force on Genetic Testing [8], genetic screenings entail:

The analysis of human DNA, RNA, chromosomes, proteins, and certain metabolites in order to detect inherited disease-related genotypes, mutations, phenotypes, or karyotypes, for clinical purposes...[including] risk of disease, identifying carriers, establishing prenatal and clinical diagnosis or prognosis. Prenatal, newborn, and carrier screening, as well as testing in high-risk families, are included. (¶9)

2.2. Benefits

Genetic screenings are increasingly practical diagnostic and predictive tools [10]. Through screenings, healthcare providers are able to identify whether patients carry, or have a heightened risk of developing, various genetic disorders; estimate patients' likelihood of developing late-onset disorders [2]; and predict parents' likelihood of passing genetic disorders to their progenies [3]. Knowledge of genetic disorders empowers patients to plan for their futures and help relatives who may unknowingly be at risk [4].

2.3. Limitations

Benefits notwithstanding, genetic screenings have numerous limitations [7]. Holtzman and Shapiro [8], Caplan [7], and Chial [11] noted that carrier tests, which screen for Mendelian disorders, are powerful predictors of disease development but poor predictors of severity. Furthermore, carrier tests sometimes overlook Mendelian disorders. Of the estimated 1,800 protein-encoding human genes believed to cause Mendelian disorders, only about 300 have been identified [12]. In most cases, the known identities correspond to common Mendelian disorders, which attract more attention and funding [11]. Additional genetic factors can make detecting some Mendelian disorders difficult [8]. Cystic fibrosis, for example, can result from over 600 mutations, some of which elude screenings [8, 13].

The limitations are more pronounced in predictive tests, which cannot assess patients' risk of developing somatic diseases [9]. Since predictive tests are probabilistic and administered to asymptomatic patients, even the best screening cannot conclusively determine that a patient will develop a disorder [2, 7]. Hence, predictive screenings can encourage unnecessary prophylaxes like mastectomies [7]. This is not to diminish the value of predictive screenings; indeed, they regularly save lives.

2.4. Risks

The risks of genetic screening stem not only from their limitations, but also from the potential of their results to have far-reaching psychological and social impacts [4, 6]. While positive test results often ease patients' anxiety [3], they can cause psychological harm and prompt drastic

lifestyle changes [6, 7]. Moreover, positive tests can thrust a patient into the stressful position of delivering unwelcome news to relatives, potentially fostering anger and resentment [8]. In extraordinary cases, the results of a genetic screening can indicate parental consanguinity, in which case healthcare providers are faced with the ethical dilemma of whether the parents have a right not to know the results [14]. Even negative tests can be harmful, as they sometimes elicit survivor guilt [4]. These risks must be communicated unequivocally, and healthcare professionals must be capable of providing necessary counseling and follow-up support [7].

3. Public Perception of Genetic Screenings

Although public opinions concerning the medical benefits of genetic screenings and public understanding of genetics are favorable [15, 16], ethical concerns persist. A complex nexus of influences shapes public perception of genetic screenings. Among the most influential factors are historical instances of genetic discrimination [2]; skewed risk perception of unfamiliar technologies [16] and disorders [17]; sociocultural tropes regarding the concept of personhood [7, 8], which can pressure parents to terminate pregnancies when a screening detects a “dehumanizing” disorder [17]; and personal experience with people who express genetic disorders [17]. Understanding these influences would help policymakers tailor initiatives and communications that increase support for genetic screenings as public policy.

3.1. Historical Instances of Genetic Discrimination

Throughout America’s history, people with qualities deemed undesirable have been particularly vulnerable to discrimination. Western cultural values, Caplan [7] observed, hold cognitive function and individual autonomy as thresholds of personhood – those who lack these facets risk being deprived of fundamental human rights. Indeed, proponents of the expressivist objection contend that negative perceptions of disorders can render everything else about a person unimportant, thereby implicitly devaluing the disabled [17]. These attitudes have contributed to policy initiatives that are widely considered unethical [2]. In the 20th century, for instance, roughly 60,000 Americans were involuntarily sterilized under eugenics programs aimed at reducing chronic illness, cognitive deficiencies, and criminality [2]. In the 1990s several state judges attempted to limit reproduction amongst mothers convicted of child abuse by making them choose between jail and mandatory Norplant contraceptives [2]. Such programs and sentences are at odds with widely accepted views on reproductive rights, which hold that only couples—not governments or courts—can dictate the number and timing of their children [18].

Moreover, historic precedent suggests that discrimination inadvertently caused by genetic screenings could disproportionately affect minorities. Genetic screenings for sickle cell anemia in the 1970s disproportionately exposed African Americans to discrimination and coercion, since refusing a screening was sufficient grounds to bar a child from public school [2]. Concomitant with the 1990s’ Norplant sentencings, some observers suggested that Norplant could “reduce the underclass” [19] by preventing pregnancies among black welfare mothers. At best, this suggestion endorses reproductive constraints as a socioeconomic solution. At worst, it conjures dystopian images of Huxley’s Epsilons [20] or GATTACA’s undesirables [21].

This history of discrimination legitimizes concerns about the rise of genetic screenings as genomic science advances. If scientists eventually discover the genetic signatures associated with stigmatized disorders (e.g., antisocial personality disorder), relatives, employers, and governments may treat those persons differently. A 2013 study by Haga and colleagues [15] found that even college-educated Americans who firmly understand the benefits of genetic screenings are troubled by the prospect of genetic discrimination. Of 300 people surveyed (65 percent of whom were college graduates), 51.3 percent of respondents believed that genetic screenings would influence one's ability to obtain health insurance, and 16 percent believed that genetic screenings could affect one's ability to find employment [15]. A 2012 study by Henneman and colleagues [16] comparing Dutch biotech perceptions in 2002 and 2010 found that respondents understood the benefits of genetic screenings, yet concerns of their potential to dichotomize society had increased during the study's 8-year span. Even if such concerns are unfounded, their persistence dampens public enthusiasm for genetic screening initiatives.

3.2. Risk Perception

The way humans measure and perceive risk could slow the implementation of genetic screenings as public policy. Scientists and non-scientists have different definitions of risk, and effective communication between experts and the public is necessary to bridge the divide between perceived and actual risk [16]. This disparity often results in public perception of high risk where it is actually low (e.g., nuclear-radiation poisoning) and low risk where it is actually high (e.g., operating a motor vehicle) [16]. The disparity is driven, in part, by familiarity – few people are versed in the nuances of nuclear radiation, while most are familiar with automobiles [16]. Since genomics is a new and arcane field, the public likely perceives inflated risks. In doctrinally risk-averse sectors like the life-insurance industry, the perfect genome myth (i.e., the belief in a quintessential, defect-free human genome) encourages policies that are hostile toward genetic indicators of disease [9], even though predictive tests cannot definitively forecast a disorder's development and no test can predict a disorder's severity [8].

3.3. Sociocultural Stigmas & Genetic Diversity

Sociocultural stigmas concerning genetic screenings discourage public adoption of sequencing policies [7, 8]. If easily identifiable disorders also carry social stigmas, patients may be reluctant to undergo testing that could foment social backlash. Furthermore, sociocultural norms may stifle genetic diversity by encouraging abortions based on a society's concept of personhood, as Billings and colleagues [9] predicted. Caplan [7] confirmed Billings' [9] prognostication decades later: In 2014, 9 of 10 positive prenatal Down's syndrome screenings in the U.S. and the UK led to abortions, and Down's had declined by 15 percent in the U.S. since 1989 despite forecasts that year of a 25 percent increase by 2014. In the UK, the expected increase in that 25-year span was 48 percent; the observed increase was 1 percent [7]. This trend seems antithetical to the United Nations Convention on the Rights of Persons with Disabilities, Article 18 [22], which implores signatory nations to respect the dignity of diverse people of all abilities. Nevertheless, Western cultural views on intellectual function and individual autonomy as facets of personhood have made abortion the overwhelming response to positive Down's screenings.

Among families in which a loved one has a genetic disorder, different phenomena emerge. In a study of 41 families and individuals living with Spinal Muscular Atrophy (SMA), Boardman [17] found that close experience with the disorder influences one of two mindsets. When the person who has SMA lives past childhood and enjoys a full life, Boardman's [17] interviewees (N = 41) tended to reject genetic screenings, which, they contended, implicitly devalued the lives of disabled persons and encouraged abortions based on a single trait without regard for deep, personal complexity (i.e., the expressivist objection). Conversely, an interviewee who lost two children to Type I SMA before their first birthdays saw genetic screenings as morally obligatory to prevent the suffering that his children experienced [17]. Experiential contact, then, appears to have a moderating effect on the decision to seek genetic screenings unless the disorder is expected to be so severe as to cause considerable suffering and infantile death [17]. This is noteworthy for two reasons: first, it is a testament to the contention that a disability does not devalue human life or make people less enriching or important to their families; second, it suggests that this moderating effect may be vanishing in countries like the U.S. and the UK, where the tendency to terminate pregnancies following positive genetic screenings for stigmatized disorders [7] makes experiential contact increasingly unlikely. Reconciling this paradox between the value of genetic diversity and some societies' pursuit of genetic uniformity must be regarded as an urgent bioethical concern.

Conclusions

As genetic screenings improve, ethical implications of public-screening policies must be scrutinized. Mandatory screenings would lead to unnecessary prophylaxes [7], cause serious psychological harm [6], and expose patients to discrimination [7-9]. Although some countries have passed laws against genetic discrimination (e.g., America's Genetic Information Nondiscrimination Act of 2008 [23]), recourse in discrimination cases requires self-advocacy [9]. Minorities who tend to disproportionately lack time and financial resources for self-advocacy could, therefore, be particularly incapable of seeking legal recourse for discrimination [9]. Other developed countries, for instance Canada, lack codified genetic-discrimination protections [24].

Managing risk perception through education and by cultivating a track record of nonmaleficence will be crucial to implementing screenings as public policy. Public risk perception rarely aligns with actual risk, and in the case of emerging technologies like genetic screenings the public probably overestimates risk [16]. Governments must work to ensure that public-relations initiatives do not overstate the safety of genetic screenings, since no genetic screening is free of risk.

As high-throughput and parallel-processing technologies improve, the volume of genetic information sequenced stands to exponentially increase [10, 25]. Bioethicists must be at the public-policy vanguard to mitigate the risks described herein. In an age of unprecedented genomic knowledge, communicating the benefits, limitations, and risks of genetic screenings to the public; safeguarding against discrimination; and protecting genetic diversity will be defining bioethical challenges of the times.

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