Ethical Implications of Genetic Testing and Selection

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Over the last century, technological advancements have revolutionized medical care and ushered in a new era of potential. One such area that has advanced dramatically in recent decades is the field of genetic sequencing and testing. Our understanding of the human genome, the impact of genetics on diseases, and the creation of gene editing technology known as CRISPR have pushed humans to the brink of being able to genetically identify those at high risk for diseases, improve family planning, and even select specific genetic traits for their children. As these technologies evolve, addressing the ethical implications and societal impact becomes increasingly important. My initial interest in this topic began in 1997 with the release of the movie *Gattaca*. This film portrays a not-too-distant future where society is composed of valids, humans genetically selected at conception by their parents, and invalids who were conceived naturally. This dichotomy creates a world where invalids are heavily discriminated against, and the plot centers around various ethical implications of genetically selective reproductive technologies.

Genetic testing involves examining a person's DNA and has become an essential tool for scientific research and a pivotal practice in medical care. It is increasingly used to diagnose diseases and identify genes linked to an increased likelihood of specific conditions. Typical uses of genetic testing today include testing embryos for genetic deficiencies such as trisomy 13, trisomy 18, and trisomy 21. Another common use is testing for women with a family history of breast cancer. Women who test positive for the BRCA gene are up to five times more likely to experience breast cancer than those without it. The knowledge from this testing brings on a series of difficult decisions for those it affects. Should a woman that tests positive for BRCA mutation decide to have children knowing there is a 50% chance she passes the gene to her offspring?

Should a woman pregnant with a fetus that tests positive for trisomy 21, also known as Down syndrome, use that information to elect abortion? Should a well-off couple use CRISPR technology to choose the eye color of their unborn child? The ethical implications and debates arising from genetics are complex and deeply intertwined with personal values and societal norms, challenging us to carefully weigh the potential benefits against our moral considerations and unintended consequences.

Audience

The intended audience for this paper encompasses a wide range of individuals and groups with varying interests in the fields of genetic testing and selection. First and foremost are prospective parents and families. Those dealing with hereditary genetic conditions will find exploring these topics highly relevant as they are directly impacted by the advancements and controversies surrounding genetic testing. Those intrigued by designer babies should be informed about the potential unintended consequences of genetically selecting traits for their unborn children. Legal professionals and policymakers must also consider the content of this paper, as the evolving nature of genetic science and societal norms necessitates the reevaluation of existing laws and the creation of new laws and regulations. Healthcare professionals, especially family planners, nurses, and OB-GYNs, play a critical role in reproductive health education and the potential application of genetic technologies. As genetic science becomes more prevalent, these professionals will be at the forefront of patient care. They must be well-informed of the latest developments and ethical implications as they educate patients and guide them through the complex landscape of genetic testing and selection concerning OB care. This paper serves as a resource for these professionals, providing an understanding of the many aspects of genetic testing and selection, its ethical implications, and the more significant debate surrounding these

issues. Lastly, whether driven by personal interest, hopes to eradicate hereditary conditions, or a desire to engineer future generations genetically, the general public will find this paper an accessible overview of the potential benefits and myriad ethical and societal pitfalls.

Cons of Genetic Testing and Selection

As Muys et al. (2019) noted, accurate genetic testing of a fetus has drawbacks. A common form of testing is chromosomal microarray analysis (CMA). CMA scans the entirety of the fetus' genome to identify the presence of copy number variants (CNVs). CMA has a high resolution of testing, meaning it can assess for the existence of aneuploidies and identify the genetic potential for late-onset diseases. This introduces an ethical and moral dilemma for physicians and patients alike. In prenatal testing, patients can terminate a pregnancy even if there is no certainty that the infant will be affected, such as in the case of the BRCA gene, or parents could decide to continue the pregnancy. Either choice can cause much stress, anxiety, and emotional distraught that can last a lifetime. If they continue with the pregnancy, they may spend untold years worrying about what may or may not happen later in their child's life. Similarly, suppose they choose to terminate the pregnancy. In that case, they may live with thoughts that they potentially terminated what could have ended up being a child that never developed cancer and lived a full and healthy life. If laws and regulations limit what physicians can share about genetic testing and late-onset disease risk, then there are concerns about paternalism and removing parental choice.

Informed consent becomes a concern when making decisions based on genetic testing. A study by Tucker and Christian (2022) focused on expectant mothers' reactions after disclosure of nonlethal fetal anomalies. Of note in this study is that women were often presented with difficult-to-understand genetic data and asked to make medical decisions based on

incomplete information. Even when this data revealed certainty that the fetus had an anomaly, mothers were still forced to weigh the various risks associated with medical intervention versus the risk of allowing natural processes to play. Furthermore, these women stated that others reacted negatively to information regarding the nonlethal fetal anomalies causing these women to feel as if they were stigmatized and that this information "was a stressful, negative, and distressing aspect of their pregnancy, a pregnancy forever changed and that the distress related to disclosure continued into postpartum" (Tucker and Christian, 2022, p.225).

In cases like these, intentional genetic selection may be an option in the near future. As noted by Hartman et al. (2018), gene editing in utero can more thoroughly alter the genomic sequence before adverse effects related to the targeted disease can ever take hold. This treatment could dramatically change the course of gene-related diseases and illnesses for humanity. However, this begets the question of how far is too far. Once technology advances to the point of editing genes in utero to improve health, it won't be long before an enterprising capitalistic company starts offering designer babies. This is already beginning to occur. A quick perusal of the internet can find multiple companies specializing in vitro fertilization (IVF) that also offer preimplantation genetic diagnosing (PGD) so that parents can select the embryo with the lowest chance of developing genetic disorders and increase the likelihood of having a child of a specific sex. A major caveat is the expense. Services like this are only available to the well-off. In a future where the rich can genetically select their children to be tall, good-looking, physically fit, and intelligent while the poor are stuck, having conventional children can further exacerbate the gulf between the proletariat and the bourgeoisie. Never mind that this opens the door for discrimination based on nothing more than genetic potential chosen by one's parents before birth.

Pros of Genetic Testing and Selection

While the concerns presented by Muys et al. (2019) highlight valid ethical considerations, there are undeniable benefits to genetic testing and selection that must be considered. The high resolution of CMA can be seen not just as a source of stress but as an empowering tool that offers prospective parents insights into their child's genetic profile. Knowledge about genetic risks regarding late-onset diseases or aneuploidies equips parents to make informed decisions regarding their family's future. These decisions can range from termination of the pregnancy to early intervention planning for specialized care.

Regarding informed consent, the argument that parents would be unable to understand the medical implication of genetic testing results is not just lazy but perhaps underestimates a parent's abilities to engage with such complex issues. The solution shouldn't lie in shunning these technologies but focus on thorough education and counseling to ensure that genetic screening is well understood. Far from endangering informed consent, these types of testing enhance autonomy by providing parents with what would otherwise be hidden information. As with any other procedure or medical information, parents can easily choose what information they wish to receive because of testing and what information they want not to be made aware of, thus allowing for culturally congruent care to fit their unique values and needs. Arguments that this is unfair to the medical professional forced to hold the secret fail to consider that these same medical professionals are expected to withhold treatment from patients at their request, even if the physician believes it is not the right choice. In utero genetic testing and screening can have a tremendous positive impact on public health. Israel instituted a genetic screening program for reproductive health in 2013. Subsequently, there was a 57% decrease in children diagnosed with spinal muscular atrophy (SMA) from 2014 through 2017 (Singer & Sagi-Dain, 2020).

The concept of genetic editing and selection, while a potential minefield of ethical dilemmas, offers promising medical interventions that could revolutionize healthcare. Gene editing to improve health is a massive step toward eradicating debilitating genetic diseases, creating a society with less suffering and a reduced medical burden which can dramatically reduce overall healthcare costs. Regarding designer babies, there are legitimate uses of trait selection. Gene selection can be used in careful family planning in families with one child and the wish to have a second child of the opposite sex. In a world where overpopulation is gaining traction as a concern, this could significantly reduce the number of children a couple needs to have to fulfill their family planning desires. Concerns of worsening the socio-economic divide can be easily resolved by creating responsible regulations and policies to guide their ethical application and instituting funding programs to aid those who can't afford it.

Summary and Reflection

There are many ethical concerns regarding genetic testing, the selection of the unborn child, and many opportunities for abuse and unethical uses. The potential benefit of such powerful technology is too significant to ignore. Consumers of healthcare should be empowered to make the best decisions for themselves and their loved ones, and the information and capabilities provided by technologies such as CRISPR and CMA do just that. Concerns that parents are ill-equipped to understand genetic data and make informed decisions are overly paternalistic and anathema to the principles of autonomy, informed consent, and personal agency, which are bedrock principles of ethical medical practice and patient-centered care.

Through researching and writing this paper, it has become apparent that the ability to enhance human life and minimize suffering is profound yet comes with inherent risks that must be managed. To mitigate those risks and enhance the benefits, we must begin to engage with this

topic more deeply on a societal level. Education is paramount for the general public, prospective parents, policymakers, and healthcare professionals. The author believes that an informed society can make responsible choices, create balanced regulation, and prevent the divide between genetic ‘haves’ and ‘have-nots.’ However, education and policymaking need to happen now to prevent capitalistic entities from turning genetic testing and selection into a for-profit industry and to protect healthcare consumers' medical rights and freedoms.

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