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Why Genetic Testing Should be Mandated

As the medical field continues to advance, genetic testing has become an increasingly important matter in many areas including disease management, family planning, and abortion. Genetic testing consists of analyzing one's genome for mutations that could potentially cause disease. By identifying abnormalities in chromosomes, DNA or proteins, individuals are more prepared to make decisions regarding their personal health care and future plans. Genetic testing is extremely beneficial and is very important in the future of medicine and public health. It provides information that can be useful for advancing treatments, creating management plans, and guiding family planning. Genetic testing should be mandated because of its ability to promote individual health in adults and children, and the major impact that it has on aiding in the push to create a healthier public.

What is genetic testing?

Genetic testing dates back to 1947 which was a focus on disability prevention. The focus then shifted to reproduction, followed by areas such as oncology and cardiology[1]. Genetic testing has always been present within the medical field, but the shift from disability to understanding the human genome on a more specific level makes outcome measures much more sophisticated. Genetic testing is used to detect abnormalities in DNA, chromosomes, and proteins that could lead to the development of a disorder.

There are multiple types of genetic tests which are used for different reasons. The most common types of tests are diagnostic and predictive tests. Diagnostic genetic testing is used to confirm a disorder in a symptomatic person[2]. This type of test provides a clear answer as to whether an individual has a genetic disorder. A predictive test is used for asymptomatic family members that are at risk for a genetic disorder due to familial genetic mutations. Within the category of predictive genetic tests, there are presymptomatic and predisposition tests. A presymptomatic test can determine if there is a genetic mutation present in an asymptomatic person, which will confirm that the individual will develop the disorder later in life[2]. Presymptomatic tests plays an important role in family planning because individuals who have a positive presymptomatic test result for a disorder such as Huntington's disease may decide to change their plans for the future to prevent the disease from being passed on. A predisposition test differs from a presymptomatic test because it determines if there is a genetic mutation present that increases one's likelihood of developing a disorder but does not confirm that the condition will be developed.

Carrier testing is used to determine if an individual has a genetic mutation for a disorder that is autosomal recessive[2]. Often, two mutated genes are required for a genetic disorder to develop; carrier testing determines if an individual carries a gene for a certain disorder. This type of test is important in the context of family planning because it can determine the chances of a child being born with a genetic disorder if the carrier status of the parents is known. Prenatal testing is performed during pregnancy to determine the child's risk of developing a genetic disorder. It differs from carrier testing because a prenatal test is used for prenate's at any point during a pregnancy whereas carrier testing is usually used for individuals who are family planning. Prenatal testing is controversial because unwanted test results are a common cause of

abortion. It sparks an ethical debate regarding whether abortions should be performed due to the knowledge of a genetic disorder. When considering public health, all types of genetic tests are beneficial in different aspects and provide individuals and their families with information to make properly informed decisions.

What are the benefits of genetic testing?

Genetic testing offers a wide array of benefits to those who are symptomatic or asymptomatic. A symptomatic person has a disease or disorder and presents with symptoms for it whereas an asymptomatic person does not present with symptoms. A diagnostic genetic test guides affected individuals in making decisions about management strategies and the possible interventions available for the particular disorder[3]. For example, a genetic test can determine whether cancer is due to an inherited gene which can alter treatment choices. This knowledge is important for being able to create the best plan to treat the disease. It can also prepare individuals for other health concerns that are commonly linked with the genetic disorder. Diagnostic genetic testing provides a clear answer which is important for the individual to guide treatment plans that will be most effective and plan for the future.

For asymptomatic individuals with familial risk, predisposition genetic testing is the most cost-effective and informative test that is available[3]. Asymptomatic individuals do not show symptoms for disease, so genetic testing is important because they will be properly informed about their risk for disease and preventative measures that can be taken. The knowledge of an increased risk of developing a disorder is very beneficial because it allows the individual to make choices that can lower his/her risk of developing the disorder/disease. More frequent screening, lifestyle changes, medications, and surgeries are steps that can be taken to

decrease one's risk of developing a disease[4]. Genetic testing allows for individuals to be proactive about their life and is imperative for promoting health. A genetic condition is also an important factor to consider when planning for future children; having knowledge about a genetic condition whether the individual is symptomatic or asymptomatic provides information that is necessary when deciding whether to have children if they have a higher risk of developing a genetic disorder.

Genome sequencing can also result in "secondary" or "incidental" findings, which are important to discover because these findings often involve disorders that cause cancer or cardiovascular disease[3]. These unexpected findings are important to understand because they can encourage preventative measures which otherwise would not be taken. Genetic testing in asymptomatic individuals is important because it not only clarifies an increased susceptibility to a disease, but also can detect diseases that are very unexpected. The awareness of one's risk of disease is important to establish appropriate treatment and management plans for individuals who present with a disease/disorder and individuals who are at an increased risk of developing a disease/disorder.

Although controversial, genetic testing within the pediatric population can provide improvements in child health. The justification behind mandating pediatric genetic testing is that society is obligated to promote health in children which can be done through early detection of disorders. It is argued that mandating screening is more important than parental rights to refuse, because an early intervention will promote child welfare[5]. Many diseases can be managed if detected early in life so mandating genetic testing allows for the child to receive the medical care that is necessary to benefit health. Mandating predictive testing for late-onset disorders such as Alzheimer's disease is discouraged for pediatric populations because there is usually no benefit

for the child. For some late-onset conditions, knowledge of the risk of disease can be extremely beneficial; for conditions such as hypertension, diabetes, and coronary artery disease, lifestyle changes can be made to reduce one's risk. Although a child is not capable of understanding his/her predisposition to a certain disorder, it is important for the family to understand it because there might be preventative measures that can benefit the child. Even though genetic testing can detect disorders that are currently not treatable or preventable, it is important to test children for these disorders because it allows for preparation and planning for the future of the child.

Newborn screening for genetic disorders is critical in promoting health. The purpose of the screening is to limit morbidity and mortality that is caused by specific inherited diseases[5]. When it comes to a newborn population, genetic testing is a critical part of health evaluations because early detection can greatly impact the future of the child. Newborn genetic testing can prevent death or disability because the early detection allows for changes in medical plans to be made. Genetic testing in newborn's can determine their risk of developing a disorder such as Type 1 Diabetes, which can lead to the implementation of a treatment plan early on to decrease the severity of the disorder[6]. Although it is often argued that newborns should not be tested for diseases that do not have available treatment, knowledge of a late-onset disease can psychologically benefit the parents. Emotional and practical preparation is important when understanding the possibility of a child developing a disorder later in life and the preparation can encourage a healthy lifestyle. Furthermore, the medical field is constantly advancing which gives hope to the development of treatments that can possibly cure or manage disorders that are currently incurable.

Why should genetic testing be mandated?

Genetic testing offers individuals with insight into their health which can guide major health care decisions. Mandating genetic testing can provide benefits to people of all ages. Currently, Newborn Genetic Screening (NBS) can be refused due to religious beliefs and only tests for a few diseases such as phenylketonuria, congenital hypothyroidism, and sickle cell disease. Mandatory genetic testing with no exceptions promotes child welfare because all the tests that are currently mandatory test for disorders that are treatable or can be successfully managed if detected early on. The purpose of public health is to prevent disease, promote healthy lifestyles, and prolong life; this includes preventing childhood illness, so it is necessary to mandate genetic testing of all children, especially when many of the disorders are treatable. Although current medical practice accepts the refusal of treatment due to religious beliefs, newborns lack autonomy which is the reason parents should not be able to refuse genetic testing; the goal of physicians and medical practitioners is to ensure the health and wellness of the newborn, so genetic testing for a wide array of diseases should be mandated because it is in the best interest of the child. It is unfair to take away a child's right to treatment because of the beliefs of the parents. Also, by expanding newborn genetic screening programs, physicians will be able to detect susceptibility to numerous diseases as opposed to only testing for those that are treatable. It is important to test for diseases that are not currently treatable because it increases the awareness about the wide array of diseases that are still not fully understood.

Preventative Genome Sequencing (PGS) which is a type of genetic test that determines the risk of genetic disorders in individuals who are not considered "high risk". The purpose of PGS is to identify those who unknowingly carry genetic mutations for preventable diseases. PGS programs can benefit public health by determining the individuals who are at risk for a

potentially preventable disease[7]; they can minimize health harms and decrease the prevalence of certain diseases and disorders. By mandating tests such as PGS, public health is promoted because individuals who unknowingly carry mutations will now be able to seek treatment to prevent the disease. With all members of society being tested, physicians and other health practitioners will be able to treat a larger population for preventable diseases which decreases the prevalence of disease in society. Although mandating genetic testing could be considered a violation of individual rights, when considering public health, this violation is dismissible due to the collective public health goals. Mandating PGS programs are especially fundamental for benefitting public health because they identify those who are at risk for preventable diseases. It is necessary to detect these diseases early on as it will increase the success of the treatment.

It is argued that "caution is essential" when discussing the use of genetic testing within a public health setting. A predictive genetic test is only able to determine if there is a mutation that increases the risk of developing a disease later in life, so it does not provide a definitive answer that would allow for treatment. Is it necessary to mandate genetic testing if it is only predictive, or if it determines the diagnosis of a disorder that is not treatable? Genetic testing provides the individual with the knowledge that is important when making decisions about their future, such as having children. Although it may seem as if a predictive test would not benefit the health of the individual, the indirect benefit is the ability to make informed decisions about one's future. Even though positive test results do not guarantee that treatment is available, receiving the information about one's health is important for mental preparation and can also encourage a healthy lifestyle to manage the disease.

Mandating genetic testing is also important because it will promote awareness and encourage research about diseases and disorders that do not currently have cures. There is

currently a project initiated by the Centers for Disease Control and Prevention (CDC) which supports a process that analyzes genetic tests in a public health practice, rather than solely a clinical setting[8]. This shift from clinical to public health will promote research that will reduce morbidity and mortality among the population. The purpose of the shift to public health is to improve quality of life through the detection of disease; detecting diseases and disorders early on is essential in promoting the well-being of populations, regardless of whether there is a cure. By testing larger populations, medical professionals will be better able to understand the prevalence of disease in society and make connections when possible to determine cause of disease and external factors that may be affecting the development of disease.

It is well established that individuals who test positive for a genotype will not always develop the disease. Predictive testing provides insight regarding one's susceptibility, but an increased susceptibility does not guarantee that the disease will occur at some point. By mandating genetic testing, health care professionals will better understand environmental factors that play a role in disease development. Determining susceptibly allows for the identification of environmental factors that can lead to disease among individuals who have a predisposition[9]. Genetic influence along with environmental interaction will be able to be studied to a greater degree which allows for a better understanding of genetic disorders. Understanding the connection between genes and environment is crucial for developing treatment and management plans that consider multiple factors rather than being solely based on genes. Including genetic testing in the public health policy will work towards the goal of reducing morbidity and mortality associated with genetic disorders. Mandating genetic testing creates the opportunity for advancement in medicine regarding the understanding of environmental effects on genetic susceptibilities.

Mandating genetic testing will also play a role in the effectiveness of vaccinations. Current medical practice administers the same vaccines to everyone in the population without the consideration of adverse reactions[13]. It is assumed that everyone is at the same risk of developing the specific disease which leads to everyone receiving the same dosage and number of dosages to develop immunity to the disease. This assumption is incorrect because it disregards the individuality of disease risk based on genetic predispositions. By mandating genetic testing, physicians will be properly informed about the immunology aspect of the genome which allows for individualized vaccines that will be most beneficial. Although vaccines work efficiently to benefit public health, it will be just as beneficial to personalize health treatments which includes immunologic preventative measures. Genetic testing will provide the necessary information to ensure that the vaccines being administered are most effective for the individual. A personalized vaccine will enhance the outcomes and minimize the risk of failure or serious complications. Vaccines will be more effective because they will be developed with the consideration of individual reactiveness to vaccine antigens and the level of need for immunity to infectious disease[13]. By recognizing genetic differences, clinicians will be better able to make more informed decisions to benefit the patient. It also creates opportunities to advance vaccines and their effectiveness in society which will benefit the public. Vaccines can be developed based on algorithms that consider individual genetic restrictions and differences that alter responses to standard vaccines. The specificity of vaccines development will provide individuals with immunization that will be most beneficial to them, which improves public health.

Ethical Issues with Mandating Genetic Testing

There are many ethical issues within the context of genetic testing for diseases and disorders that do not have a cure. Genetic test results can often be misinterpreted by physicians which can lead to many more tests being ordered when they may not be necessary[9]. These tests usually have risks associated with them which can lead to complications. The lack of education for genomic medicine in non-specialized physicians can majorly impact the recommended treatment plans for individuals who may not even be at risk for the disease/disorder. Genetic testing will be most beneficial when physicians are able to provide their patients with information that is correct, useful, and offers guidance that provides a clear benefit. It is important to better educate physicians on genetic testing and counseling, so they can provide their patients with accurate information without overestimating or underestimating the patient's risk of the disease or disorder.

Complex genetic information regarding the future of someone's life can be very unsettling, especially when the information is about someone's unborn child. Individuals and families are often forced to make "tragic" choices after receiving negative genetic information about their child. Parents have a choice to either raise a child with a genetic disorder knowing that he/she might be susceptible to injustices, such as social inequities, or choose to terminate the pregnancy. The ethical dilemma lies in the ability to terminate a pregnancy due to the knowledge of a genetic disorder. Currently, serum screening and ultrasound screening are performed around 20 weeks of gestation to assess the risk of a fetal genetic disorder[10]. Depending on the results, more testing is recommended to confirm a disorder. Although many states restrict late-term abortion with no exceptions, it is legal in some states. Many argue that it is unethical to terminate a pregnancy after 20 weeks because the fetus is considered viable, but it

also can be considered unethical to raise a child that will have a poor quality of life. It can be argued that it is beneficial to end a late-term pregnancy due to a genetic disorder because the health implications will result in a debilitating life, but the true importance of mandating genetic testing lies in the parent's right to all information that can guide decisions. Genetic testing will provide parents with the information that is necessary when deciding whether to terminate a pregnancy because of a major disorder or raise the child. It is the right of the parent's to have as much information as possible about the health status of the fetus in order to make a decision about their future.

Many issues arise when discussing the obligations of the physician and the patient in regards to genetic test results. Physicians often feel as if it is their responsibility to provide genetic counseling because they are knowledgeable about the patient and their background. The downfall with physicians providing counseling is that they are not required to be trained in genetic testing and counseling so there is often a lack of knowledge[11]. This can be detrimental when providing options for the patient as the lack of knowledge may drive a patient to make an unnecessary decision. For genetic testing to truly benefit the public, physicians need to be well educated in genetics and counseling because they are the individuals who provide patients with plans for treatment. By increasing education in genetic testing and counseling, physicians will be better able to inform their patients about treatment plans and preventative measures that can be established.

There are also moral theories that play a role in answering important questions regarding predictive genetic testing and what should be done with the results. From a utilitarian perspective, determining one's susceptibility to a genetic disorder is necessary to promote the good of society[10]. Individuals who know of a disorder can prevent it from being passed onto

future generations by choosing to not have children. By preventing the spread of disease/disorders, the common good is being promoted and public health is improved. This will also save money for treatments that are often paid for by the public. It is beneficial to spread awareness about genetic diseases because the knowledge can prevent the disorder from affecting a greater population and burdening both individuals and society with the cost of treatment.

Another moral theory comes into play when considering personal autonomy. From a libertarian perspective, it is the right of the family members not to know about a genetic disorder. With the knowledge about their risk of a disorder, individuals might make life altering decisions that may or may not be necessary[10]. A libertarian would argue that it is the right of the individual to make decisions about the knowledge of their disorder without interference from physicians, counselors, and family members. This argument is faulty because it removes individual justice of family members from the issue at hand. Family members should have the right to know about their susceptibility to disorder. It is important to acknowledge that many genetic disorders are familial, so if one family member is tested, it is important to inform the rest of the family about the results. Without knowledge of a predisposition to a disorder, family members of an individual who has a genetic disorder do not have the information that is needed to seek treatments; even if treatments are not available, research about genetic diseases is ongoing and there is always a chance that a new treatment will be discovered. If family members are not required to be informed about their possible risk of a genetic disorder, their ability to seek out preventative measures is taken away.

The purpose of genetic testing is to detect diseases and disorders in order to promote the health and well-being of individuals. By mandating genetic testing, the health of individuals is promoted by providing information that can be beneficial in developing treatments plans,

management strategies, and guide family planning. Family planning leads into promoting public health, which encompasses many populations and improves human health and quality of life. In order to successfully promote public health, individuals who are susceptible to genetic disorders must understand their risk of passing on disorders. Mandating genetic testing in newborns is also necessary because it provides physicians and parents with the information that is needed to either begin treatment or simply be aware of the possibility of disease development. Although mandating genetic testing may seem extreme, it is for the common good and can promote health in multiple aspects.

Bibliography

1. Resta RG. What have we been trying to do and have we been any good at it? A history of measuring the success of genetic counseling. *European Journal of Medical Genetics*. 2019;62(5):300-307. doi:10.1016/j.ejmg.2018.11.003.

2. Pagon RA. Genetic testing. *Western Journal of Medicine*. 2001;174(5):344-347. doi:10.1136/ewjm.174.5.344.

3. Correction: Precision Medicine for Clinicians. *Annals of Internal Medicine*. 2019;170(12):912. doi:10.7326/l19-0314.

4. Genetic Testing for Inherited Cancer Susceptibility Syndromes. *National Cancer Insitute.* March 2019 . https://www.cancer.gov/about-cancer/causes-prevention/genetics/genetic-testing-fact-sheet#q6.

5. Ethical Issues With Genetic Testing in Pediatrics. *Pediatrics*. 2001;107(6):1451-1455. doi:10.1542/peds.107.6.1451.

6. Kerruish NJ. Newborn screening: new developments, new dilemmas. *Journal of Medical Ethics*. 2005;31(7):393-398. doi:10.1136/jme.2004.008219.

7. Lázaro-Muñoz G, Conley JM, Davis AM, Riper MV, Walker RL, Juengst ET. Looking for Trouble: Preventive Genomic Sequencing in the General Population and the Role of Patient Choice. *The American Journal of Bioethics*. 2015;15(7):3-14. doi:10.1080/15265161.2015.1039721.

8. Marzuillo C, Vito CD, Dandrea E, Rosso A, Villari P. Predictive genetic testing for complex diseases: a public health perspective. *Qjm*. 2013;107(2):93-97. doi:10.1093/qjmed/hct190.

9. Khoury MJ. From genes to public health: the applications of genetic technology in disease prevention. Genetics Working Group. *American Journal of Public Health.* 1996;86(12):1717-1722. doi:10.2105/ajph.86.12.1717.

10. Oliveri S, Pravettoni G, Fioretti C, Hansson MG. Let the Individuals Directly Concerned Decide: A Solution to Tragic Choices in Genetic Risk Information. *Public Health Genomics.* 2016;19(5):307-313. doi:10.1159/000448913.

11. Rebouche R, Rothenberg KH. Mixed Messages: The Intersection of Prenatal Genetic Testing and Abortion. *Howard Law Journal*. 2012;55(3):983-1126. Accessed October 20, 2019.

12. lda KG. Ethical issues in predictive genetic testing: a public health perspective. *Journal of Medical Ethics*. 2006;32(3):143-147. doi:10.1136/jme.2004.010272.

13. Poland GA, Ovsyannikova IG, Jacobson RM. Personalized vaccines: the emerging field of vaccinomics. *Expert Opinion on Biological Therapy*. 2008;8(11):1659-1667. doi:10.1517/14712598.8.11.1659.