

# **FEAR OF GENETIC DISCRIMINATION**

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By

Tony Albini

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On my honor as a University student, I have neither given nor received unauthorized aid on this assignment as defined by the Honor Guidelines for Thesis-Related Assignments.

ADVISOR

Catherine D. Baritaud, Department of Engineering and Society

In 1990, the United States government created the Human Genome Project to form a dedicated body whose purpose was to map the entire human genome. By 2000, scientists had sequenced nearly the entire human genome (Collins & McKusick, 2001, p. 540). As DNA sequencing technology continued to improve, the ability for people to have their own DNA tested increased as well. Private companies began to provide options for people to obtain genetic testing information in the mid-2000s (Borry, Cornel, & Howard, 2012, p. 101). Through genetic testing, people could better modify risk factors according to genetic risk stratification, receive earlier care for diseases, or have “designer” drugs created specifically for them based on their genes (Collins & McKusick, 2001, p. 540). Although genetic testing provides great promise, public hesitation and fear regarding its consequences exist. Fear of genetic discrimination from health insurance companies is a common reason for people to not support genetic testing or to forego the option entirely (Allain, Friedman, & Senter, 2012, p. 640). Genetic testing offers potential life-altering benefits, but the presence of public fear could restrict the technology’s capability. The STS research involved in this project will analyze the public’s fear surrounding genetic testing and a potential mitigation method.

The technical side of the project aims to create a better way for physicians to determine treatment plans for patients with early onset scoliosis (EOS), which is principally caused by genetics in roughly two thirds of surgical cases (Gao et al., 2014, p. 325). Patients that can be diagnosed due to their genetic data will be affected by the ways insurance companies will be allowed to use this information. Currently, there is not an exact guideline for physicians on how to treat EOS (Cunin, 2015, p.S117). Through providing the total lung capacity, physicians would know the optimal time for surgical intervention. Both projects aim to provide better treatment to patients, especially those who suffer from genetic diseases. The paper presented here will use the

Social Construction of Technology (SCOT) (Pinch & Bijker) in order to outline the social groups that affect the future of genetic testing (1987). Through engineers generating communication by creating focus groups, the fear surrounding genetic testing should decrease.

### **FEAR OF GENETIC DISCRIMINATION**

Although genetic testing is currently only used for specific genes and rare conditions, the National Institutes of Health predicted that the price for genetic testing will continue to decrease (National Institutes of Health, n.d.). As expenses decrease, more people will be able to afford a genetic test. Through genetic testing, people can identify diseases they already have or are at more risk of developing. This information could lead to early treatment of diseases or allow people to make lifestyle decisions that could mitigate the problems associated with various diseases (Belisle-Pipon, Vayena, Green, & Cohen, 2019, p. 1198). Genetic testing also provides the possibility of unique and improved treatment for each patient (Nill, Lacznik, & Thistle, 2019, p. 105).

Although genetic testing provides great promise, people are hesitant to undergo testing due to fear of genetic discrimination. Specifically, people expect that they and their family will be forced to pay increased amounts for health insurance (Wauters & Van Hoyweghen, 2016, p. 279). This fear leads to some people foregoing genetic tests despite the potential benefits (Allain et al., 2012, p. 640). Even those who know they are predisposed to a certain condition may make the conscious choice to not get tested. In a specific case study on breast cancer, a group of women who had over 20% chance of developing breast cancer decided to not get tested, specifically citing the potential for insurance genetic discrimination (Armstrong et al., 2003, p. 362). However, given these fears, Armstrong et al. (2003) found no evidence to show that insurance companies were using genetic information as a tool to change insurance premiums (p.

362). This raises the question why does the public fear genetic discrimination from insurance companies if there is a lack of evidence?

On the other side of this problem, insurance providers fear that without genetic testing they put themselves at financial risk. Adverse selection is the instance where people who are at greater risk will continue to buy insurance while those at low risk are less likely to get insured (Nill et al., 2019, 114). This situation creates financial risk for companies because, in an extreme case, they will only be providing insurance to those who will need to cash in on their plans to cover their medical expenses. In the above case concerning breast cancer, the women involved were likely to alter their insurance plans due to their genetic test results (Armstrong et al., 2003, p. 363). Therefore, insurance companies may have credible concerns for market failure.

Alternatively in 2000, Zick, Smith, Mayer, and Botkin stated that the potential for adverse selection is non-existent (p. 37). This study documented the genetic tests and responses of women who were examining the BRCA1 gene linked to breast cancer. Women who tested positive for the BRCA1 gene mutation were no more likely to change their insurance plans than the women who tested negative (Zick et al., 2000, p. 37). Armstrong et al. (2003) acknowledges this difference by stating the number of women who would change their plans could be miniscule compared to those who do not and would not cause adverse selection (p. 363). These findings should limit the concerns of insurance companies.

If the current climate surrounding genetic testing continues to exist, the public risk losing the benefits genetic testing can provide. Another large barrier to the success of this technology is the lack of understanding of genetic testing legislation. For instance, the United States government passed the Genetic Information Nondiscrimination Act (GINA) of 2008, which explicitly protects people from discrimination from health insurance companies. The act states

that insurance companies cannot use genetic testing information to deny coverage or make premium decisions (National Institutes of Health, 2020). Even after the enactment of this policy, people still chose to not get tested due to fear of discrimination (Allain et al., 2012, p. 640). Allain et al. (2012) stated that a lack of knowledge of the current legislature could be the reason for the continued presence of fear (p. 641). Additionally, misunderstanding exists among the government officials whose job is to enforce insurance legislation (Golinghorst & Prince, 2019, p. 3). Different states have different laws on how genetic information can be handled by health insurance companies. Golinghorst and Prince (2019) suggested this complicated legislative reality has led to their misunderstanding (p. 5). There is also little evidence of communication between the public and the policy enforcers. The fear exists, but state insurance commissioners do not hear of it directly from their constituents. A line of communication is needed between the state commissioners and the public. Additionally, it is imperative that both the public and policy enforcers obtain a better understanding of the genetic testing legislature for the promises of genetic testing to substantialize.

## **ENGINEERS FORCE COMMUNICATION**

To ensure genetic testing reaches its fullest capabilities, a closer look at how society interacts with this technology is necessary. The engineers who are developing genetic testing technology will need to ensure its success. The Social Construction of Technology (SCOT) framework will be used to illustrate the social relation engineers have in this problem (Pinch & Bijker, 1987). According to the SCOT framework, the engineer or developer does not produce the technology in a vacuum. Instead, relevant groups of people also affect the outcome and development of technology (Pinch & Bijker, 1987). Technological development is dependent on

the feedback loop between the engineers and the relevant groups. The success of genetic testing relies on the creation of a SCOT model, as shown in Figure 1.

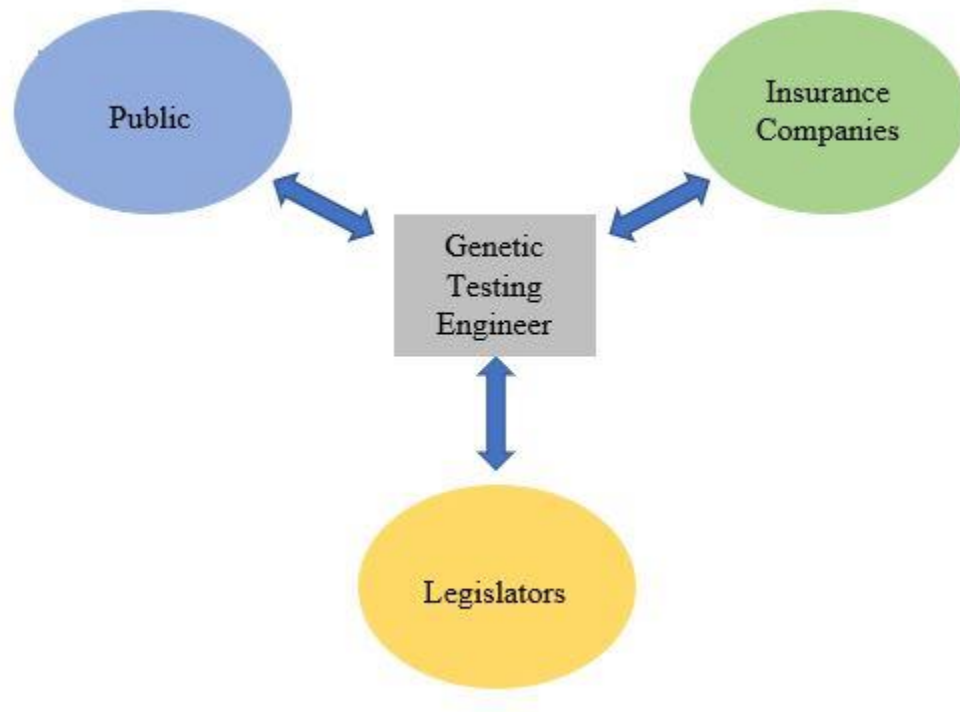


Figure 1: Genetic testing SCOT model: The engineer at the center is expected to take each actor's interests into account to produce a successful genetic testing product (Adapted by Anthony Albini (2020) from W. B. Carlson, 2009).

The Social Construction of Technology (SCOT) model presented above identifies the public or policyholders, insurance companies, and legislators as relevant social groups. Therefore, these groups will affect how engineers develop genetic testing technology. As mentioned previously, these social groups have already altered the technology. Genetic testing is at a point where society may lose its potential because of the actions of the social groups identified. Engineers will need to keep in mind the values of each of the groups in this model to allow genetic testing to succeed. As engineers continue to develop genetic testing to the point it becomes cost effective for widespread usage, they will need to effectively communicate with each group. The policyholder's suspicion of suffering from genetic discrimination, the health

insurance industry's fear of market failure, and the legislative wish to protect its constituents need to all be considered.

### **FOCUS GROUPS: THE SOLUTION**

Rogers et al. (1996) outlines a study of the STOP AIDS campaign where the creation of focus groups allowed information to spread through targeted communities (p. 10-11). A system that the engineer should try to obtain through creating focus groups in which the three groups can communicate their concerns and values is shown in Figure 2 below.

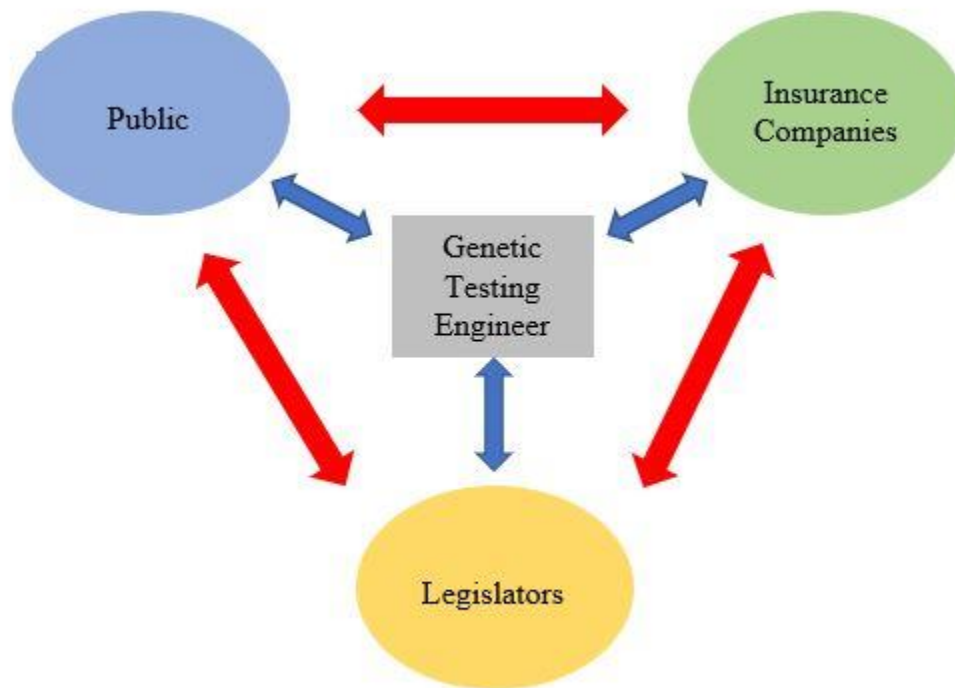


Figure 2: Genetic testing SCOT model with improved communication: Similar to the previous SCOT figure except the red arrows signify the engineer focusing on improving communication between the social groups (Albini, 2020).

The focus of the engineer should be to learn the values of each group through creating communication networks between the groups. As Golinghorst and Prince (2019) stated, if communication between the state commissioners of insurance and the public increases, the benefits of genetic testing can be allowed to occur (p. 6). The public could learn of the protection

the Genetic Information Nondiscrimination Act provides from state commissioners, which could diminish their fear. Increase of feedback should not end here, legislators should also consider the values of the insurance industry. Additionally, insurance companies will need to consider the values of their policyholders and vice versa. As Zick et al. (2000) found, insurance companies could learn that their policyholders are not likely to change their insurance plans based on genetic testing (p. 37). Therefore, their fear of economic failure should decrease. Further, the public can learn that insurance companies do not have the power to alter their premiums. The groups will become more informed, while their fears should decrease. With respect to the AIDS pandemic, once relevant social groups communicated, the overall public health improved (Rogers et al., 1996, 10-11).

An additional factor that has enabled the spread of beneficial health practices is the inclusion of peer opinion leaders (Valente & Fosados, 2006, p. S28). These leaders are identified as trusted individuals within relevant social groups. According to Valente and Fosados (2006), studies have shown that identifying trusted members of relevant social groups is effective at creating change (p. S29). In order to choose the peer opinion leaders, first the relevant social groups need to be selected. As stated above, these groups are the policyholders, the legislators, and the insurance companies. A survey would be given to each group and those who receive the most positive nominations will be selected as peer opinion leaders. (Valente & Fosados, 2006, p. S29). Once the engineers have identified peer opinion leaders, they could then take part in effective discussion during the focus groups. After the focus groups, opinion leaders are expected to then spread the information received from the focus groups to their respective social groups. Individuals would listen to the leaders and could adopt new practices. If the focus groups are successful a succession of adoption would occur as shown in Figure 3 on page 8.



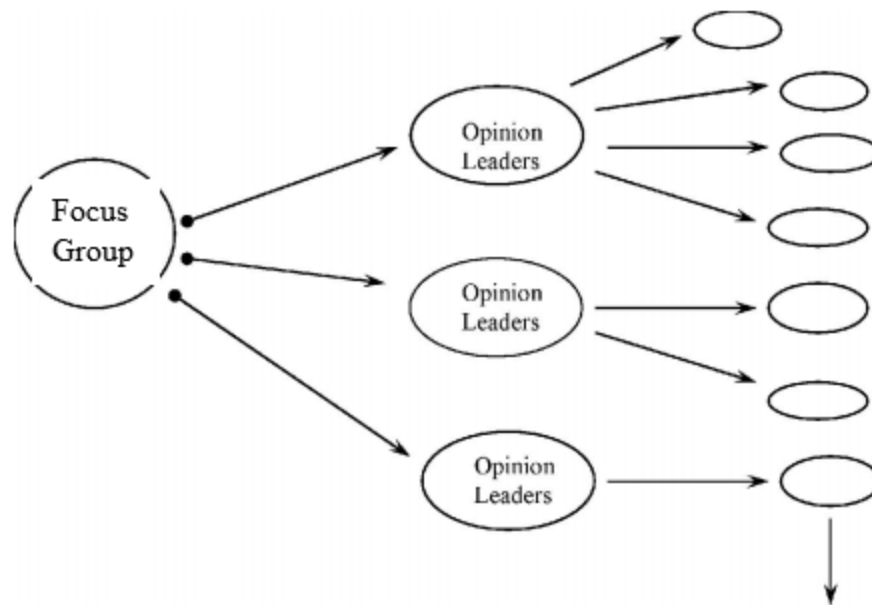


Figure 3: Model of adoption: This shows how the first adopters of the new health practice will be the opinion leaders who attend the focus groups. The opinion leaders then transfer the learned information to the members of their respective social groups (Adapted by Anthony Albini (2021) from Valente & Fosados, 2006).

The figure illustrates the importance of the opinion leaders. The fear of the leaders who represent the policyholders and the insurance companies should diminish. Additionally, the legislators should be better equipped to enact effective legislation. From this point, the opinion leaders would express their views to their respective social groups. After this, the public health benefits of genetic testing could achieve their potential.

Valente and Fosados (2006) outlined an additional technique for the dispersion of information. This technique is called mass media campaigns, which involves opinion leaders obtaining all their information from the media. This technique follows a similar path as displayed in Figure 3, except the information originates from media rather than focus groups. In this case, another difference is opinion leaders are those who pay increased attention to media (Valente & Fosados, 2006, p. S24). Mass media campaigns are not as effective as the use of focus groups

and carefully identified opinion leaders. This failure is two part. One, the reaction to information is difficult to predict and control. Second, the absence of understanding the relevant social groups does not tend to change the practices of individuals (Valente & Fosados, 2006). By allowing opinion leaders among social groups to communicate, individuals with the groups are more likely to change their behavior.

## **FUTURE IMPLICATIONS AND RESEARCH**

The Social Construction of Technology allows engineers to understand the social groups who will influence genetic testing technology. Engineers can then create focus groups and generate effective communication. The success of these focus groups should lead to reduced public fear surrounding genetic testing. Policyholders may feel more open to getting tested and learn that legislation exists to protect them. Additionally, legislators can learn the fears of their constituents, which include both the policyholders and insurance companies. Then, they can create and enforce better legislation. The fears insurance companies hold regarding adverse selection could also be eliminated. Hopefully, increased understanding would allow public health to greatly improve through the creation of personalized medicine and preventative treatment.

Future research could examine if specific diseases would benefit more from these methods. If true, researchers can look to create social networks with those who would benefit the most from genetic testing. As Armstrong et al. (2003) wrote, scientists already know there are specific genetic mutations that increase one's chance of developing breast cancer (p. 360). Researchers could examine these disease groups to develop quicker progress. Further, a demographic study on the fear of genetic testing could offer insight. If specific races or genders are more likely to not get tested, focus groups could concentrate on them. Finally, studies could incorporate genetic counselors as an additional social group. Genetic counselors provide

expertise and guidance for understanding genetic test results. The relationship between policyholders and genetic counselors could provide useful information.

## WORKS CITED

- Albini, A. (2020). *Genetic testing SCOT model*. [Figure 1]. Prospectus (Unpublished undergraduate thesis). School of Engineering and Applied Science, University of Virginia. Charlottesville, VA.
- Albini, A. (2020). *Genetic testing SCOT model with improved communication*. [Figure 2]. Prospectus (Unpublished undergraduate thesis). School of Engineering and Applied Science, University of Virginia. Charlottesville, VA.
- Albini, A. (2021). *Model of adoption*. [Figure 3]. Research Paper (Unpublished undergraduate thesis). School of Engineering and Applied Science, University of Virginia. Charlottesville, VA.
- Allain, D., Friedman, S., & Senter, L. (2012). Consumer awareness and attitudes about insurance discrimination post enactment of the Genetic Information Nondiscrimination Act. *Familial Cancer, 11*(4), 637-644. doi:10.1007/s10689-012-9564-0
- Armstrong, K., Weber, B., Fitzgerald, G., Hershey, J. C., Pauly, M. V., Lemaire, J., . . . Asch, D. A. (2003). Life insurance and breast cancer risk assesment: Adverse selection, genetic testing decisions, and discrimination. *American Journal of Medical Genetics, 120A*(3), 359-364. doi:10.1002/ajmg.a.20025
- Borry, P., Cornel, M. C., & Howard, H. C. (2010). Where are you going, where have you been: a recent history of the direct-to-consumer genetic testing market. *Journal of Community Genetics, 1*(3), 101-106, doi:10.1007/s12687-010-0023-z\
- Collins, F. S. & McKusick, V. A. (2001). Implications of the Human Genome Project for Medical Science. *JAMA, 285*(5), 540-544, doi:10.1001/jama.285.5.540
- Cunin, V. (2015). Early onset scoliosis - Current treatment. *Orthopaedics & Traumatology: Surgery & Research, 101*(1), S109-S118, doi:10.1016/j.otsr.2014.06.032
- Gao, X., Gotway G., Rathjen, K., Johnston, C., Sparagana, S. & Wise, C. A. (2014). Genomic analyses of patients with unexplained early onset scoliosis. *Spine deformity, 2*(5), 324-332, doi:10.1016/j.jspd.2014.04.014
- Golinghorst, D. R., & Prince, A. E. R. (2019). A survey of U.S. state insurance commissioners concerning genetic testing and life insurance: Redux at 27. *Journal of Genetic Counseling, 1*-8. doi:10.1002/jgc4.1197
- National Institutes of Health. (2020, September 16). Health Insurance (Title 1). In *Genetic Discrimination*. Retrieved from <https://www.genome.gov/about-genomics/policy-issues/Genetic-Discrimination>

- National Institutes of Health. (n.d.). Genetic Testing: How it is Used for Healthcare. In *NIH Fact Sheets*. Retrieved from <https://archives.nih.gov/asites/report/09-09-2019/report.nih.gov/nihfactsheets/ViewFactSheetef83.html?csid=43&key=G#G>
- Pinch, T. J., Bijker W., & Hughes, T. P. (1987). The Social Construction of Facts and Artifacts. *The Social Construction of Technological Systems*. 17-50.
- Rogers, E. M., Singhal, A., Quinlan, M. M., Stacks, D. (Ed.) & Salwen, M. (Ed.). (1996). *An integrated approach to communication theory and research*. New York, NY: Routledge.
- Valente, T. W. & Fosados R. (2006). Diffusion of innovations and network segmentation: the part played by people in promoting health. *Sexually Transmitted Diseases*, 33, S23-S31. doi: 10.1097/01.olq.0000221018.32533.6d
- Wauters, A., Van Hoyweghen, I. (2016). Global trends on fears and concerns of genetic discrimination: a systematic literature review. *Journal of Human Genetics* 61(4), 275-282. doi:10.1038/jhg.2015.151
- Zick, C. D., Smith, K. R., Mayer, R. N., & Botkin, J. R. (2000). Genetic testing, adverse selection, and the demand for life insurance. *American Journal of Medical Genetics*, 93(1), 29-39. doi.org/10.1002/1096-8628