**Face2Gene: Using Facial Recognition to Aid in Diagnosing Rare Genetic Disorders**

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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

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**Abstract**

Face2Gene utilizes facial recognition technology to analyze a "selfie" of a patient, then comes up with a list of most likely diagnoses. With over 6,000 rare genetic disorders that are often accompanied with a variety of subtle but distinct facial cues, diagnosticians need assistance in swiftly and accurately providing a diagnosis for people with rare genetic disorders. As this application has been built on a machine learning model, it has been molded to suit the accuracy and diversity needed by medical professionals and patients alike. As such, Face2Gene provides a rich subject for analysis under the framework social construction of technology (SCOT) because it is an application that has been shaped by human action. This paper will provide background information on Face2Gene, then analyze how the application has grown since its launch in 2014 under the SCOT pillars of interpretative flexibility, wider context, and closure & stabilization.

## Introduction

Facial recognition technology has come a long way since 1964, when scientists first trained computers to recognize the human face (NEC, 2020). The vast improvements in camera quality, computing power, and machine learning algorithms over the past few decades have made facial recognition immensely powerful. While utilization of facial recognition technology is most often associated with law enforcement, it has proved to be beneficial in a vast majority of industries. For example, facial recognition is being used in the healthcare industry to speed up patient check-in at medical facilities, track a nonverbal patient's pain via their expression (and adjust their medication accordingly), and even to aid in diagnostics (Crompton & Diamond, 2019).

Facial recognition has the potential to solve a major problem in the healthcare industry by assisting in diagnosis of rare genetic disorders. The number of rare genetic disorders has surpassed 6,000, and each new disorder comes with an array of subtle symptoms of which diagnosticians cannot always detect nor recall. As the number of people who currently live with rare genetic disorders exceeds 300 million, there is a critical need for swift diagnoses of these disorders. That being said, patients with rare genetic disorders usually wait between 5.6 and 7.6 years to receive a proper diagnosis (Vandeborne et al., 2019). This is where facial recognition technology can come in handy, as 30% to 40% of these genetic disorders involve detectable changes to the face or skull (*Medical products: Facial recognition technology to diagnose rare genetic diseases*, 2015).

Face2Gene is the one of the first applications that utilizes facial technology to diagnose these types of rare disorders. In fact, its creators were the inventors of the first facial recognition technology that actually outperformed humans. Face2Gene allows smartphone users, physicians

and patients alike, to take self-photographs and generate a list of the most likely diagnoses based on detected facial features. Since Face2Gene is built atop machine learning, the model increases in accuracy and range the more that it is utilized. (Wallner, 2017).

The application Face2Gene is a prime example of a technology that has been shaped by its users, making it a prime technology for study under the framework social construction of technology (SCOT). This research paper will first provide background information on the SCOT framework, then will detail how Face2Gene has been molded to fit its users' unique needs via the SCOT pillars: interpretive flexibility, wider context, and closure/stabilization. With an analysis under the SCOT framework, it will become clear that Face2Gene is a technology shaped by human action because of how its users have enhanced the accuracy and diversity of the application since its launch.

## Conceptual Framework: SCOT

SCOT is a framework that emerged in 1987 from *The Social Construction of Technological Systems*, written by Wiebe Bijker and Trevor Pinch. The framework initially consisted of four related components: interpretive flexibility, relevant social group, closure and stabilization, and wider context. Interpretive flexibility suggests an open design process, with intergroup negotiations forming a product that is meaningful for all user groups. These user groups are called relevant social groups. A relevant social group encompasses one particular interpretation of the technology; in other words, members of a group assign the same meaning to the technology. The design phase is finally complete once each relevant social group has decided that the technology will meet their needs. The third component of SCOT, closure and stabilization, pertains to conflicts in the design of a technology (in this case, the application Face2Gene). If a relevant social group sees a problem with the current design, the design phase is

not over. Closure is finally achieved when the design phase has ended, which can only occur when the technology is considered noncontroversial by all relevant social groups. Pinch and Bijker categorized two kinds of closure and stabilization: closure by redefinition and rhetorical closure. In closure by redefinition, closure is achieved when the problem is redefined so that the problems posed by social groups are no longer relevant. In rhetorical closure, a decision is made that problems no longer exist; in other words, all groups agree to consider the problem solved in its original context. Lastly, wider context encompasses the cultural and political environment in which development occurs. It is essentially the background information necessary to understand the motives and interactions between relevant social groups, as well as external constraints on the design. It will be clear why SCOT is an appropriate framework for analyzing Face2Gene in the subsequent sections of this thesis, in which the application is explained within each pillar of SCOT.

Many scholars have since critiqued SCOT for its over simplicity. For example, all relevant social groups rarely have an equal say in the design process, nor are all members of a group identical in their beliefs. This is particularly true with the development of Face2Gene's machine learning model, as they began training the model with only a few dozen clinicians. As such, the training of the Face2Gene model in the three years between its beta launch in 2014 and public launch in 2017 was limited in perspective and scope (Mack, 2017). As such, the entire relevant social group of patients and their families did not get a say in the design process of the application. Additionally, the relevant social group of medical professionals was not necessarily given equal say, as the vast majority of that group did not have the chance to beta test of the application. As such, Face2Gene is not entirely adherent to this particular SCOT concept. That being said, the application's founders and initial clinician beta testers made an intentional effort

to diversify the application, such that the Face2Gene database included phenotypic/genotypic information for over 7,000 rare genetic disorders at its public launch (Mack, 2017). By training Face2Gene to recognize rare disorders that they themselves may not have ever needed to diagnose/treat, the stakeholders that had a voice in the design process also incorporated the needs of patients and clinicians beyond their own circles.

Another concept was later added to the SCOT framework called technological frame, which attempts to address these criticisms by providing room for more analysis of the structure of thinking within a relevant social group (Klein and Kleinman, 2002). Without this concept, there was little room for the technical background to be incorporated into a SCOT analysis. While the technological frame adds more realistic complexity to analysis, it will not be mentioned in this particular thesis. Instead, the subsequent analysis of Face2Gene will address the four original pillars of SCOT. After defining Face2Gene under each of these pillars and drawing the paper to a close with a conclusion section, it will be clear how the application has been shaped by human action rather than itself making an impact on society.

## Interpretative Flexibility

The relevant social groups must first be defined, in order to demonstrate how Face2Gene applies to other pillars of SCOT. The relevant social groups for the application Face2Gene include: the developers of Face2Gene (a startup company called FDNA), medical professionals tasked with diagnosing rare genetic disorders, and their patients (as well as patients' loved ones) who seek diagnosis. Note that these groups are inferred based on the major users of the application and may not be entirely comprehensive (but will be suffice for this analysis). These groups were inferred based on the major users of the application and those benefitting from its success.

These relevant social groups see Face2Gene as a means to different ends. For the developers of Face2Gene, they see their application as a means to aid medical professionals in diagnostics and patients in getting answers to their symptoms. The "About" section of the FDNA site includes the statement "Since its founding in 2011, FDNA continues to aid clinicians, researchers and genetic testing labs in finding answers and treatments for hundreds-of-millions of patients globally living with a genetic disease" (*About FDNA*, 2021). That being said, the founders of FDNA also saw a gap in the market that would allow them to achieve personal career success and worldwide recognition. On their website, FDNA states "Our MISSION is to disrupt clinical genomics by integrating NGP into the genetic testing workflow" (*About FDNA*, 2021).

For medical professionals, Face2Gene is an invaluable aid to diagnosis. The meaning of the application hinges upon its ability to make suggestions on the subtle facial cues that can be hard for physicians to detect or connect to one of the thousands of rare genetic disorders with such features. In an article that was published in the British Journal of General Practice, general practitioner and author Willian Evans writes "Knowledge of all 6000-8000 rare diseases, increasing each week, is impossible for any one individual. The response 'How can we know about all rare disease?' is not only true but also feeds into the cognitive barrier that prevents clinicians contemplating a rare disease at all" (Evans, 2018). Karen Gripp, MD and medical geneticist as Nemours/Alfred I. duPont Hospital for Children and professor of pediatrics at Thomas Jefferson University, stated in a 2020 interview "Sometimes the typical facial features [of a genetic disorder] are present, but they are so mild that a human expert has trouble identifying them. An algorithm is at times superior at identifying these" (Grifantini, 2020).

For patients with rare genetic disorders, as well as their loved ones, they see Face2Gene as an unbiased, reliable tool for compiling a list of likely diagnoses with just a selfie as input. As stated by FDNA founder Dekel Gelbman, "Patients living with a genetic disease often times go years without receiving a diagnosis, which means they are also not receiving the appropriate treatment or therapies that are available for some of these syndromes. By aiding in reaching a faster, more accurate diagnosis, patients can seek appropriate treatments and care that can improve their quality of life and extend their lives" (AI Time Journal Editorial Staff, 2019). As the various relevant social groups attached to Face2Gene are assigning different meanings to the application, it becomes necessary for all of these separate use cases and interests to be equally accounted for in the design process. Since it is rarely the case that all stakeholders have equal role in the design process and because Face2Gene unlike most technologies is actually actively shaped as it is utilized by stakeholders (the model strengthening to fit all users' needs as it is utilized), Face2Gene is still applicable under SCOT's interpretative flexibility.

## Wider Context

The SCOT pillar referred to as "wider context" encompasses all sociocultural and political backgrounds taking place at the time of the development and adoption of the technology. This is where it is especially clear that Face2Gene was not a technology with a purpose that was fixed during the design phase. The application was launched in 2014 in "stealth mode" to gather enough data to train the system, with a few dozen beta users sharing patient photos. By 2017 when the application was officially launched to the public, the application's database already had information for over 7,000 rare diseases. Face2Gene expressed a desire to create the world's largest repository of rare disease big data through users inputting selfies to

Face2Gene. Evidently, the founders of the application saw the value in allowing users to morph the application's model to fit their own needs (Mack, 2017).

A multitude of researchers and physicians have since published results of utilizing Face2Gene to diagnose various disorders. In January of 2016, the first study was published based on initial findings in the accuracy of Face2Gene in recognizing Cornelia de Lange syndrome. The average detection rate of medical experts was measured to be around 77%, while the new Face2Gene technology had a detection rate of 94%. (Basel-Vanagaite et al., 2016). Another study utilizing Face2Gene was released several months later, in December of 2017, by a group of researchers that ran tests of its ability to detect Down Syndrome. The application showed recognition of Down Syndrome in Caucasians with 80% accuracy, compared to Africans at 36.8% accuracy. The researchers then added more training data to the application that included cases of Africans with and without Down Syndrome and were able to increase the recognition to 94.7% accuracy. While this study revealed bias in the original data set supplied alongside Face2Gene, it also revealed how further training for particular ethnic groups makes the application incredibly powerful for any group. (Lumaka et al., 2017). In November of 2017, another group of researchers released results of a study applying Face2Gene to detecting two disorders: Emanuel (ES) and Pallister-Killian Syndrome (PKS) which further proved the breadth of knowledge that Face2Gene is capable of holding (Liehr et al., 2018). In December of 2017, just months after the Face2Gene application was launched, a group of researchers published a study in the Official Journal of the American Academy of Pediatrics detailing the use of Face2Gene to diagnose fetal alcohol spectrum disorders (Valentine et al., 2017). FDNA also published their own study in January 2019, utilizing Face2Gene's image analysis framework, DeepGestalt, with proof of the platform outperforming clinicians in three experiments. This

particular study highlights improvements in the DeepGestalt model, including the dataset's growth to 17000 images representing 200 syndromes. The increased efforts to grow the training set resulted in 91% top-10 accuracy in identifying a person's genetic disorder in the same study, which utilized 502 different images to come up with this result. (Gurovich et al., 2019). Through studies such as these, it is evident how the underlying algorithm of Face2Gene is capable of being morphed to fit the needs of its users both in accuracy of diagnosis and breadth of knowledge.

## Closure and Stabilization

SCOT also emphasizes closure and stabilization as signs of a technology shaped by society. As mentioned in the Conceptual Framework section, are two types of closure under the SCOT framework: rhetorical and redefinition of the problem. Face2Gene has experienced closure under redefinition of the problem, since it is merely an *aid* to diagnosis rather than a tool to provide a certain, singular disorder as the cause of a patient's symptoms. The context has been switched to aiding diagnosticians, rather than replacing them. Donald Basel, MD and medical director of the Genetics and Genomics Program, said about Face2Gene "It never gives you a diagnosis. It gives you an answer to consider". With the list of the ten most likely possibilities for diagnosis provided by the Face2Gene analysis, as well as the patients' medical history, Basel can make more confident and speedy diagnoses. According to Basel, "Machines can never replace humans. The power of the machine is only as good as the information going in and the people who interpret it. The machine can only take all of the data and process it, but it can't prioritize it". As of 2019, Basel and his colleagues had utilized Face2Gene to diagnose over 1,000 patients (Etter, 2019). Clearly more than Basel and his colleagues have accepted

Face2Gene; it has been adopted by over 70% of the world's geneticists across 2,000 clinical sites and in over 130 countries (*About FDNA*, 2021).

It should be noted that relevant social groups such as patients, their loved ones, and medical professionals have questioned the privacy of Face2Gene. Face2Gene has a comprehensive Data Sharing & Protection Policy that addresses any ethical concerns once raised by relevant social groups. To begin, patients' personal identifiers such as names, addresses, and social security numbers are not collected by the application. There are still concerns around the facial images and associated test results or any other information inputted by the physician (including, but not limited to, clinical observations and family history). As such, some may argue that Face2Gene does not make sense as an object for SCOT analysis as it has not been accepted entirely by the relevant social group encompassing patients and their loved ones, calling into question whether Face2Gene is an application shaped by its users. There is certainly a valid counterargument that Face2Gene founders did not incorporate users' opinions into the initial design of the application, and if they had then perhaps the application would not have utilized facial recognition at all. That counter argument certainly implies that users did *not* shape Face2Gene, nor is it an example of SCOT. That being said, Face2Gene's founders have since met the concerns of users to achieve closure for their application. In order to address privacy issues, Face2Gene's developers installed a "de-identification" process for facial photos in their data set so that it is not possible for someone who gains access to the application database to discern someone's identity. The application is applicable to HIPAA, EU data protection regulations, and meets the concerns of the relevant social groups (*Data Sharing & Protection Policy,* 2017). As such, a consensus has been established, achieving closure amongst the relevant social groups and ending the design phase of Face2Gene.

## Conclusion

As technology becomes more powerful and pervasive, people view technological artifacts themselves as holding the power to shape the structure of society. That being said, this concept of technological determinism is often not accurate, especially when it comes to technologies that employ machine learning. As machine learning algorithms improve upon themselves by user input, they specifically grow in accuracy and diversity based on users' needs. As such, users hold a great deal of power in molding these applications, even if they were not directly consulted during their initial design. Face2Gene is an example of a technology that has been directly shaped by human action. Its founders incorporated the other major relevant social groups, physicians and their patients, in the design process by launching in "stealth mode" to give the application's algorithm years to be shaped by its users to best fit their needs. Now that the application is public, it still is being shaped by the multitude of studies undertaken on it, growing more accurate where it is most needed by society. Through this analysis of Face2Gene under the SCOT framework, it is clear that Face2Gene is an application that has been shaped by human action because of how its users have molded the application to suit their needs.

In the future, it is likely that more applications of machine learning are created and subsequently popularized to meet society's needs. Face2Gene itself will continue to increase in accuracy the more data that it is provided (a.k.a. the more that its users utilize it), and it will also grow in scope as it is provided with data for new kinds of rare disorders. As CEO of FDNA Dekel Gelbman said in a 2017 interview with StartUs magazine, "The majority of clinical geneticists around the world are already using our technology. It is also used by a large body of researchers, drug developers and genetic testing labs working to find answers and treatments for hundreds-of-millions of patients globally. In the future, every person's genome will serve as their

medical record. Our technology will help patients are their caregivers understand their health better and come up with a personalized care plan – this is our role in the future of precision medicine" (Wallner, 2017). Face2Gene will continue to be a prime example of SCOT as it is being directly shaped by society's needs.

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