

Creating a Temperature-Tracking Door Locking System for COVID-19
(Technical Paper)

Face2Gene: Using Facial Recognition to Aid in Diagnosing Rare Genetic Disorders
(STS Paper)

A Thesis Prospectus Submitted to the

Faculty of the School of Engineering and Applied Science
University of Virginia • Charlottesville, Virginia

In Partial Fulfillment of the Requirements of the Degree
Bachelor of Science, School of Engineering

Amanda Rein
Fall, 2020

Technical Project Team Members

Andy Hui
Greg Vavoso
Jack Shefer
Matthew Bain

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

Introduction

1 in 3 patients with rare genetic disorders wait at least two years to receive a diagnosis according to The Genetic Alliance UK. A 2019 study in the United States and United Kingdom estimated that it takes even longer, between 5.6 to 7.6 years on average. Physicians simply cannot keep up with the 6,000 and counting rare genetic disorders, despite many of them possessing characteristic facial cues (Vandeborne et al., 2019). A mobile application called Face2Gene is utilizing facial recognition technology to drastically reduce the amount of time it takes for physicians to diagnose these rare disorders. Not only does this application of facial recognition save physicians time; patients and their loved ones also save enormous amounts of time, and money, that would have been spent traveling to specialists and undergoing expensive testing. This sociotechnical project will analyze Face2Gene as a technology that has been shaped by societal needs, rather than shaping society itself.

Automation technology is being applied to save time in other areas of healthcare, as well. During the COVID-19 pandemic, individuals not trained in medical professions have had to donate their time to enforcing public health policies. Office managers, security guards, and retail workers are just a few types of people that are manning doorways to scan entrants' temperatures in an attempt to prevent a contagious individual from infecting the space. Automation technology in the form of this proposed Capstone project will free up these individuals' time by automating the process of scanning a person's temperature and controlling their access to the space accordingly.

Technical Topic (Capstone)

The goal of this technical project is to help combat the spread of the novel coronavirus (COVID-19), a deadly illness with over eight million reported cases and more than two hundred thousand deaths in 2020 (CDC, 2020b). As economies start to reopen, concern for public health has given rise to the demand for technology that automates finding and monitoring cases (Morrissey, 2020). Contact tracing mobile applications have emerged to try and meet this need (COVIDWISE, 2020). These applications keep track of nearby devices in order to build network of which users have been in close proximity to others. Thus, when a user tests positive, they can privately and anonymously notify all other devices (a.k.a. users) that were nearby. However, these technologies are limited by the number of users they can attract and the willingness of these users to report positive test results. One of the major goals of this project is to create a technology that does not rely on user motivation. Rather than having users actively enroll in public safety measures, the default state should be participation.

There are many stakeholders in this project, incentivized to ensure public safety and automate enforcement of these measures. Brick-and-mortar businesses, such as restaurants and retail stores, are likely candidates. Their motivations are two-fold. First, in order for customers to return, they must feel safe; therefore, increasing safety steps may increase in-person traffic and subsequently profit. Second, business owners are willing to implement these safety measures to protect themselves and their employees from illness and potential legal liability (Elejalde-Ruiz, 2020). This growing desire for new safety measures applies to office spaces, as well, as employers look to keep themselves and their employees healthy. Additionally, since these brick-and-mortar businesses and office spaces are often privately-owned businesses, they are free to enforce limits on who can enter their property. Other possible stakeholders are healthcare centers and educational institutions. Healthcare centers, such as the UVa Elson Student Health Center,

have been asking visitors to wait outside until someone arrives to manually measure their temperature before admittance. This process of waiting to be scanned before entering relies upon visitors having the patience to wait outside, rather than entering through one of the other entrances. This is an inefficient system, especially for the staff of the healthcare center who must constantly operate the entrance.

This project will prototype an automated temperature screening system to control who can enter a space. This system will be attached to a door-frame. Visitors will approach door and scan their temperatures by holding their wrists under a non-contact temperature sensor. If their temperature is within a predetermined bound, the user will be admitted to the facility; if not, the door will be automatically locked to prevent their entrance. Their temperature reading will be sent via Bluetooth to a server that will store the reading. This collection of stored data will be rendered on a web application that tracks usage and temperature measurements of people attempting to enter the space through that doorway, enabling the owners of the space (and other stakeholders) to have an overall idea of the health of its visitors. Since this web application will display the number of people that have been admitted to the space, it may also be helpful for determining the number of people in the space at a particular time (i.e. for students wanting to avoid peak times at the gyms or libraries). While temperature is only one indicator of a person's wellbeing, it is still one of the most powerful tools for detecting if someone carries a communicable disease, making this system a highly effective way to automate screening the health of individuals before they are admitted to a space (CDC, 2020a).

STS Topic

Over 300 million people currently live with a rare genetic disorder. While this statistic only amounts to approximately 5% of the world population, these disorders significantly reduce the

quality of life for hundreds of millions of people. Diagnosis of these disorders is incredibly difficult due to their uncommon nature, as well as their deceptively-common symptoms (Nguengang et al., 2020). Patients with rare genetic disorders do not receive a proper diagnosis for several years on average, as physicians cannot maintain a knowledge of the symptoms associated with the ever-growing number of rare disorders (Evans, 2018). As such, it is not just patients and their loved ones that are stakeholders in the development of faster, more accurate ways to diagnose rare genetic disorders; medical professionals are also eager to correctly and efficiently diagnose their patients' disorders. While genetic testing is an option for diagnosing genetic disorders, it is very expensive and time-consuming, especially without an idea of the few most likely culprits. There are rare disorder search engines on the Internet that aid in the diagnosis process, as well, but they rely upon the physician's ability to detect all symptoms accurately.

Recent advances in facial recognition technology have brought great hope to increasing the speed and accuracy of diagnosis of these rare genetic disorders. While 30 to 40% of genetic disorders involve detectable changes to the face or skull, these facial cues may be subtle and not well-known to medical professionals (*Medical products: Facial recognition technology to diagnose rare genetic diseases*, 2015). One particular application, developed in 2016, has become incredibly useful in detecting these facial cues and subsequently suggesting likely diagnoses: Face2Gene. This application has been trained on tens of thousands of case files, growing even more accurate every time that it is used due to the nature of computer vision models (Wallner, 2017).

The adoption of Face2Gene is an example of social construction of technology (SCOT) as a technology that has been shaped by its users. SCOT is a popular STS framework which posits

that human action shapes technology. The framework was designed with four key pillars: interpretive flexibility, relevant social group, closure and stabilization, and wider context. SCOT is critiqued by a variety of scholars, including its original authors, for its grouping of society into “relevant social groups” that are assumed to be of equal importance and to have equal voice in the design process. Authors of SCOT have attempted to address these critiques of the framework being overly simplistic, adding another major tenant to SCOT: technological frame. While SCOT is still an imperfect framework, it provides for a rich analysis of the rapid growth of Face2Gene (Klein and Kleinman, 2002).

In summation, this paper will explain how Face2Gene’s development and adoption by the medical community aligns with the major components of SCOT: interpretive flexibility, relevant social group, closure and stabilization, wider context, and technological frame. By exploring these components in the context of Face2Gene, it will become clear how human action has shaped this technology.

Research Question and Methods

Research Question: “How has the facial recognition technology Face2Gene improved diagnosis of rare genetic disorders?”

This research question will be answered using an STS framework entitled social construction of technology (SCOT). First, the research question will be framed with the appropriate background information about the challenges of diagnosing rare genetic disorders, as well as the spread of facial recognition technology as a whole. After that, an explanation will be provided on how the adoption of the Face2Gene application is an example of social construction of technology. To do a full SCOT analysis, it will be necessary to first define the relevant social groups: patients with rare genetic disorders and their loved ones, medical professionals searching

for diagnoses for these patients, and the scientists responsible for creation of Face2Gene. Then, the analysis will launch into how the application's development and adoption fits into the main pillars of SCOT: interpretive flexibility, relevant social groups, closure and stabilization, wider context, and technological frame.

For interpretive flexibility, it will be necessary to include a timeline for the development and adoption of facial recognition as a whole, to point out how facial recognition has been around for decades and yet has only applied to diagnosing rare genetic disorders in the past few years (“A brief history of facial recognition”, 2020). Interpretive flexibility also involves *how* relevant social groups assign meaning to the technology. Addressing this component of interpretive flexibility will require an overview of general sentiment around Face2Gene, as well as the ways in which it has supported its relevant social groups. This will necessitate the use of sources like news and magazine articles, or even quotations from those that have benefited from the application (Etter, 2019) (Grifantini, 2020) (Mjoseth, 2017) (Vincent, 2019).

To address relevant social group, motivations of the social groups defined previously will be outlined, in order to show how they align with the adoption of Face2Gene. Again, this connection requires a general idea of the attitude around Face2Gene rather than sources that provide quantitative data (Etter, 2019) (Grifantini, 2020) (Mjoseth, 2017) (Vincent, 2019).

To address closure and stabilization, it will be important to note how Face2Gene received HIPAA and EU data privacy compliance via sources published by the founders of Face2Gene. Additionally, Face2Gene's method of “de-identifying” the facial photos in their data set in order to maintain individuals' privacy will be highlighted to make it clear how Face2Gene received acceptance by relevant social groups and therefore was able to be adopted by the medical community (“Data Sharing & Protection Policy, 2017) (Martinez-Martin, 2019).

For the wider context, relevant sociocultural and political background at the time that Face2Gene was being released will be laid out. This part of the analysis will include background from news articles and blog posts about the climate surrounding Face2Gene. Most importantly, the analysis of Face2Gene as a form of SCOT will be solidified by case analyses of how the application has been used to aid in diagnosis of a wide variety of genetic disorders, extending beyond its original intent and abilities. A multitude of studies published in medical journals regarding the utilization of Face2Gene to aid in diagnosis of rare genetic disorders have already been collected (Basel-Vanagaite et al., 2016) (Jin et. al, 2020) (Kosilek et al., 2015) (Liehr et al., 2018) (Nguengang et al., 2020) (Stephen et al., 2017) (Valentine et al., 2017).

Finally, resources that highlight the launch and marketing strategy of Face2Gene will detail the technological frame. Including this particular pillar of SCOT will mitigate criticisms of the framework by providing additional detail about the technological strategy utilized by Face2Gene's founders that enabled its success ("2017 Year in Review", 2018) ("2019 Highlights", 2020) ("FDNA and Microsoft Collaborate to Enhance Genomics Technology Through AI", 2018) ("GDNA and PerkinElmer Announce Collaboration to Offer Enhanced Genetic Testing Augmented by AI and Facial Analysis", 2019) ("FDNA Partners with Blueprint Genetics to Spotlight RASopathies During the Year of Discovery", 2017) (Wallner, 2017) ("What to do and See at ACMG 2017 in Phoenix, AZ", 2017) ("What to Do and See at ESHG 2017 in Copenhagen", 2017). Through the analysis of Face2Gene from each of the pillars of SCOT, it will be clear that Face2Gene has been shaped by its relevant social groups.

Conclusion

These two projects address critical applications of technology in healthcare, the first an analysis of facial recognition as a tool to aid in diagnosing rare disorders and the second a

prototype for an automated door locking system that prevents people from entering a room if their body temperature is irregular. The analysis of a facial recognition mobile application that aids in diagnosing rare genetic disorders focuses on a technology that provides answers to millions of people suffering from undiagnosed (and therefore untreated) disorders. The analysis of Face2Gene provides evidence for how technology can be shaped by society to fit its needs. Similarly, the team creating the door locking system will prioritize mitigating public health concerns, in this case focused on the current COVID-19 pandemic. The invention will ensure that indoor spaces do not include individuals with high body temperatures, as high fevers are often indicative of infectious diseases such as COVID-19. In summation, both of the endeavors included in this portfolio focus on technology as a means to improve global public health.

References

Capstone

CDC. (2020a, February 11). *Coronavirus Disease 2019 (COVID-19)*. Centers for Disease Control and Prevention. <https://www.cdc.gov/coronavirus/2019-ncov/hcp/guidance-risk-assessment-hcp.html>

CDC. (2020b, March 28). *COVID-19 Cases, Deaths, and Trends in the US | CDC COVID Data Tracker*. Centers for Disease Control and Prevention. <https://covid.cdc.gov/covid-data-tracker>

COVIDWISE. (2020). Virginia Department of Health. <https://www.vdh.virginia.gov/covidwise/>

Elejalde-Ruiz, A. (2020, May 4). If you get sick with COVID-19, is your employer liable? As businesses prepare to reopen, worker safety is a priority. *Chicago Tribune*.

<https://www.chicagotribune.com/coronavirus/ct-coronavirus-employer-liability-workplace-exposure-20200501-dye6husnszchpnpaadiensn2ja-story.html>

Morrissey, J. (2020, June 16). Fighting the Coronavirus With Innovative Tech. *The New York Times*. <https://www.nytimes.com/2020/06/16/business/fighting-covid-19-innovative-tech.html>

STS

2017 Year in Review. (2018, January 31). <https://www.fdna.com/blog/2017-year-in-review/>

2019 Highlights. (2020, January 20). *FDNA*. <https://www.fdna.com/blog/2019-highlights/>

A brief history of facial recognition - NEC New Zealand. (2020, May 26). *NEC*.

<https://www.nec.co.nz/market-leadership/publications-media/a-brief-history-of-facial-recognition/>

Basel-Vanagaite, L., Wolf, L., Orin, M., Larizza, L., Gervasini, C., Krantz, I. D., & Deardoff, M.

A. (2016). Recognition of the Cornelia de Lange syndrome phenotype with facial

dysmorphology novel analysis. *Clinical Genetics*, 89(5), 557–563.

<https://doi.org/10.1111/cge.12716>

Data Sharing & Protection Policy. (2017, March 28). *Face2Gene*.

<https://www.face2gene.com/data-sharing-protection-policy/>

Etter, N. S. (2019, April 9). *Facial recognition and the future of diagnosis*. Children's Wisconsin.

<https://childrenswi.org/newshub/stories/face2gene-facial-recognition-diagnosis>

Evans, W. (2018). Dare to think rare: diagnostic delay and rare diseases. *British Journal of General Practice*, 68, 145. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5916061/>

FDNA and Microsoft Collaborate to Enhance Genomics Technology Through AI. (2018, October 16). *FDNA*. <https://www.fdna.com/blog/fdna-and-microsoft-collaborate-to-enhance-genomics-technology-through-ai/>

FDNA and PerkinElmer Announce Collaboration to Offer Enhanced Genetic Testing Augmented by AI and Facial Analysis. (2019, June 25).

<https://www.businesswire.com/news/home/20190625005184/en/FDNA-and-PerkinElmer-Announce-Collaboration-to-Offer-Enhanced-Genetic-Testing-Augmented-by-AI-and-Facial-Analysis>

FDNA Partners with Blueprint Genetics to Spotlight RASopathies During the Year of Discovery. (2017, February 15). *FDNA*. <https://www.fdna.com/blog/rare-diseases-blog-post/>

FDNA Presents Rare Disease Technologies at the 2017 Precision Medicine Summit at Boston Children's Hospital. (2017, September 18). *FDNA*. <https://www.fdna.com/blog/fdna-presents-rare-disease-technologies-2017-precision-medicine-summit-boston-childrens-hospital/>

- Grifantini, K. (2020, May 13). Detecting Faces, Saving Lives: How facial recognition software is changing health care. *IEEE Pulse, March/April 2020*.
<https://www.embs.org/pulse/articles/detecting-faces-saving-lives/>
- Jin, B., Qu, Y., Zhang, L., & Gao, Z. (2020). Diagnosing Parkinson Disease Through Facial Expression Recognition: Video Analysis. *Journal of Medical Internet Research, 22*(7).
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7382014/>
- Klein, H. K., & Kleinman, D. L. (2002). The Social Construction of Technology: Structural Considerations. *Science, Technology, & Human Values, 27*(1), 28–52.
- Kosilek, R. P., Frohner, R., Wurtz, R. P., Berr, C. M., Schopohl, J., Reincke, M., & Schneider, H. J. (2015). Diagnostic use of facial image analysis software in endocrine and genetic disorders: review, current results, and future perspectives. *European Society of Endocrinology, 173*(4).
<https://ej.e.bioscientifica.com/downloadpdf/journals/eje/173/4/M39.pdf>
- Liehr, T., Acquarola, N., Pyle, K., St-Pierre, S., Rinholm, M., Bar, O., Wilhelm, K., & Schreyer, I. (2018). Next generation phenotyping in Emanuel and Pallister-Killian syndrome using computer-aided facial dysmorphology analysis of 2D photos. *Clinical Genetics, 93*(2), 378–381.
<https://doi.org/10.1111/cge.13087>
- Lumaka, A., Cosemans, N., Mampasi, A. L., Mubungu, G., Mvuama, N., Lubala, T., Mbuyi-Musanzayi, S., Breckpot, J., Holvoet, M., Ravel, T. de, Buggenhout, G. V., Peeters, H., Donnai, D., Mutesa, L., Verloes, A., Tshilobo, P. L., & Devriendt, K. (2017). Facial dysmorphism is influenced by ethnic background of the patient and of the evaluator. *Clinical Genetics, 92*(2), 166–171. <https://doi.org/10.1111/cge.12948>

- Martinez-Martin, N. (2019). What Are Important Ethical Implications of Using Facial Recognition Technology in Health Care? *AMA Journal of Ethics*, 21(2), 180–187.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6634990/>
- Medical products: Facial recognition technology to diagnose rare genetic diseases.* (2015, December 1). UK Research and Innovation. <https://mrc.ukri.org/news/browse/medical-products-facial-recognition-technology-to-diagnose-rare-genetic-diseases/>
- Mjoseth, J. (2017, March 23). *Facial recognition software helps diagnose rare genetic disease.* National Human Genome Research Institute. <https://www.genome.gov/news/news-release/Facial-recognition-software-helps-diagnose-rare-genetic-disease>
- Nguengang Wakap, S., Lambert, D. M., Olry, A., Rodwell, C., Gueydan, C., Lanneau, V., Murphy, D., Le Cam, Y., & Rath, A. (2020). Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *European Journal of Human Genetics*, 28(2), 165–173.
<https://doi.org/10.1038/s41431-019-0508-0>
- Stephen, I., Hiew, V., Coetzee, V., Tiddeman, B., & Perrett, D. (2017). Facial Shape Analysis Identifies Valid Cues to Aspects of Physiological Health in Caucasian, Asian, and African Populations. *Frontiers in Psychology*, 8, 1883.
<https://www.frontiersin.org/articles/10.3389/fpsyg.2017.01883/full>
- Valentine, M., Bihm, D. C. J., Wolf, L., Hoyme, H. E., May, P. A., Buckley, D., Kalberg, W., & Abdul-Rahman, O. A. (2017). Computer-Aided Recognition of Facial Attributes for Fetal Alcohol Spectrum Disorders. *Pediatrics*, 140(6). <https://doi.org/10.1542/peds.2016-2028>
- Vandeborne, L., van Overbeeke, E., Dooms, M., De Beleyr, B., & Huys, I. (2019). Information needs of physicians regarding the diagnosis of rare diseases: a questionnaire-based study in

Belgium. *Orphanet Journal of Rare Diseases*, 14(1), 99. <https://doi.org/10.1186/s13023-019-1075-8>

Vincent, J. (2019, January 15). *Facial recognition and AI could be used to identify rare genetic disorders*. The Verge. <https://www.theverge.com/2019/1/15/18183779/facial-recognition-ai-algorithms-detect-rare-genetic-disorder-fdna>

Wallner, S. (2017, October 3). *Precision Medicine Startup FDNA Introduces Next-Generation Phenotyping (NGP)*. <https://magazine.startus.cc/precision-medicine-startup-fdna-introduces-next-generation-phenotyping-ngp/>

What to Do and See at ACMG 2017 in Phoenix, AZ. (2017, February 15). *FDNA*. <https://www.fdna.com/blog/events-blog-post/>

What to Do and See at ESHG 2017 in Copenhagen. (2017, May 22). *FDNA*. <https://www.fdna.com/blog/what-to-do-and-see-at-eshg/>