Developing a Blink-Based Communication System for ALS Patients

Investigating the Ethics and Morals of Regulating Genetic Data Privacy

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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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Introduction

Neurodegenerative diseases emerge when nerve cells in the brain and nervous system deteriorate and lose function. *Amyotrophic Lateral Sclerosis* (ALS), more commonly known as Lou Gehrig's Disease, is a progressive neurodegenerative disease affecting motor neurons located in the brain and spinal cord. ALS is characterized as a fatal neurological disease, and progressive paralysis commonly leads to death from respiratory failure within three to five years of symptom onset (Saez-Atienzar et al., 2021).

There exists no general consensus on a definitive etiological causation for ALS. However, over the last 50 years the rapid expansion of the genomic library has shed some light on potential genetic factors that might predispose an individual to developing the disease. Several independent studies have identified one commonality existing in the genetic makeup of ALS patients — a hexanucleotide repeat expansion mutation occurring on the *C9ORF72* gene (van Blitterswijk et al., 2012). Additionally, more than 50 modifying genes have been identified as possessing dangerous pathogenic variants capable of contributing to the development of ALS. However, of these identified genes more than 70% of both sporadic and familial occurrences of ALS remain unexplained (Mejzini et al., 2019; Renton et al., 2014).

ALS affects a large amount of people across the world, however prevalence in the United States is significantly higher than our European counterparts. A survey done by the National ALS Registry found that nearly 7 out of every 100,000 people in the United States will be diagnosed with ALS, with white males being nearly twice as likely to develop the disease (Mehta et al., 2023). this disease causes patients to lose the ability to initiate and control voluntary muscle movements. Bulbar neurons, those controlling common lower jaw movements such as swallowing and speech, become increasingly deteriorated in the mid and late stages of ALS and
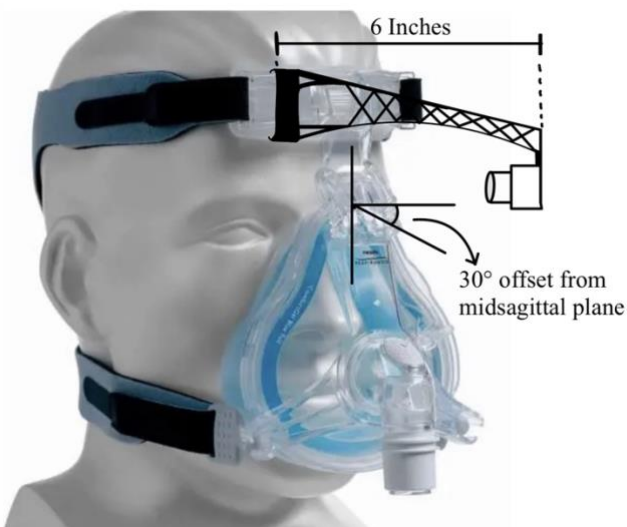
can greatly reduce the ability of affected patients to communicate effectively (Moawad, 2022). As the ability to speak decreases, the role of a caretaker becomes more and more necessary to maintain an acceptable quality of life. The Zarit Burden Interview (ZBI) was developed as a way to quantify the amount of burden experienced by caretakers when providing physical, emotional, and financial care to patients. In a study published by the Journal of Neurology, a strong correlation was found between the progression of ALS (quantified by the number of health domains for which a patient has lost independence) and the ZBI score reported by the caregiver (Burke et al., 2018). While the serious physical health hurdles that must be traversed every day by people living with ALS are very real, the responsibilities and time invested by caretakers are also equally as real. This technical project focuses on the integration of an open-source blink detection algorithm with a BiPAP mask camera to develop an effective and modular communication system that will increase the autonomy of ALS patients and decrease caretaker burden.

Blink-Based Communication System

The ability to communicate is of vital importance to people living with ALS, as it serves as a lifeline connecting them with loved ones, caregivers, and healthcare professionals. It enables them to express their needs, emotions, and preferences about important medical and financial decisions, and maintain a sense of autonomy and quality of life amidst the physical challenges posed by the disease. There are a wide variety of augmentative and alternative communication (AAC) methods available to ALS patients, with the complexity of the devices progressing alongside the progression of the disease. During early onset of symptoms, low-tech AAC systems such as picture boards rely on patients to select images or phrases to communicate, and

some digital devices can convey these user inputs via prerecorded voice output messages. As the disease progresses and the ability to speak and move freely further deteriorates, more sophisticated speech-generating devices (SGDs) are employed to communicate more complex phrases and allow users to control their external environment (Brownlee & Bruening, 2012). Nevertheless, these communication devices maintain their reliance on having a physical caretaker present in the room to act on the needs of patients. Additionally, the decrease in dexterity and range of motion experienced during the later stages of ALS can cause these methods to become less practical. One emerging field of research to address this discrepancy is oculomotor function, which helps to adjust and coordinate eye position during movement (Joyce et al., 2023).

In order to bridge the gap between the ability of a person living with ALS to communicate and the necessity of having a present and engaged caretaker in the room, this technical project will utilize an open-source, Python-based blink detection algorithm, which will be integrated with a small Raspberry Pi camera to detect voluntary versus nonvoluntary blinking. A lightweight 3D-printed mount will be designed for attaching the camera onto BiPAP masks, which are worn by patients to provide ventilatory support and improve sleep quality. Figure 1 shows a schematic illustrating the BiPAP mask and how the camera will be mounted, including the distance from the mask to the camera as well as the angle of observation of the camera. This communication system will be

Figure 1: BiPAP Camera Mount Schematic (Caylor, 2023)

implemented primarily during the later hours of the day, especially at night while the user is sleeping. If the patient needs something urgently, three quick and prominent blinks will trigger the system to alert the caretaker that the person is in need. The primary inconvenience that this technology will alleviate is the need for a caretaker to be present in the same room as the patient at all hours. The combination of mounted cameras with blink-detection will improve the quality of life for both patients and their caregivers, and give people with ALS a new freedom, the ability to promptly signal for help in real time should they need assistance.

Employment of cameras in the medical field, such as with blink-detection technologies for eye tracking or motion capture cameras for range of motion assessments in physical therapy, have brought significant improvements to treatment approaches and quality of patient life. However, many people are hesitant to sacrifice privacy in their life to any infrastructure so closely intertwined with the government. Another example of these medical privacy concerns has arisen with the recent development of gene editing technologies. The Journal of American Medical Informatics Association even published a 2014 study in which more than 83% of participants cited concerns that their genetic data should be protected (Rogith et al., 2014). As technological access to private genomic data becomes more pertinent to medical procedures, more discussions are taking place about the ethical regulation of genetic data privacy.

ETHICS AND MORALS OF REGULATING GENETIC DATA PRIVACY

As the industry nears 22 billion dollars in value by 2030 (Kumar, 2022), citizens fear the privacy of their genetic data may become increasingly vulnerable. The complex network of the healthcare industry is comprised of a variety of stakeholders. Patients—individuals seeking genetic treatment—are the direct stakeholders associated with the industry. Healthcare providers

comprise a second stakeholder group, as they deliver the treatment to patients. On the opposing team, insurers are stakeholders who determine coverage and reimbursement policies. The relationship between patients and insurers has perpetuated healthcare inequities and continues to do so with the emergence of gene editing and its high out-of-pocket treatment costs. Lawmakers make up a separate stakeholder group that passes legislation to protect individual rights. The Genetic Information Nondiscrimination Act (GINA), passed by Congress in 2008 as a federal law prohibiting discrimination by employers and health insurers based on an individual's genetic information, was one of the first milestone legislative actions taken to address access to genetic information (Feldman, 2012). The interconnectedness of these stakeholder groups, all working in the relatively uncharted grounds of gene editing, represent an important portion of the American healthcare system centered around genetic data, privacy, and discrimination.

Infrastructure is the organization and interactions of both human and nonhuman components of a system that contribute to effective functionality. In complex system such as the U.S. healthcare industry, several main elements of infrastructure can explain how humans and technology are interconnected (Star, 1999). Elements of a system can *be learned as part of membership*, meaning in order for one to understand the organizational framework of an aspect of a system, they must be naturalized as part of membership in some group. Individuals electing to undergo gene editing treatment are inherently aware of the resulting availability of their genome in the healthcare industry. However, given the hereditary nature of genomics, it is important to consider close family members as having membership in the system as well. George J. Annas, Director of the Center for Health Law, Ethics & Human Rights at the Boston University School of Public Health, published a book highlighting the discrepancies that might arise within families about how genetic information is distributed and accessed. He argues that

overlap in genomic data between family members give merit to including all family members in making decisions regarding genetic data privacy (Annas, 1999). *Scope,* the broad reach that infrastructure has on surrounding individuals and systems, is another facet of infrastructure that impacts how genetic information is collected, used, and distributed (Star, 1999). The scope of genetic data privacy is broad and encompasses access to genetic information by third parties. However, some industry professionals believe this scope might be beneficial in the protection of genetic data. Patricia Roche of the Health Law Department at the Boston University School of Public Health published a journal article proposing the utilization of a third party to collect DNA samples. For-profit companies will hire private genetic information mediators to obtain informed consent of the individual to codify the data by taking out some genetic markers and then distribute the data to researchers (Roche & Annas, 2001). The purpose of this system would be to ensure no genetic information is being used or distributed without the informed consent of the individual.

RESEARCH QUESTION AND METHODS

The scope of gene editing expands far beyond the hospital room, encompassing ethical questions about gene editing in fetuses and even implications in insurance discrimination and genetic warfare. These considerations are shaping how the gene editing industry has grown and will continue to grow. However, it is important to answer one pertinent question to guide the growth of genetic engineering technology: How can we mitigate genetic data privacy threats through ethical infrastructure and legislation?

The ethics of legislation possess a strong correlation to individual rights and liberties. Laws passed should reflect the overarching values and morals of the general public. Thus, to

address this question I plan on conducting surveys of pertinent stakeholder groups including private individuals and healthcare professionals. The survey will consist of a series of ethical questions such as: Do individual family members have the right to distribute hereditary genetic information? Should there be differentiation in privacy legislation based on the type of information (e.g., medical versus ancestral data)? Who should hold the liability for misuse of private genetic information? There will also be additional numerical response questions utilizing the Likert scale such as: On a scale of 0-7 how protected do you think genetic data privacy is?

This survey will be conducted both on grounds via student and faculty responses as well as at the hospital via healthcare professionals and patients. The numerical data will be analyzed to identify general opinions on pertinent health topics, and the short response questions will be evaluated qualitatively to identify recurring important themes about hesitations, concerns, and values of the participants. Kirsten Riggan and her colleagues from the Mayo Clinic Biomedical Ethics Research Program conducted a similar survey regarding ethical uses of genetic engineering, whose framework for data analysis will be utilized to review each survey transcript (Riggan et al., 2019).

CONCLUSION

The burden taken on by caretakers can lead to serious mental and physical setbacks including exhaustion, loss of self-identity, and even substance abuse (McAtee et al., 2021). The lack of individual autonomy for people living with ALS is a very serious difficulty, and can foster a negative patient-caregiver relationship built upon dependance (Tramonti et al., 2012). The development of a blink-based communication system will foster a safe environment to enhance patient freedom and distance the caregiver from the burden of their necessity.

As genetic engineering becomes increasingly popular in medical treatments, the implementation of ethical legislation protecting genetic data becomes paramount. Gaining an understanding about the morals and values of private individuals will ensure that legislation passed accurately reflects and protects individual liberties. Both qualitative and quantitative research will provide a variety of perspectives from pertinent stakeholders involved in the genetic engineering industry.

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