Genome-wide association meta-analysis identifies critical regulators of immune dysfunction and cell stress pathways driving cardiovascular disease and systemic lupus erythematosus
(Technical Report)

The Polarization of Responses to The First Human Clinical Trial Involving Gene Therapy
(STS Research Paper)

An Undergraduate Thesis Portfolio

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Socio-Technical Synthesis

In 1990 the Human Genome Project was funded with the goal of sequencing the entire human genome in 15 years and was completed two years ahead of schedule. While in its early days there were many critics, in hindsight, the project's overwhelming success made it clear that the benefits greatly outweighed the cost. With technological advancements in genetics, we now know so much more about the human genome and molecular biology. As a result, biomedical research and medicine is heavily relying on genetic information to understand and ultimately treat all types of medical conditions. While many medical professionals are determined to fully master and manipulate the human genome, social restrictions remain a large obstacle to genetic engineering.

Determining the genetic players in disease has become a major focus of biomedical research as it provides insight into the relevant pathophysiological mechanisms and potential areas for targeted drug intervention. Systemic lupus erythematosus (SLE) is an autoimmune disease estimated to affect nearly 1.5 million people in the United States alone. Additionally, premature heart disease is now the leading cause of morbidity and mortality in lupus patients. Unfortunately, the standard-of-care drugs for cardiovascular disease (CVD) are not effective in these patients. Identification of biological pathways contributing to CVD in SLE can provide insight into potential drug targets for improved outcomes. My technical project investigates the shared pathways underlying heart disease in lupus patients, in hopes of identifying existing drugs that can be repurposed for SLE to prevent or treat CVD. Working with AMPEL BioSolutions, I have employed a novel computational approach to map genetic variants associated with SLE and CVD to genes and molecular pathways for drug target identification.

Since the first discussions of genetic therapies in the 20th century, concerns regarding the ethics and role of bioengineering in society have been expressed. Society continues to face a severe contradiction regarding medicine created using genetic engineering techniques that remains an obstacle for the development and use of gene therapies. As such, my STS research evaluates diverse social perspectives on genetic engineering to understand the underlying forces contributing to strong opposition of such technology. More specifically, my paper is focused on the polarization of responses to the first clinical trial involving gene therapy.

Understanding the genetic variants and biological pathways contributing to disease is extremely useful in drug development. By predicting critical genes and pathways involved in the development of heart disease in lupus patients, my technical project serves as a robust discovery method for personalized medicine. While the direct goal of my technical research is insight into plausible targeted-drug treatments, not gene therapies, advancements in genetic sequencing, editing, and analysis techniques have resulted in a shift towards directly manipulating genomic and transcriptomic activity contributing to disease. However, as genetic engineering introduces complicated concerns, it is important to thoroughly evaluate the relevant societal perspectives and opposition towards the emerging technology. As such, my STS research provides a deeper understanding of the societal concerns regarding genetic engineering and offers insight into how to navigate the controversy including important considerations and restrictions for the use of gene therapies.