**Thesis Project Portfolio**

# Improving Fetal Aneuploidy Detection: A Better Bioinformatics Pipeline for Non-invasive Prenatal Testing Analysis

(Technical Report)

# Sociocultural Considerations for Equitable Non-invasive Prenatal Testing Administration

(STS Research Paper)

An Undergraduate Thesis

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**Shruthi Nyshadham**

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Department of Computer Science

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## Sociotechnical Synthesis

Non-invasive prenatal testing (NIPT) has risen in popularity over the last decade as a way to screen for potential fetal aneuploidy early in pregnancy. My technical project and my STS project attempt to improve the current state of NIPT from two different approaches. My technical research directly works on improving the existing NIPT bioinformatics algorithms, by improving its function in situations where it currently does not produce accurate results. My STS project, on the other hand, aims to better characterize the economic and cultural barriers that have led to decreased NIPT use among underserved patient populations in the United States, as well as lay out important considerations for equitable NIPT use. This approaches the improvement of NIPT not from a purely technological perspective, but instead from a social one, looking at how the structures and systems in place around the technology function, as well as where they struggle. The combined focus of both of these projects is ultimately to pave the way for increased access to accurate NIPT testing among all pregnant people who wish to learn about the genomic health of their developing baby.

The aim of my technical research project was to assign every genomic site a fetal probability score (FPS), using a combination of computational, statistical, and metagenomic analysis techniques on existing NIPT patient data. This site-specific FPS, which would indicate the likelihood of DNA from a given site being fetal DNA, could then be incorporated into existing NIPT pipelines to artificially enrich FF by giving greater weight to DNA with a higher FPS when predicting fetal aneuploidy. The goal is that this enhanced NIPT algorithm will be able to better predict aneuploidy even in cases of low FF, resulting in fewer no-call results. Cases of low FF are more common in certain already underserved populations, such as patients with higher BMI or patients of color. Therefore, this project ultimately aimed to improve NIPT and expand access to it using a technical approach.

My STS project focused on taking a Social Construction of Technology (SCOT) approach to NIPT, looking at the sociocultural considerations important to keep in mind for the successful and equitable administration of NIPT. I argued that the current integration of NIPT into the prenatal care process is inequitable and improper due to a lack of appropriate consideration for patients' sociocultural backgrounds and medical literacy. I used SCOT to analyze patient, provider, and NIPT company perspectives on NIPT to understand how their differing views on the technology has led to issues with its implementation. Through this analysis, I attempted to highlight how social guidelines and systems must be put in place around the application of the technology to ensure it is used in a way that leads to the greatest benefit, rather than being forced on the unwilling or the uninformed.

Working on better understanding and improving NIPT using both a technical and sociotechnical approach simultaneously gave me a unique perspective on this project I would not have otherwise had. Prior to working on the STS project, I had a very black and white view of NIPT; namely, that the more people using NIPT as part of their prenatal care plan, the better. Further, I strongly believed that the best way to improve NIPT was to continue fine-tuning the technology so it would work for even more patients than it already does. However, the STS project gave me a new perspective, by forcing me to look at not just the technology itself, but the infrastructure around its implementation. I learned that just improving NIPT algorithms isn't enough if underserved populations do not have access to using it, either through physical barriers or through insufficient knowledge of how it works. Working on the STS project at the same time as the technical project helped me see that technology does not exist in a vacuum, and even the best technological innovations are only as good as the systems in which they are implemented.