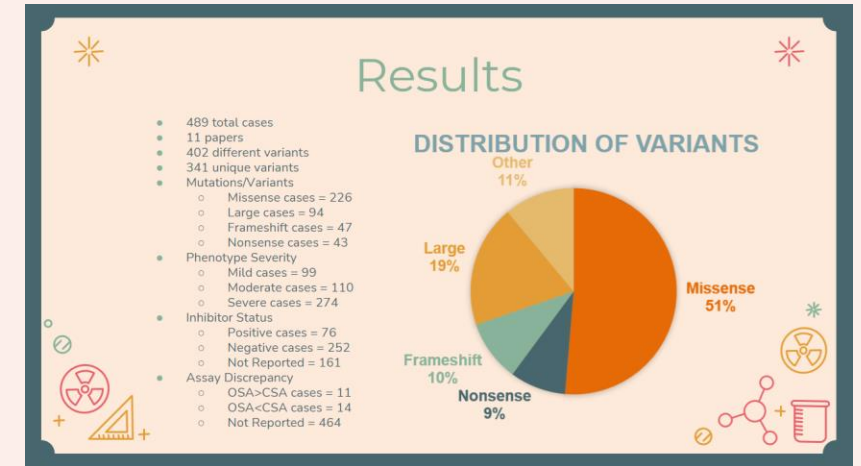


# Variants that cause Hemophilia A and B: An Exploration through Hypothes.is



- Dan Tsuma
- Biology Major
- Lab of Dr. Jonathan Berg | Department of Genetics



# Research Objective

- The objective of my project is to create annotations, through Hypothes.is, for the F8/F9 coagulation factor genes that will aid variant curation and to correlate the type of variant with the severity of Hemophilia A or B, inhibitor status, and assay discrepancy
- This research objective is relevant because annotation is important to variant curation and our understanding of the role of pathogenic variants in disease

# Results

- Results: Missense variants were about 51% of the variants I saw across 489 total cases. Missense variants were commonly associated with mild hemophilia A or B, a negative inhibitor status, and no reported assay discrepancy. Large variants were associated with severe hemophilia A or B, a positive inhibitor status, and no reported assay discrepancy. Finally, both frameshift and nonsense variants were associated with severe hemophilia A or B, a negative inhibitor status, and no reported assay discrepancy
- These results are important because they seem to suggest a correlation between the type of variant and phenotype severity, inhibitor status, and assay discrepancy. With more research, more correlations could be drawn that could help us more effectively treat patients in clinical genetics
- This is relevant not only to the scientific community, but also to the general public as we become more knowledgeable about pathogenic variants and ways to better treat disease