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#### Study on KANSL1 gene mutation and its association to diabetes

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## Study on KANSL1 gene mutation and its association to diabetes Danielle Willis and Elaine Vanterpool, PhD

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## ABSTRACT

•Diabetes is a chronic health condition affecting millions worldwide, it disrupts the body's ability to regulate blood sugar levels effectively. The process begins with the breakdown of food into glucose, which is then released into the bloodstream. Normally, the pancreas responds by secreting insulin, a hormone that facilitates the entry of glucose into cells for energy production. However, in diabetes, this process is compromised. The purpose of this study is to minimize the chances of developing the disease and understand and reduce the mortality of those with the disease. The gene I chose is KANSL1. Simple Clinvar was used to identify genes associated with diabetes. KANSL1 was identified to be associated with this disease. Polyphen 2 sift and swiss model was used to analyze the data. Clinvar determined the protein mapping and pathogenic state of KANSL1 and its mutations. The purpose of this study is to provide research to the medical community and potentially treat this disease.

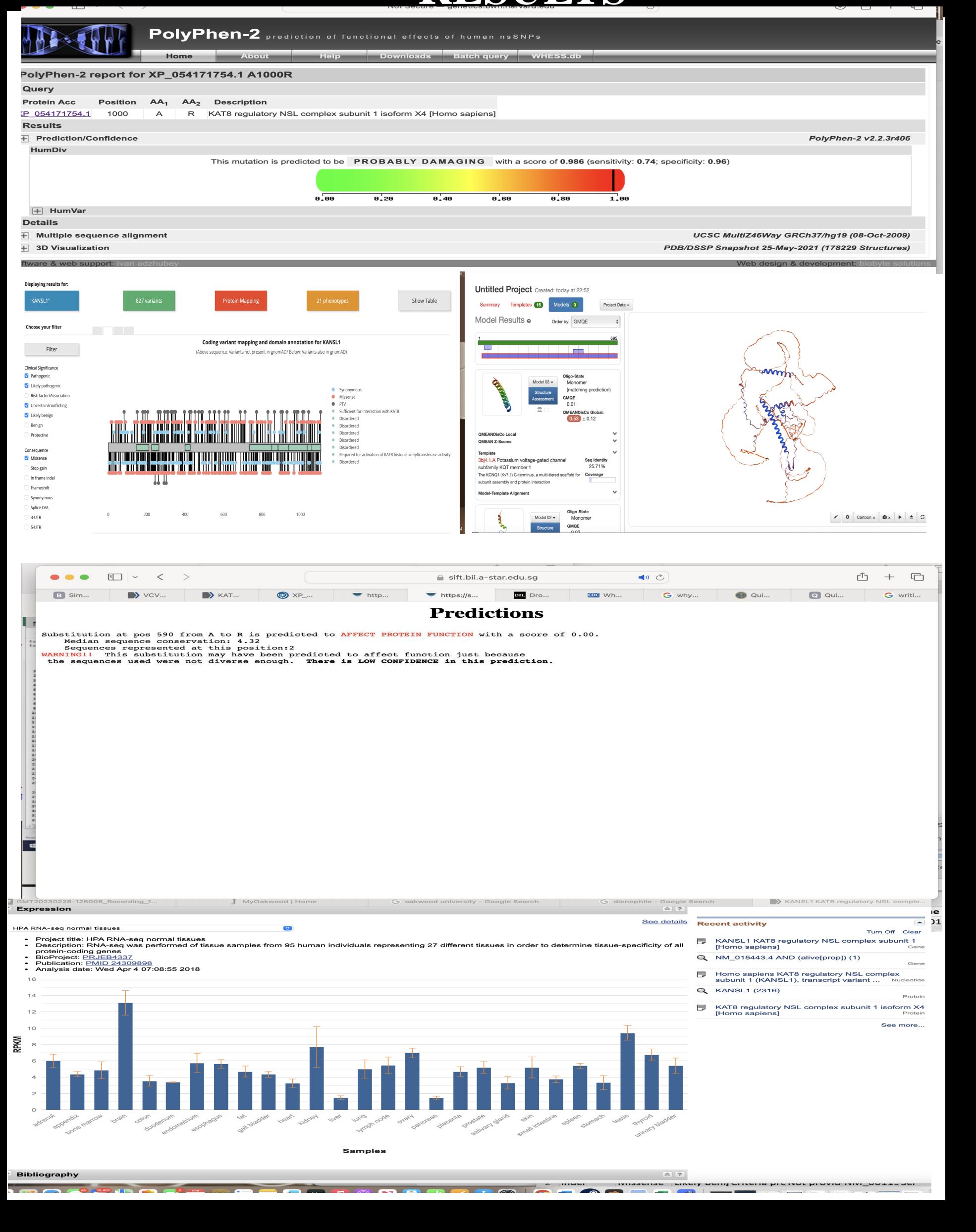
## INTRODUCTION

KANSL1 encodes a nuclear protein that is a subunit of two protein complexes involved with histone acetylation, the MLL1 complex and the NSL1 complex. The encoded protein has been implicated in a variety of cellular processes including enhancer regulation, cell proliferation, and mitosis.

## METHODS

- •Simple Clinvar was used find the gene associated with diabetes
- •Polyphen 2- FATSA sequence from simple clinvar used to analyze variants
- •Sift was used to determine if amino acid would affect proteins function
- •Swiss Modeling was used to develop an 3D model of the structure of the protein.

## RESTIAS



# DISCUSSION AND CONCLUSION

Diabetes is an chronic health condition that disrupts the body's ability to regulate blood sugar levels effectively. The gene I choose KANSL1. This gene was associated with diabetes when put through simple clinvar. The research gathered was able to determine the gene had a mutation and is pathogenic

#### REFERENCES

•Kansl1 Kat8 regulatory NSL complex subunit 1 [homo sapiens (human)] - gene NCBI. (n.d.). Retrieved from https://www.ncbi.nlm.nih.gov/gene/284058
•Simple clinvar. (n.d.). Retrieved from https://simple-clinvar.broadinstitute.org/

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