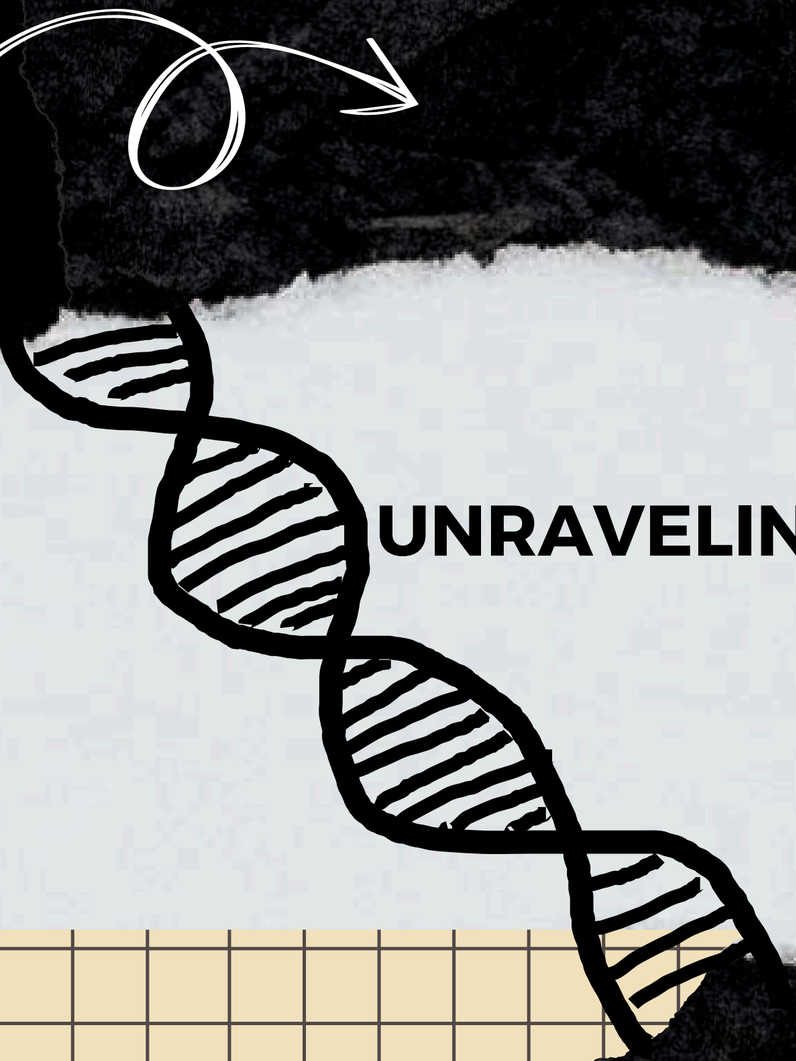
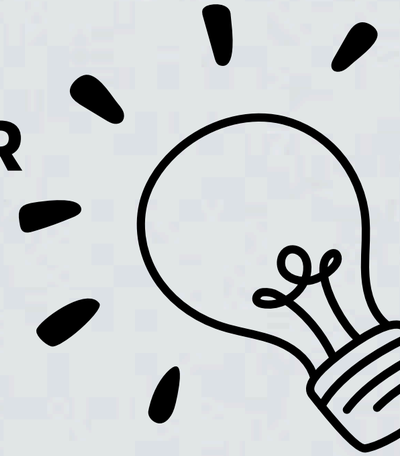
UNRAVELING ASD SUSCEPTIBILITY MARKERS FOR PREDICTIVE DIAGNOSIS

Presented By Group 7





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WHAT IS ASD?

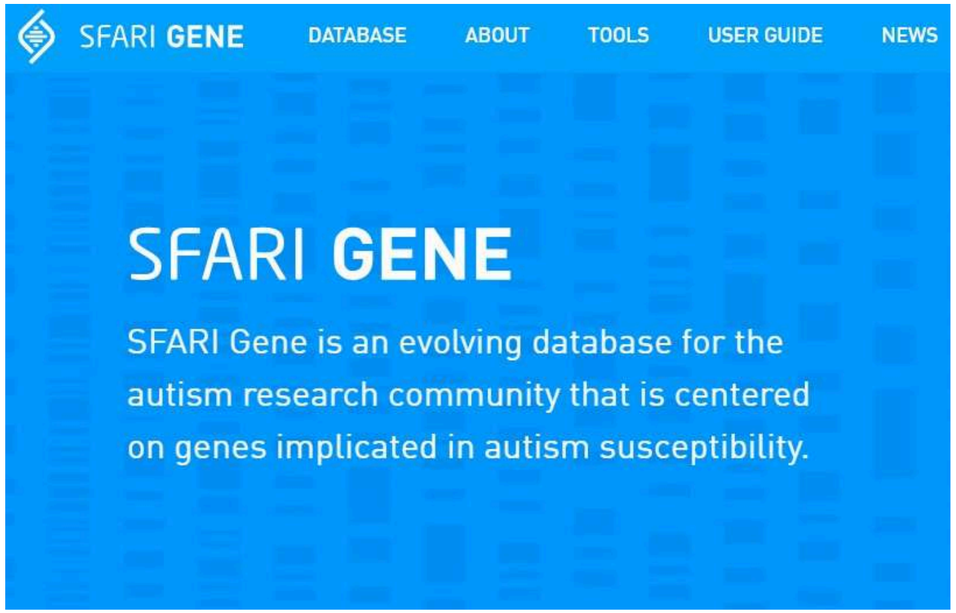
Autism spectrum disorder (ASD) is a neurological and developmental disability caused by differences in the brain. People with ASD often have problems with social communication and interaction and restricted or repetitive behavior. Their symptoms can also affect their ability to function in school, work or other areas of life.

PROBLEM STATEMENT

Our aim is to identify genes whose mutations cause ASD, looking for those mutations and using this mutation data to predict the chances of an unknown sample being affected by ASD.

POSSIBLE SOLUTIONS

We will use the SFARI gene to get the data on genes that are strong contenders for ASD. Using this data, we will identify gene markers and test for those in sample genomes to predict whether this patient is susceptible to having ASD or not.



ANALYSIS METHODS

CNV ANALYSIS

- Analyzing CNVs to obtain insights into structural variations in the genes associated with ASD, identifying genetic variations.
- These variations could help us understand the susceptibility of ASD.

SNP ANALYSIS

- SNP analysis is used as the basis of Genome-wide association studies. SNPs present in genes associated with ASD can provide insight into mutation/variations in that gene, thereby contributing to the prediction of ASD.

WORKFLOW

STEP 1

- Identifying the genes associated with ASD
- (Getting data about the genes from sources like Sfari Gene, also referring to some already existing research papers/articles.)

STEP 2

- Obtain sample genomes of humans to analyze.

STEP 3

- Analysing SNPs and CNVs using sequence analysis methods, microarray analysis, etc, to get insight into those genes in the sample genome to get a predictive verdict.

FEASIBILITY OF PROJECT PLAN

- Availability of data: Some databases like SFARI gene are publicly available, for some other databases which can contribute to extensive study require access, so we are sending out request accesses.

- Resources and tools: We will be using BLAST for sequence alignment and use dbSNP to identify SNPs.

CURRENT PROGRESS

We have identified the following datasets and tools to help us analyze the data:

- SFARI Gene
- MSSNG

As mentioned earlier in our solution, we will be analyzing the data from these sources to find mutations and look for those mutations in a sample genome.

EXISTING RESEARCHES

- Seaver Autism Centre for Research and Treatment
- MIND Institute at University of California
- Centre for Autism Research
- Autism and Developmental Medicine Institute
- John Hopkins University centre for Autism and Developmental DIsabilities

REFERENCES AND DATASETS USED

- https://medlineplus.gov/genetics/condition/autism-spectrum-disorder/#causes
- https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7369758/
- https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6710438/
- https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9923897/
- https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8119484/
- https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3513682/#:~:text=First%2C%20specific%20genetic%20syndromes%20like,involve%20synaptic%20genes%20like%20NLGN3
- https://www.youtube.com/watch?v=b9nCihlhhk0
- https://www.spectrumnews.org/news/autism-genetics-explained/
- https://gene.sfari.org/

THANK YOU