

**TEAM MARAVILLA**

# Single Base Position Mutations in Parkinson's Disease

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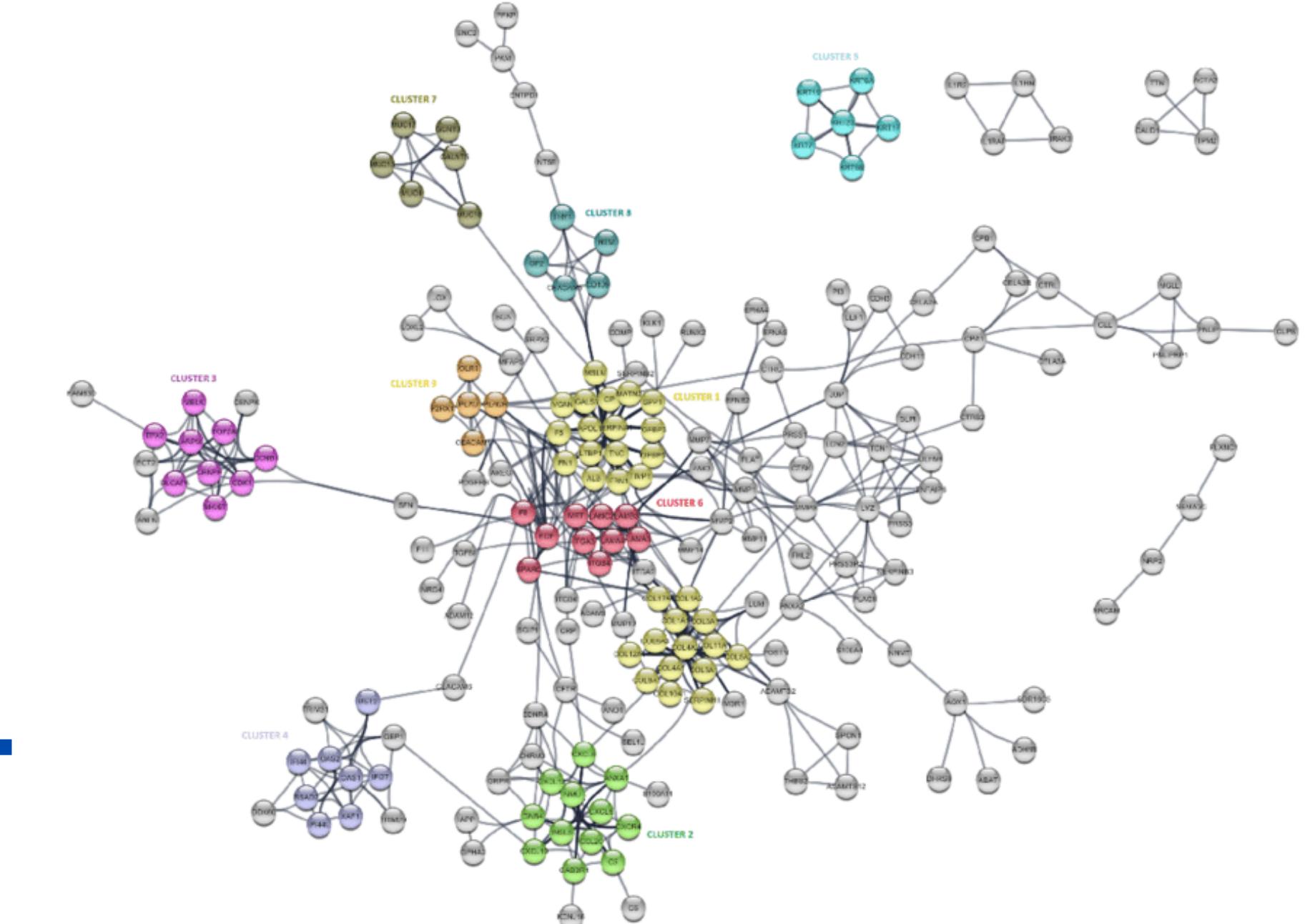
A Very Basic Bioinformatic Analysis

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# Points of Discussion

**These are the broad topics we will cover.**

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01 Parkinson's Disease

02 Single Base Position  
Mutations (SNPs)

03 Protein-Protein  
Interaction Network

04 Materials and  
Methods

# PD

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## Parkinson's Disease



### Definition

Parkinson's disease is a progressive brain disorder that causes nerve cells to break down, leading to movement and mood problems:

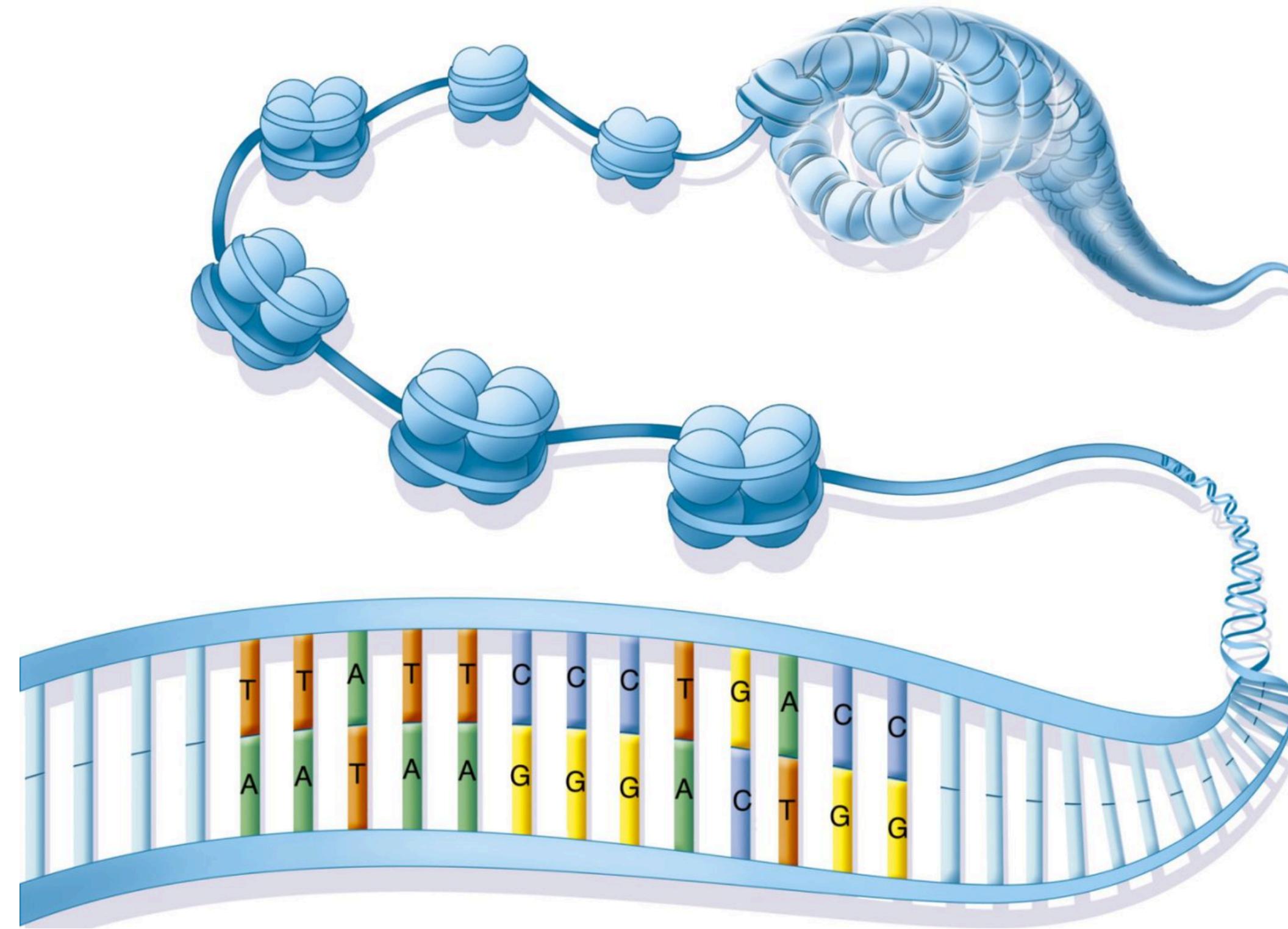


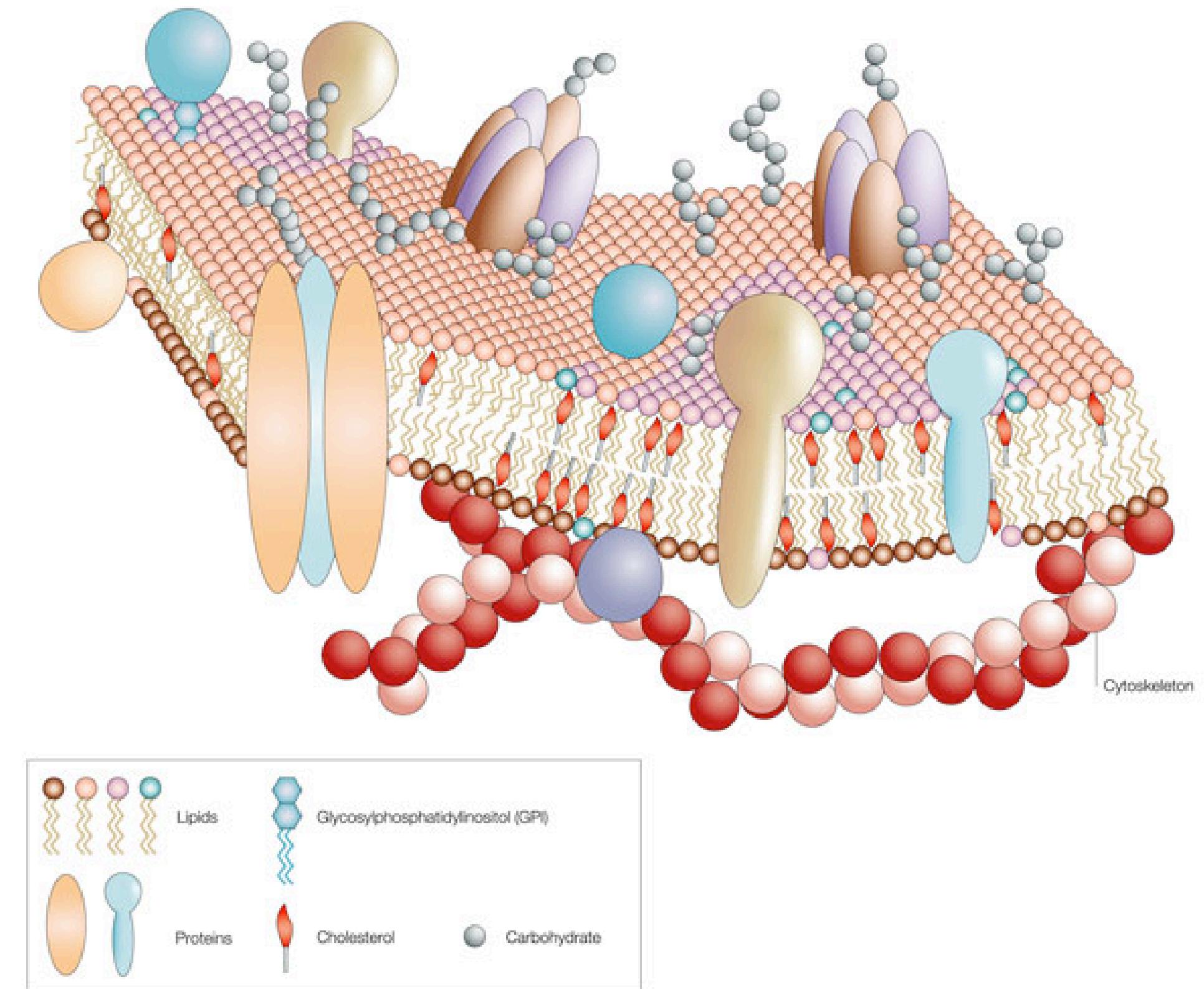
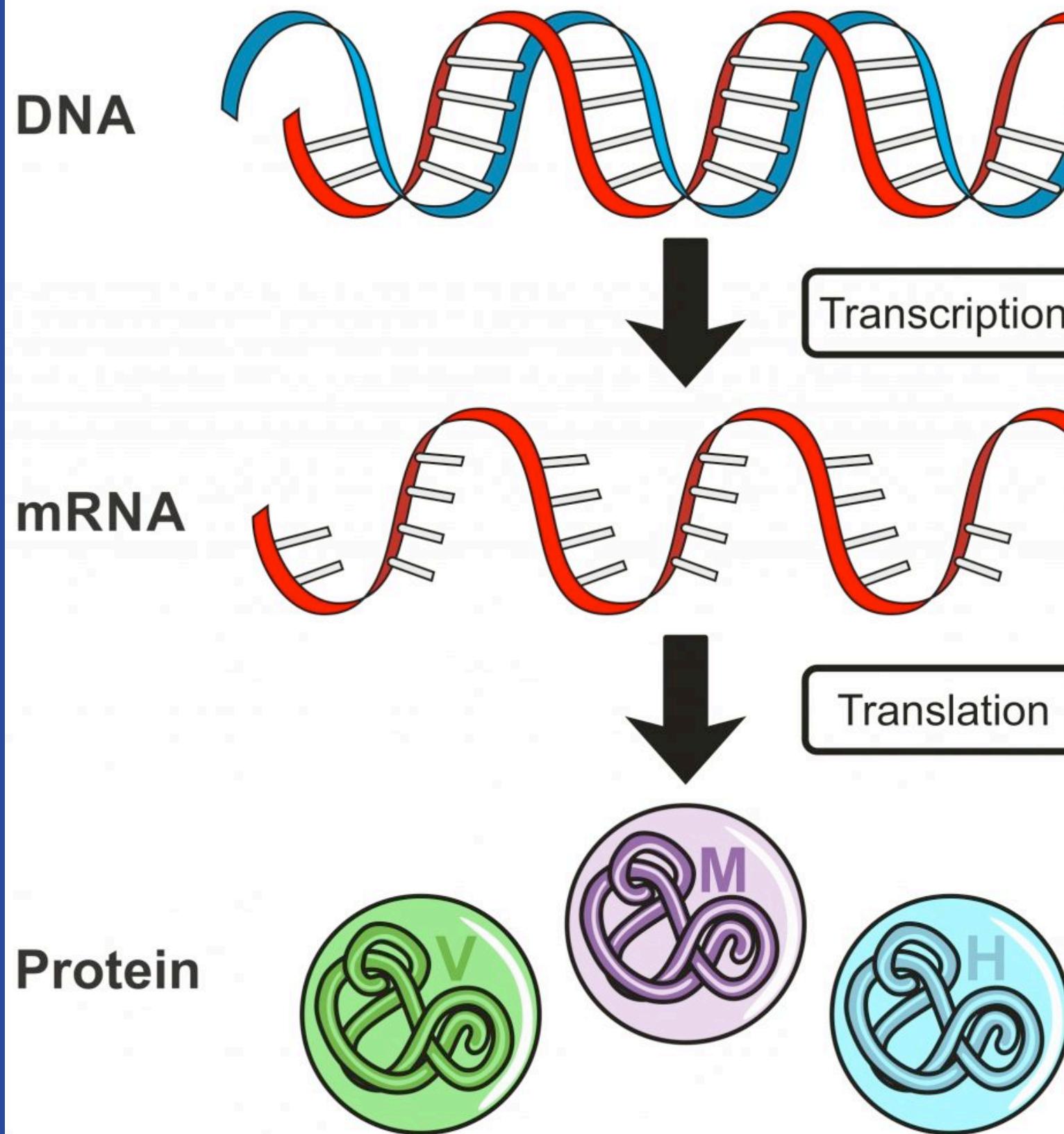
### Focus

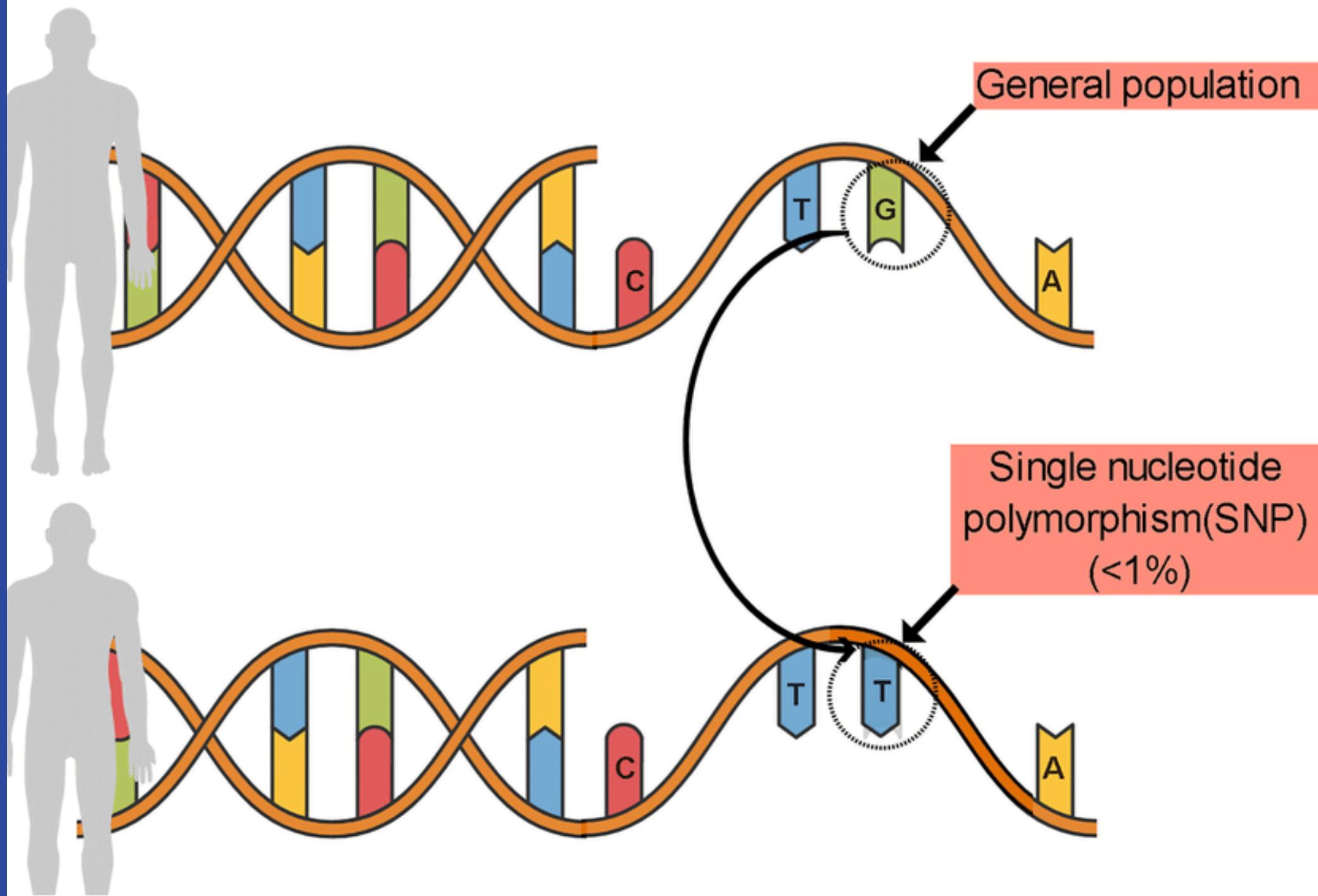
The impact of **single nucleotide polymorphisms** (SNP) related to PD in protein-protein interaction networks.

# 3,000,000,000

**Human DNA has about 3 billion base pairs.**







# SNP

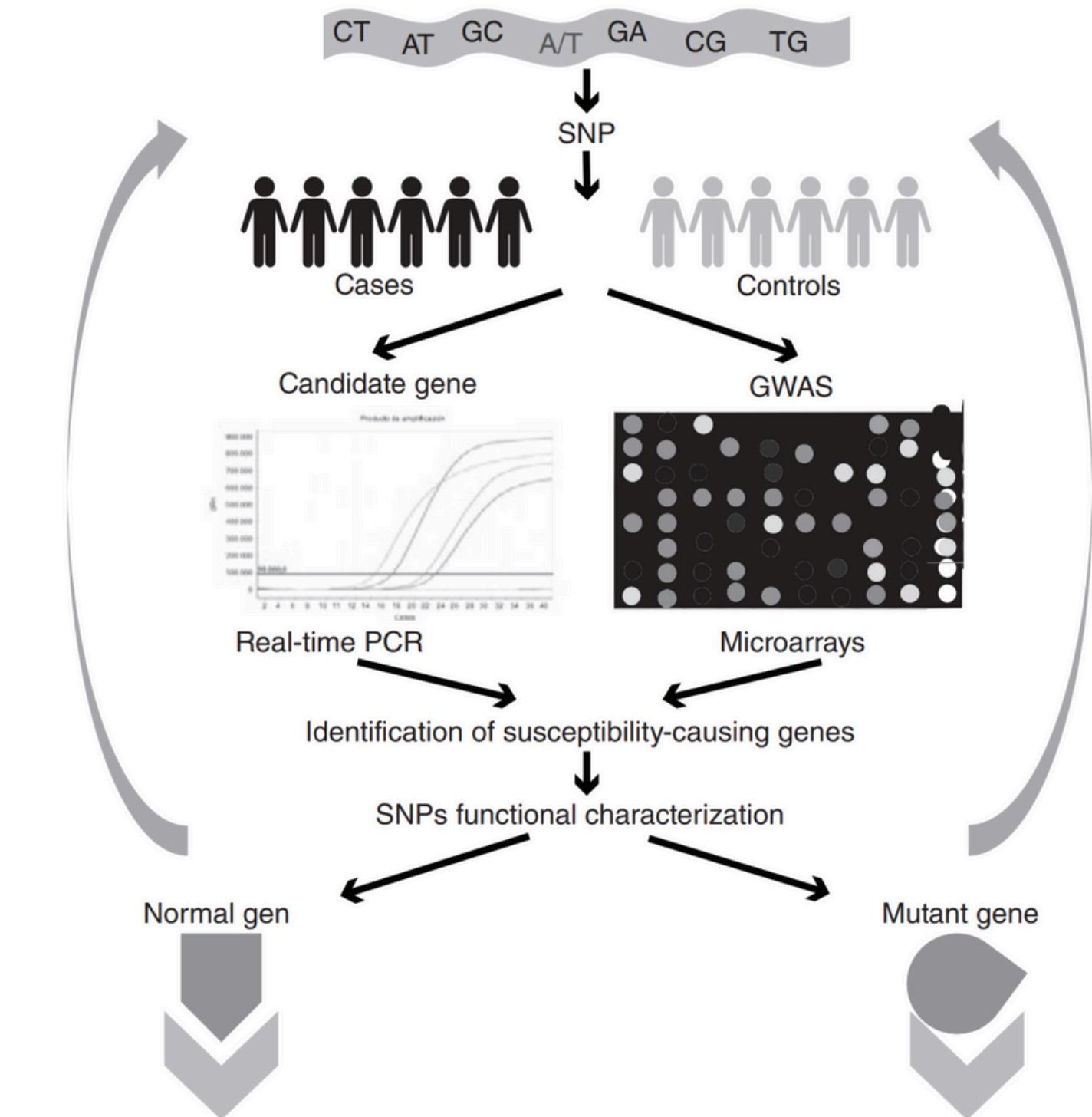
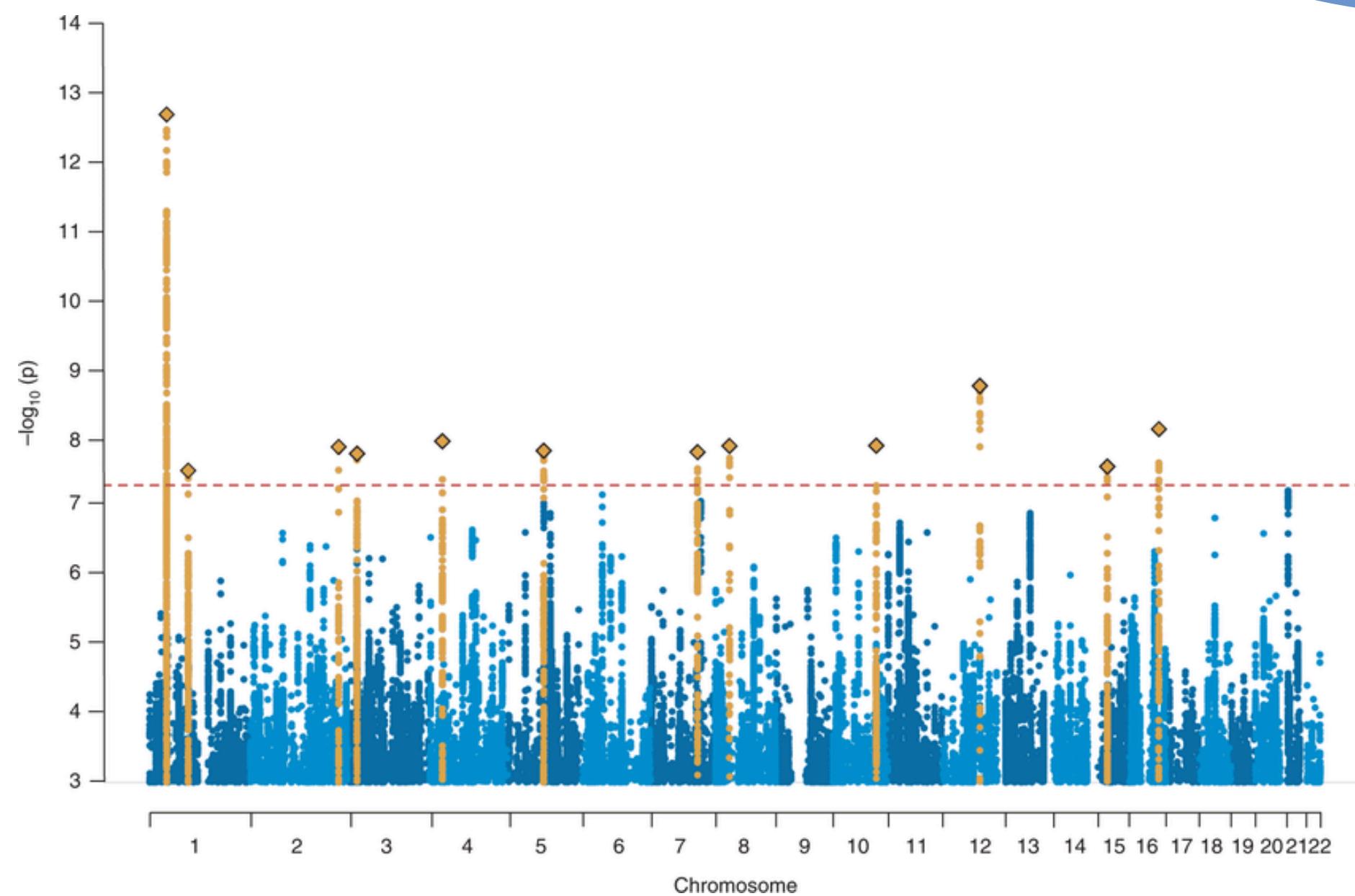
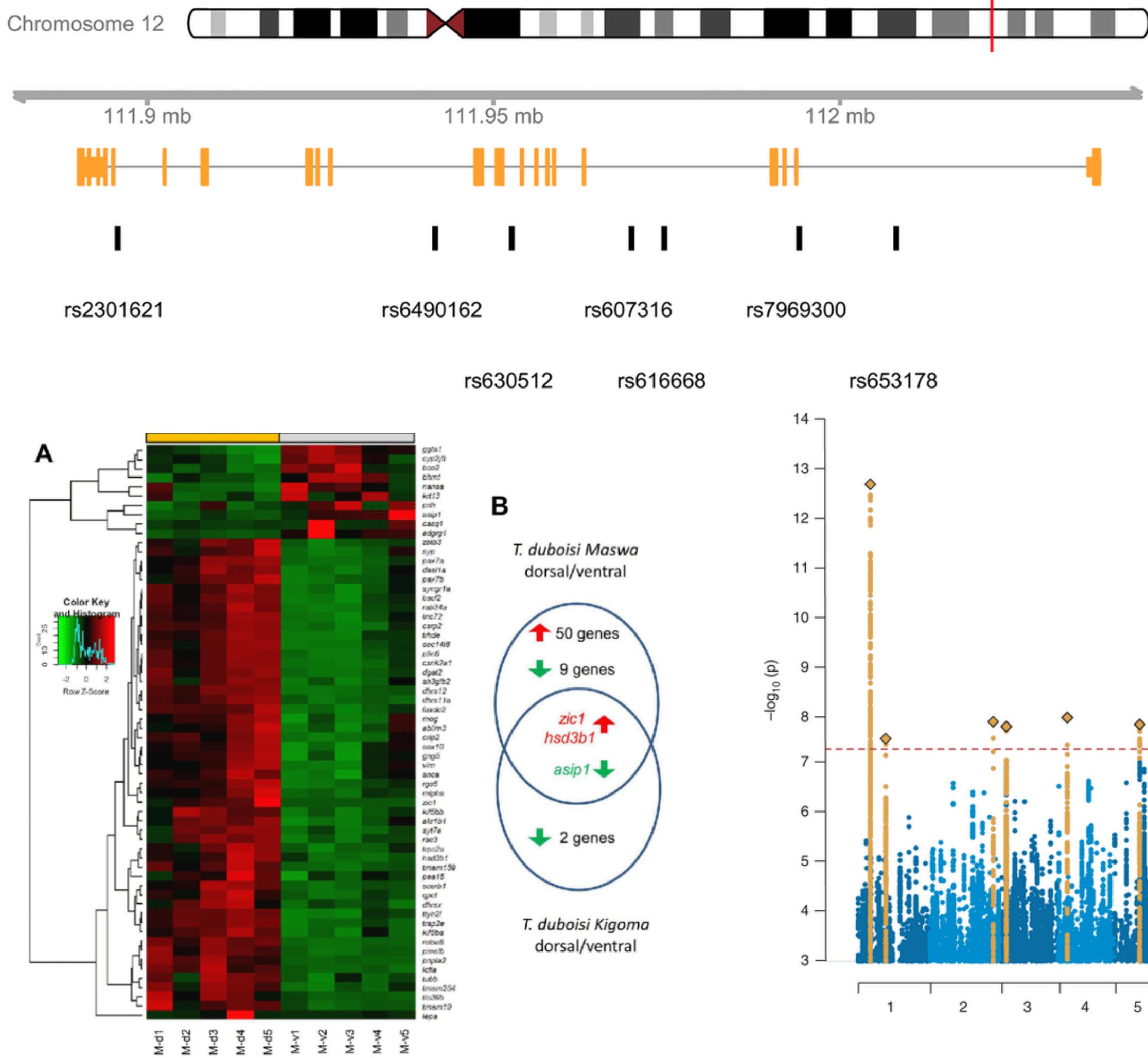
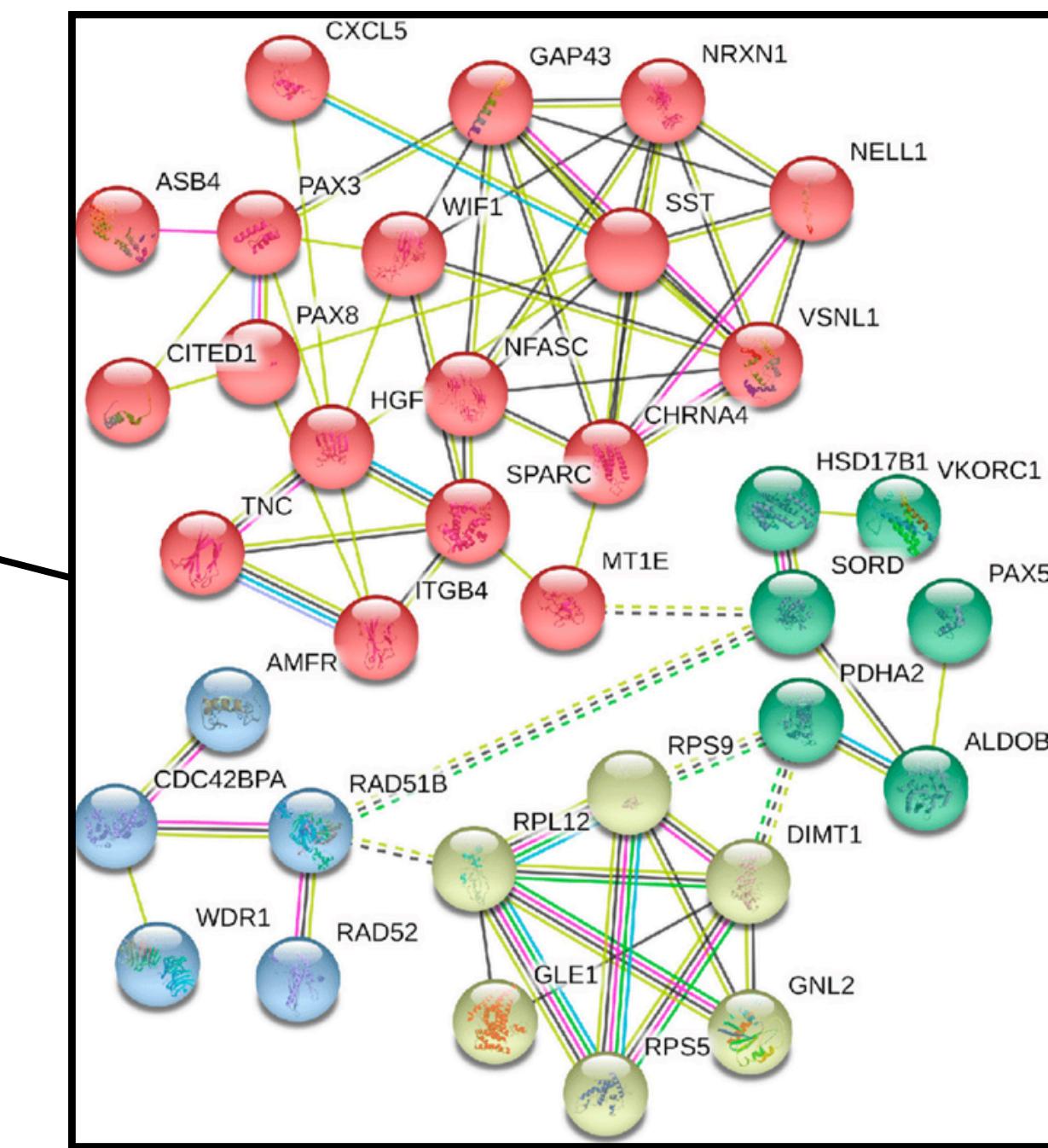
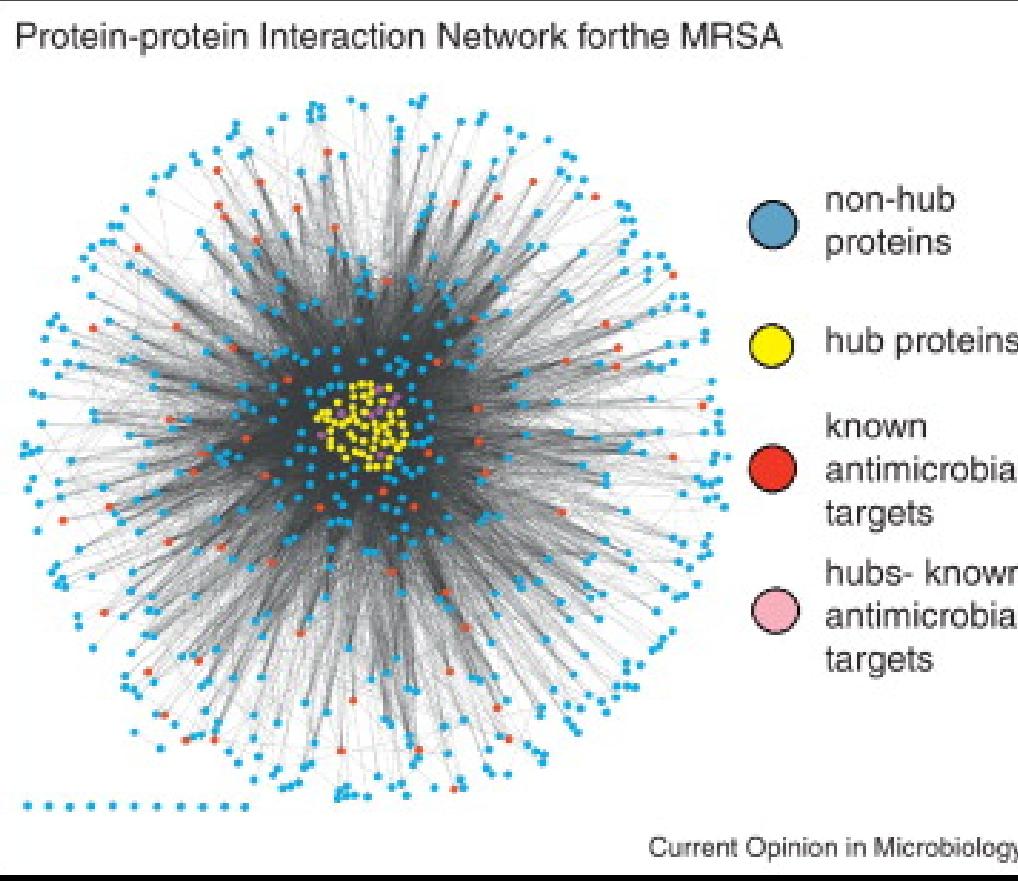
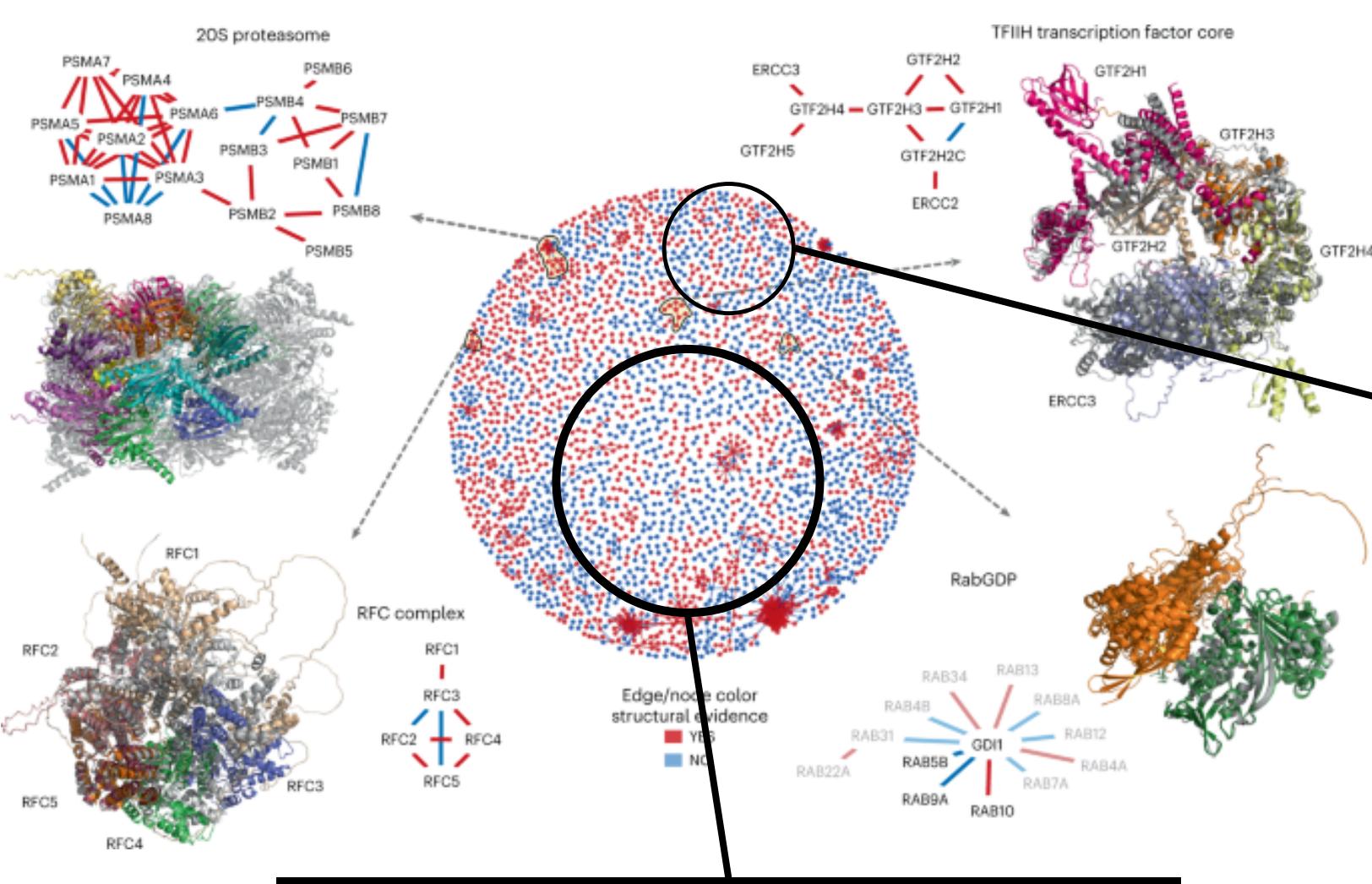


Figure 1  
gen...  
diffe...

# SNP

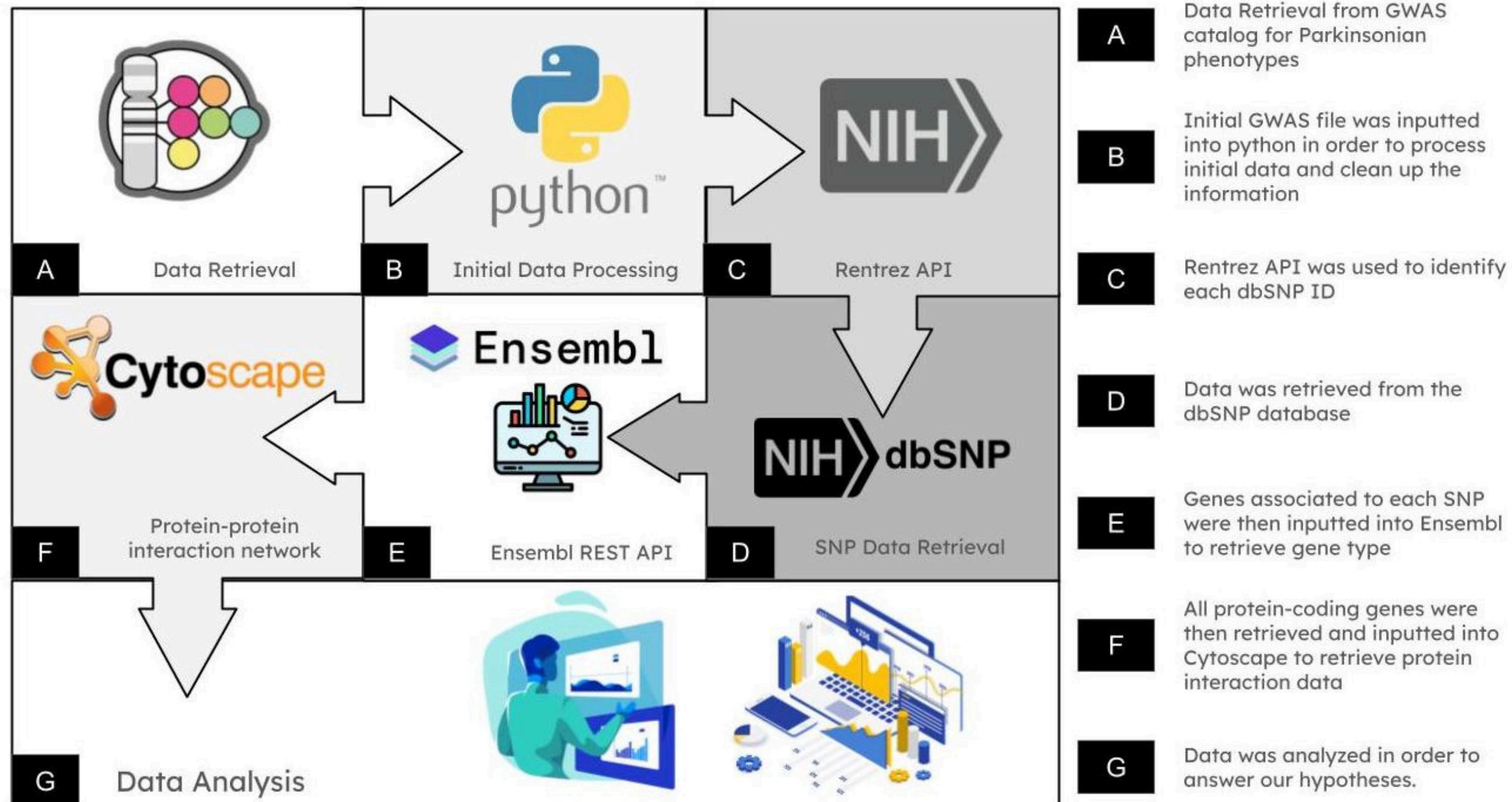




# Protein-protein interaction network

# Methods

## GWAS-to-PPI Data Workflow



## QUESTION 1: PD-GENES AND SNPs

**WHICH SNPs AND GENES ARE MOST ASSOCIATED WITH PD? WHICH POPULATIONS WERE THESE SAMPLES TAKEN FROM?**

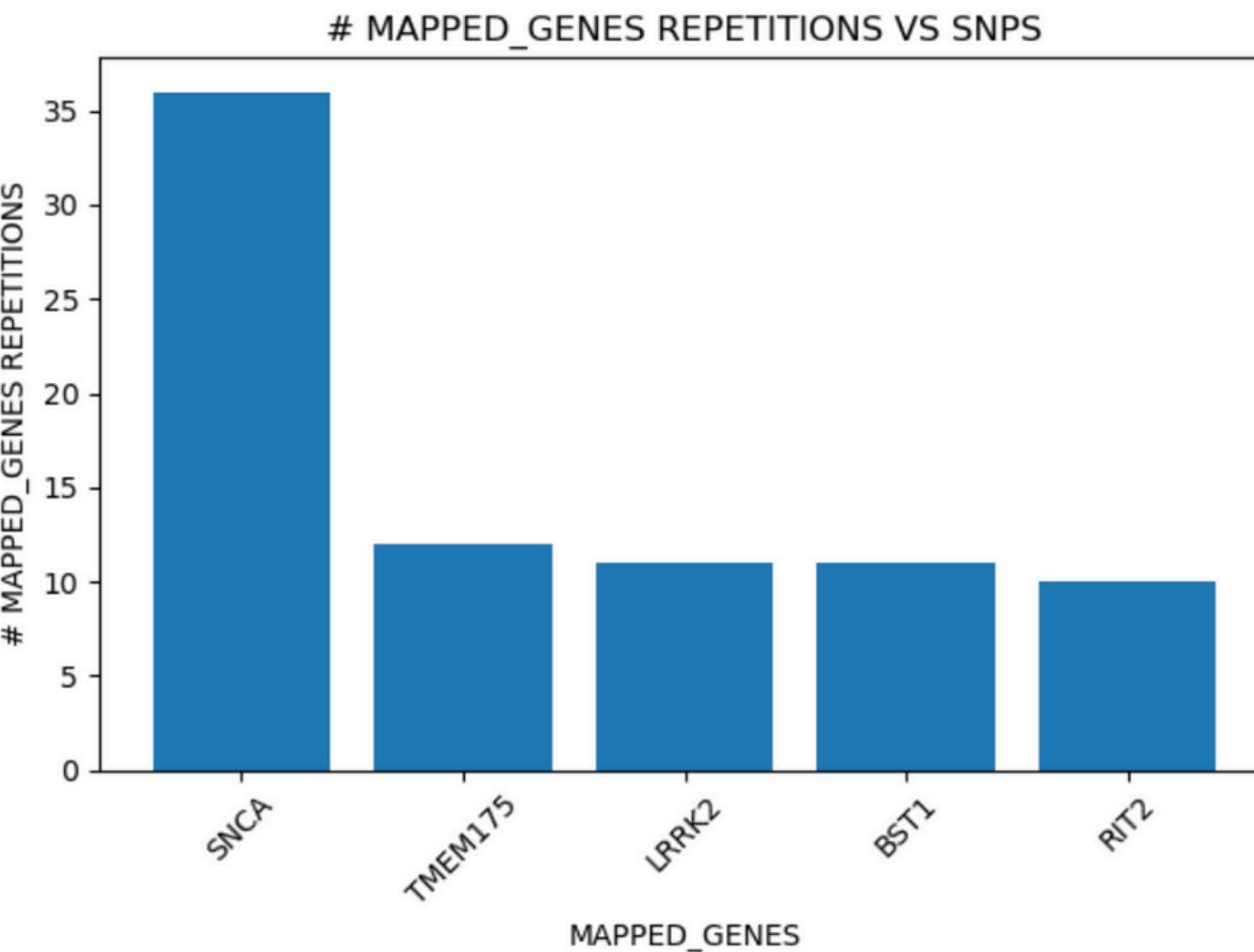
## QUESTION 2: PD-SNP GENE TYPE

**WHAT TYPE OF GENES WERE Affected BY THESE SNPs?**

## QUESTION 3: PROTEIN INTERACTIONS

**WHICH PROTEIN-CODING GENES FORM PART OF THE PPI?**

# Identify PD- related SNPs and Genes



**Unique mapped gene:296**  
**Mapped gene repetitions: 408**  
**Total of mapped genes:704**

MAPPED_GENE	# MAPPED_GENES REPETITIONS	SNPS
SNCA	36	rs356182
TMEM175	12	rs34311866
LRRK2	11	rs28903073
BST1	11	rs4266290
RIT2	10	rs4130047
MCCC1	9	rs11711441
STK39 - RN7SL813P	8	rs13016703
SH3GL2	8	rs2209440
DLG2	8	rs10501570
WNT3	7	rs199501
RORA - LINC02349	7	rs2414739
GCH1	7	rs11158026
NUCKS1 - RAB29	6	rs823118
LINC02210-CRHR1	6	rs62053943
HMGN2P18 - KRTCAP2	6	rs35749011
FAM47E, FAM47E-STBD1	6	rs6812193

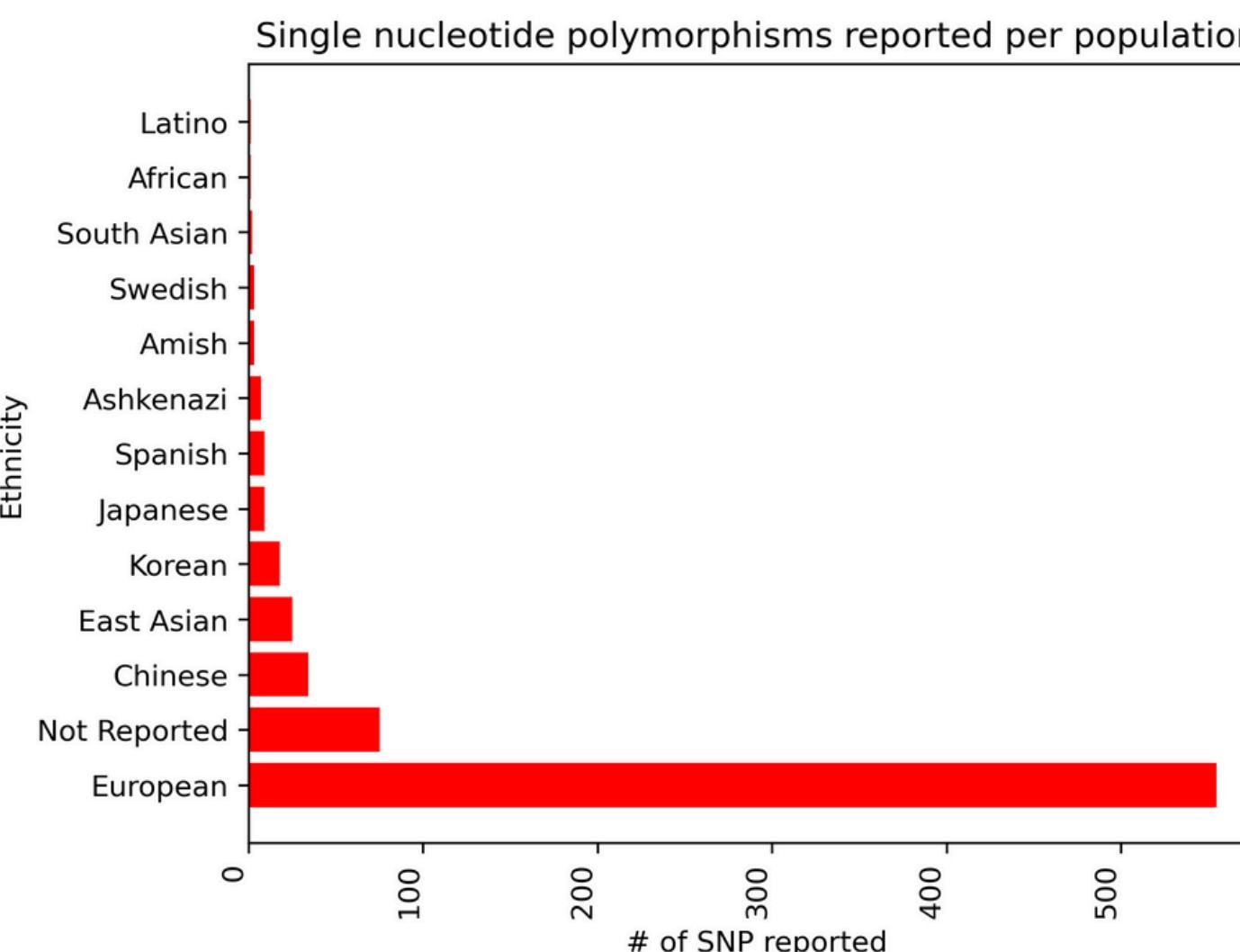
# Identify PD- related SNPs and Genes

## Sample of people presenting mutations of SNPs

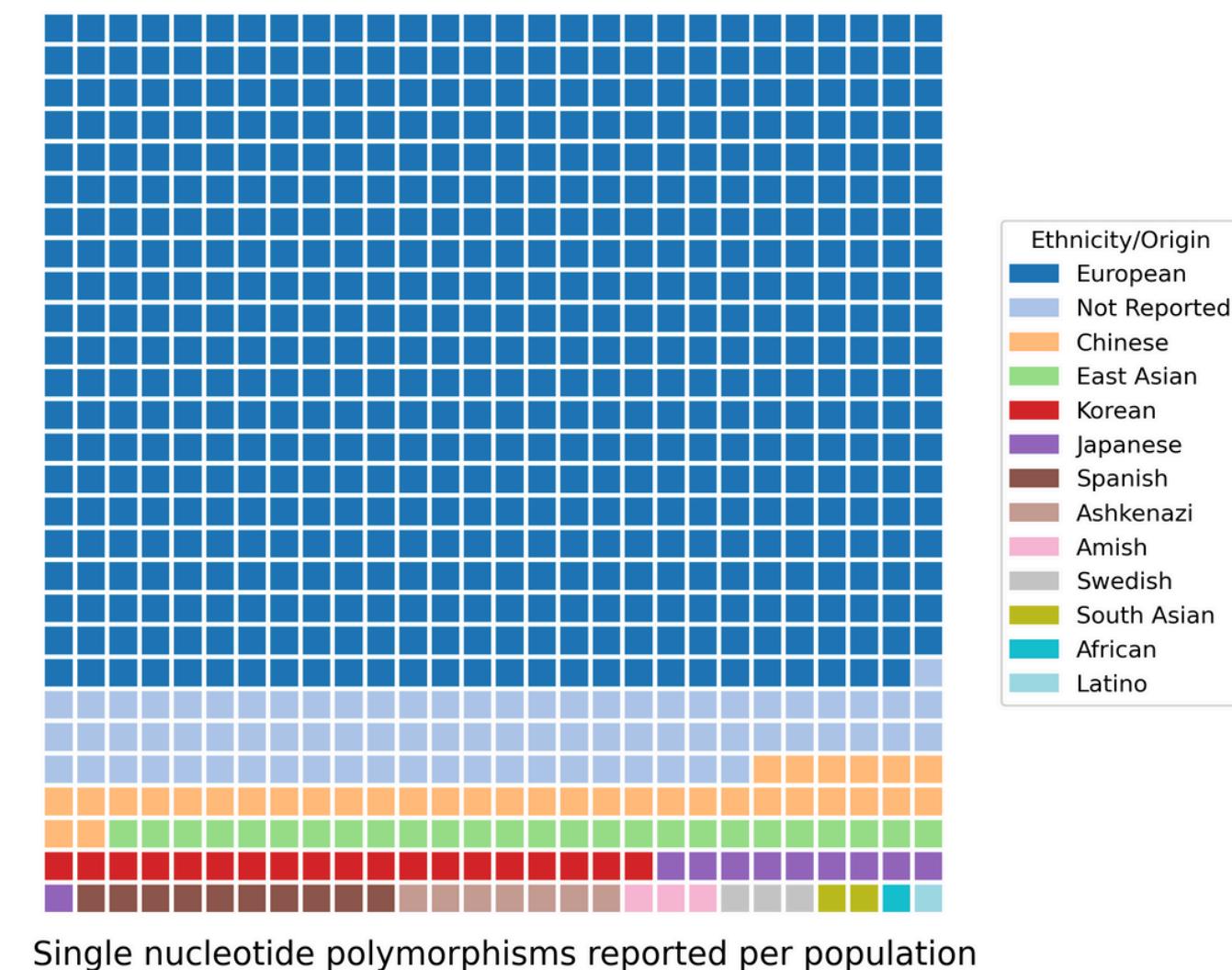
```
Ethnicity/Origin
European      555
cases,
individuals   42
East          29
Chinese        23
Korean         22
Han            18
Japanese       12
Spanish         9
Ashkenazi      9
cases           7
Swedish        4
Amish          3
South          3
least           2
African         2
Latino          1
Name: count, dtype: int64
```

	Ethnicity/Origin	count
0	European	555
1	Not Reported	75
2	Chinese	34
3	East Asian	25
4	Korean	18
5	Japanese	9
6	Spanish	9
7	Ashkenazi	7
8	Amish	3
9	Swedish	3
10	South Asian	2
11	African	1
12	Latino	1

# Identify PD- related SNPs and Genes

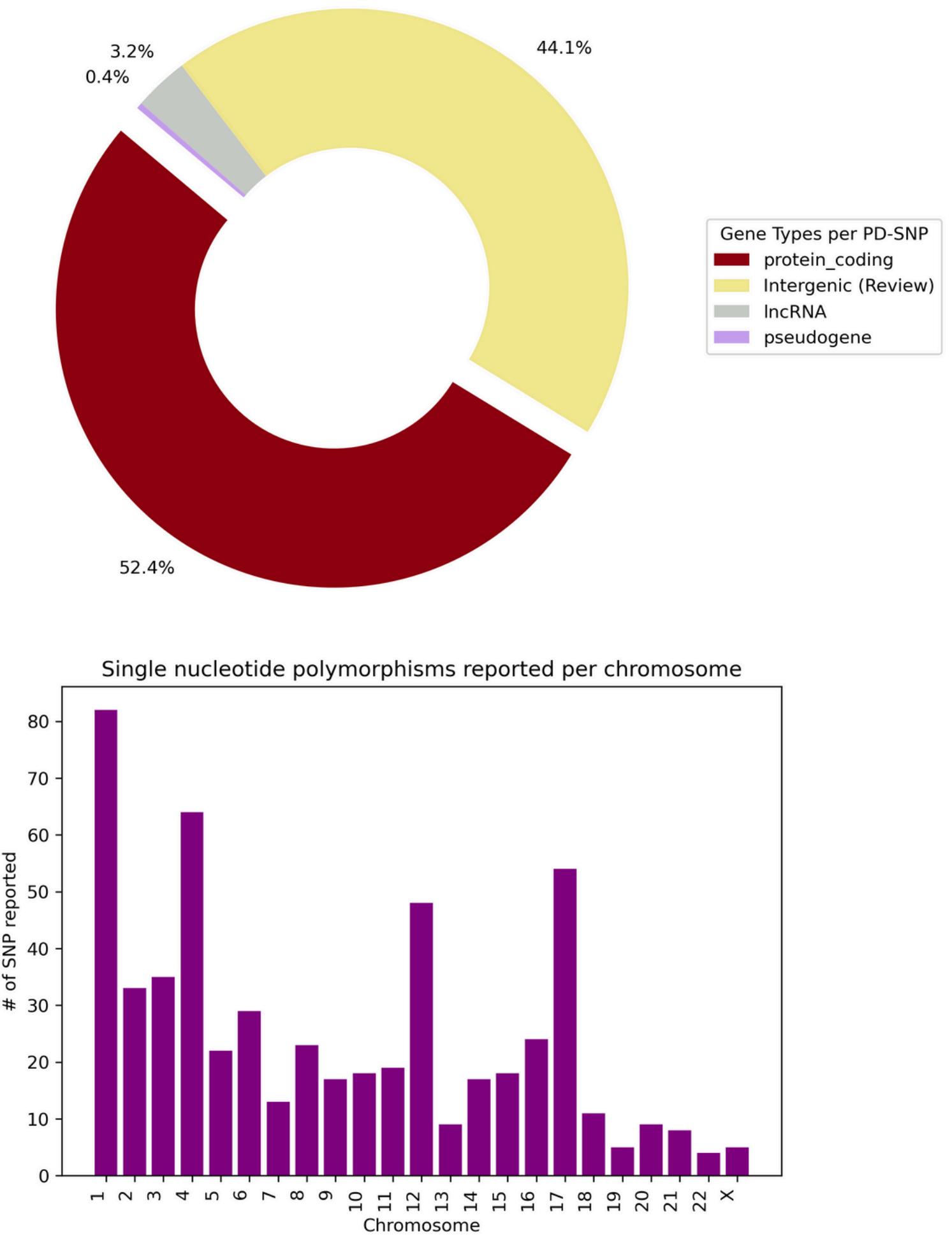


Waffle Chart

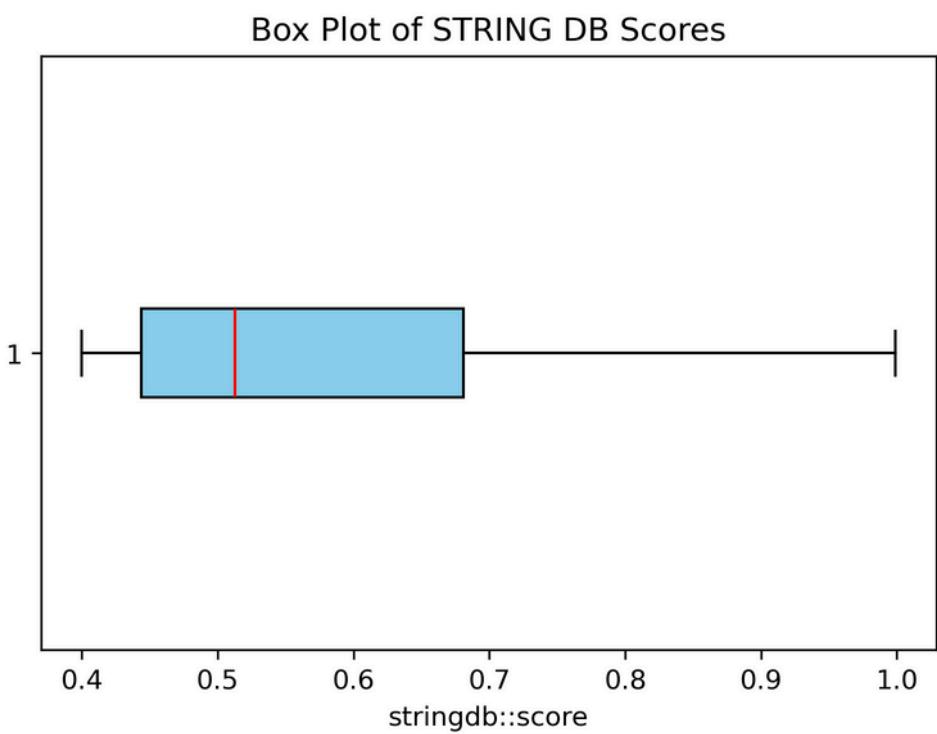
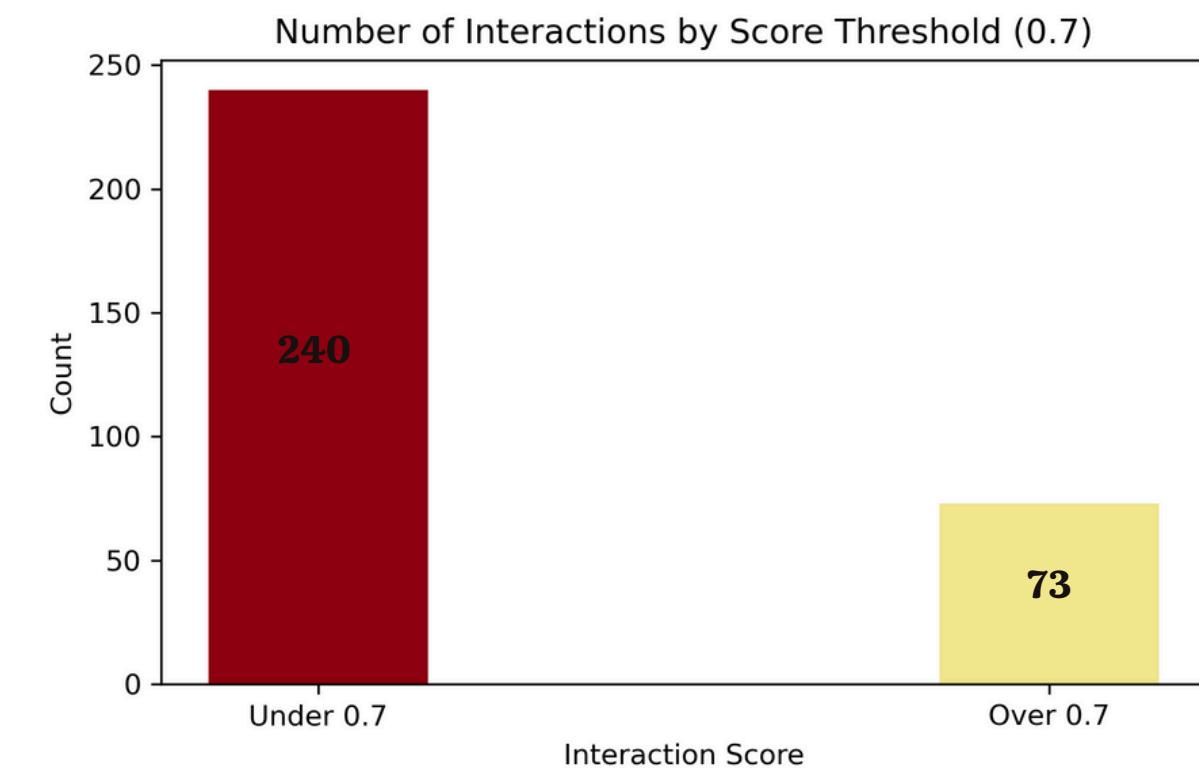
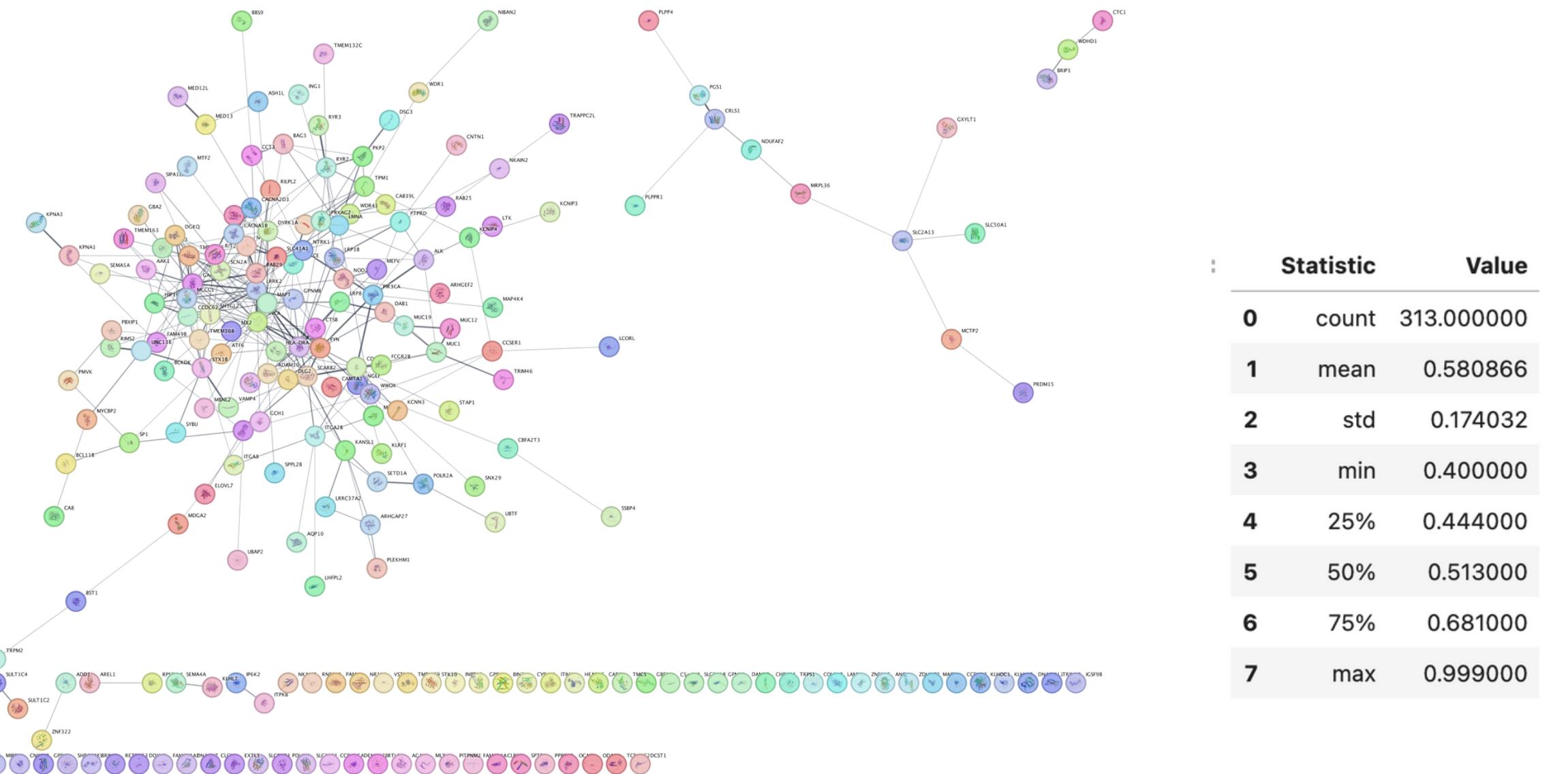


Bar Chart

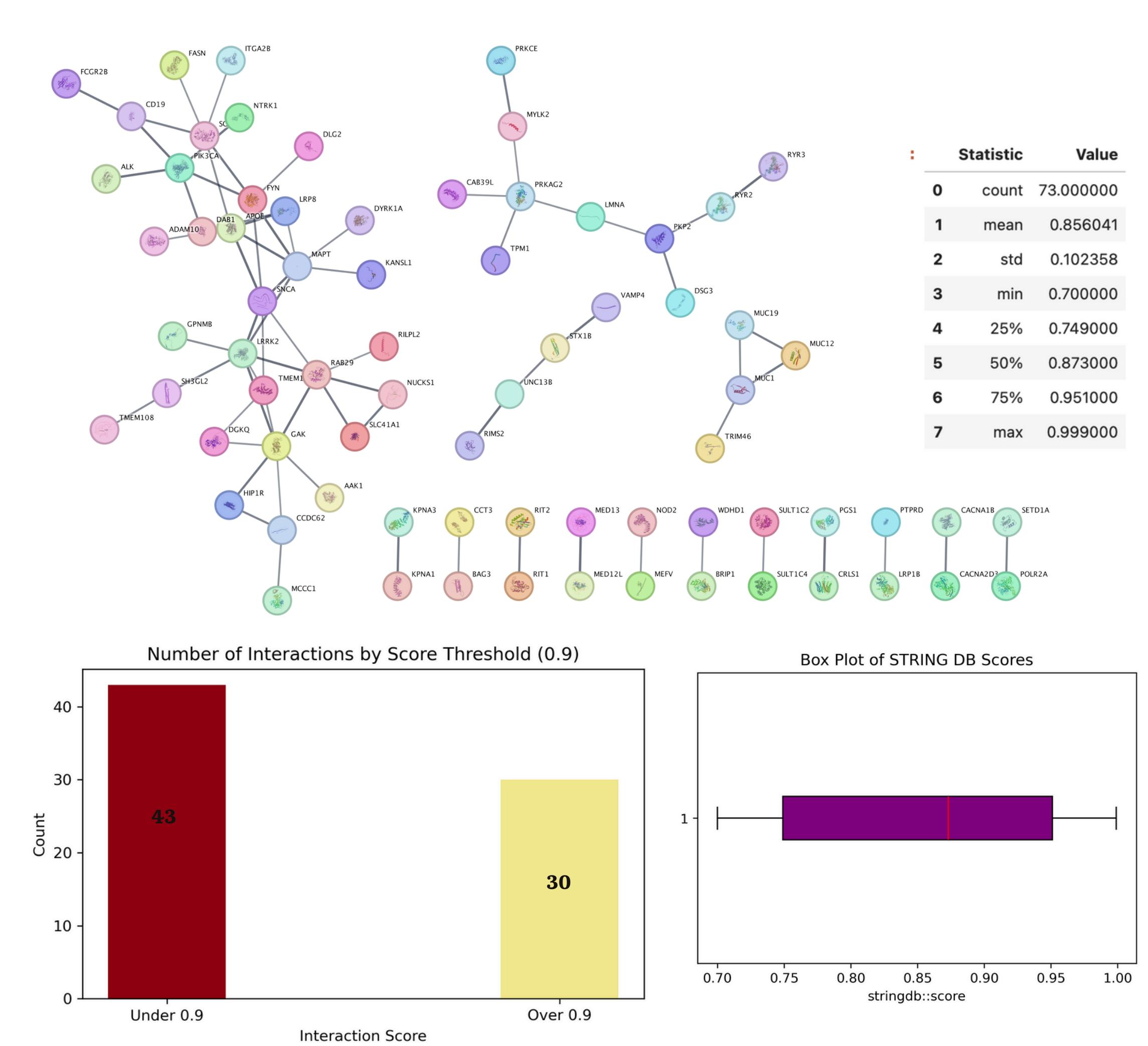
# Type of Genes Affected by PD-SNPs



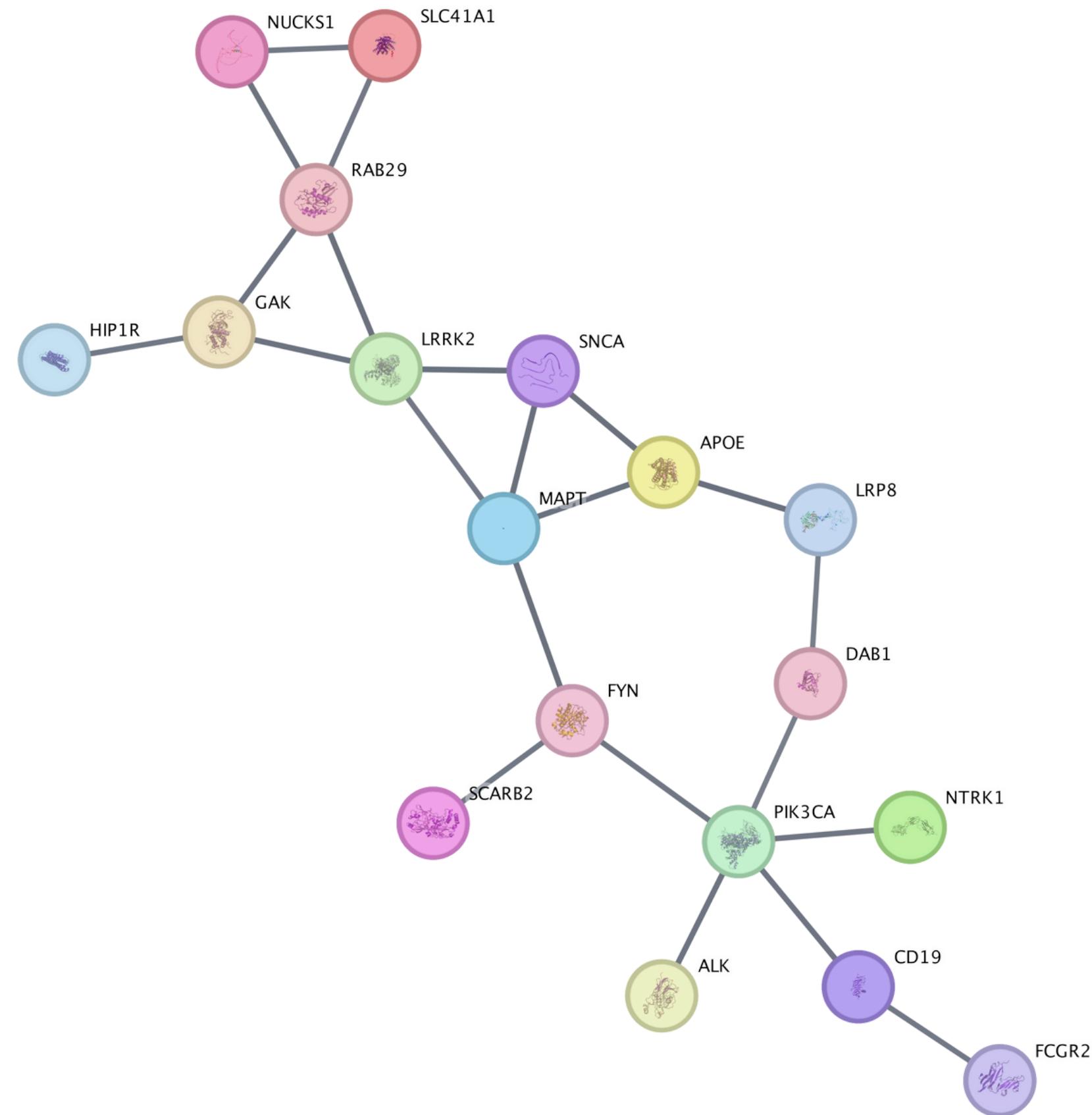
# Protein-protein interaction analysis



# Protein-protein interaction analysis



# Protein-protein interaction analysis



# Summary

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**Parkinson's Disease (PD)** is a progressive brain disorder that causes nerve cells to break down, leading to movement and mood problems.

**Single Nucleotide Polymorphisms (SNPs)** are small genetic variations in our DNA that can influence how genes function.

**A Protein-Protein Interaction (PPI) network** is a map showing how proteins work together to carry out vital cellular processes. Disruptions in these networks can lead to diseases, including PD, by impairing neuron communication or cellular maintenance.

# Conclusion

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As demonstrated in this report, most of the studies about PD are on the European ancestry group. Therefore, one conclusion is that more studies are needed in other ancestry groups.

Our data demonstrates that specific **SNPs**, such as **rs356182** in the **SNCA gene** are strongly linked to Parkinson's Disease. These SNPs disproportionately affect genes that are critical for neuron health and function, with key proteins forming hubs in PPI networks.

These findings provide valuable insights into genetic and molecular basis for Parkinson Disease, which help to create targeted therapies and further research.