*Simpósio de Susceptibilidade Genética Aplicada à Saúde*

# EXPLORATORY BIOINFORMATICS ANALYSIS OF VARIANTS IN CELL DEATH RECEPTOR GENES

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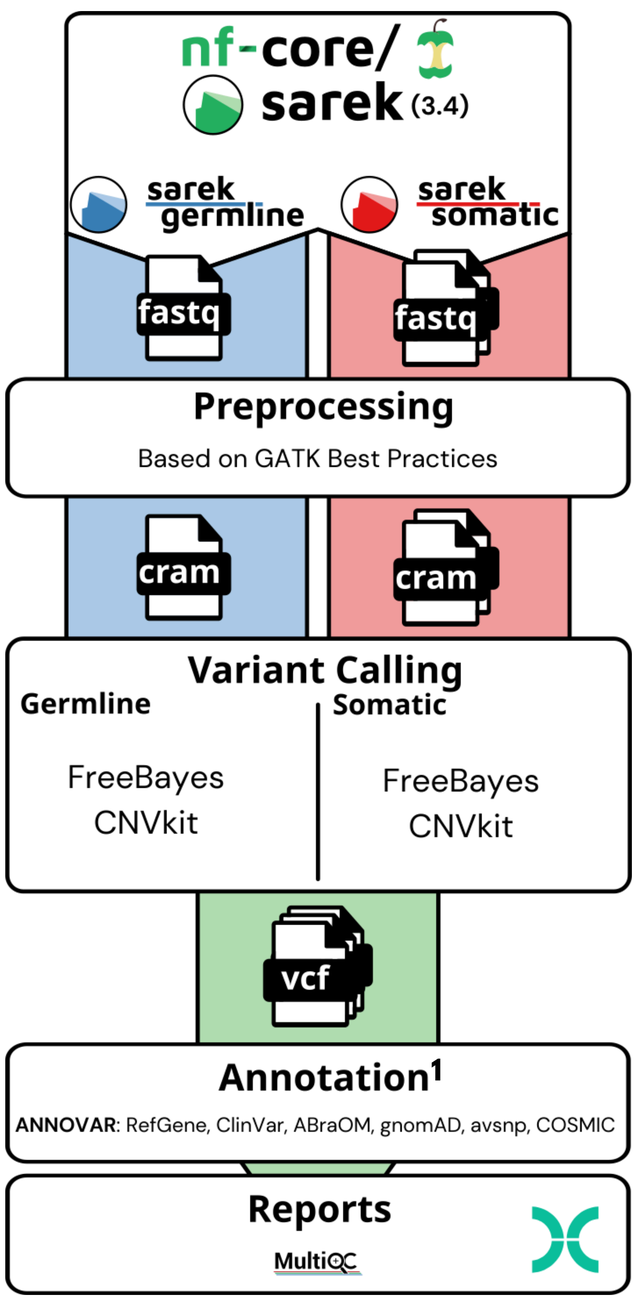
## INTRODUCTION

*Cellular receptors and sensitivities are mechanisms responsible for triggering cell death and maintaining tissue homeostasis. In cancer cells, these receptors may undergo mutations and no longer respond to cell death signals when activated. Identifying and mapping these mutations is crucial for understanding cancer biology, aiding in diagnosis, and guiding therapeutic interventions. Our* ***objective*** *is to explore and classify the distribution of mutations in cell death receptors and triggers in breast cancer patients through exome analysis.*

## METHODOLOGY

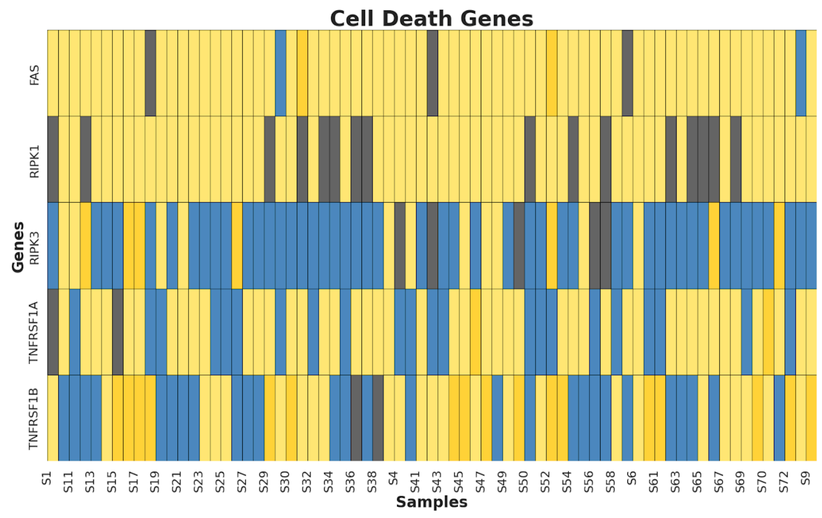



***Selection of variant related to cell death and plotting:***

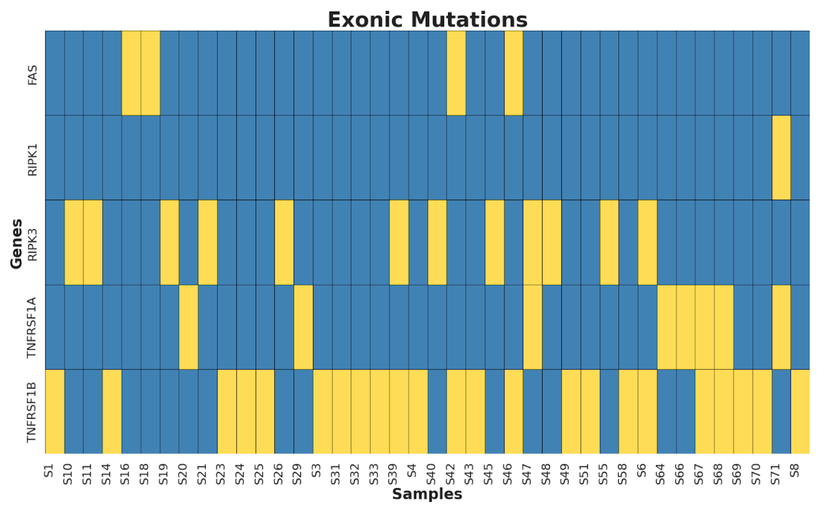
*FAS receptor*
*TNFRSF1A and TNFRSF1B*
*RIPK1 and RIPK3*

1 *GRCh38; RefGene (20211019); ClinVar (2024 0924); gnomAD 41 exome; ABraOM (2018 1204); avsnp150; COSMIC 100.*

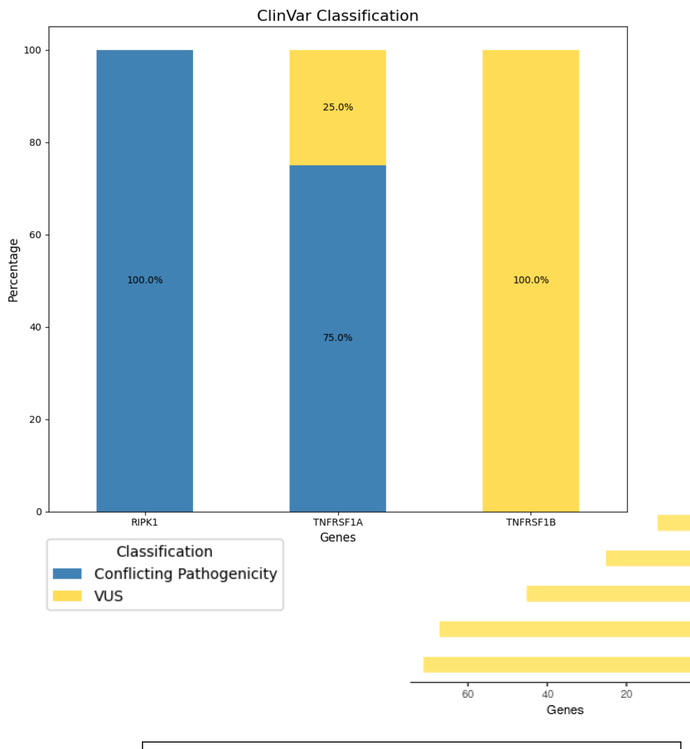
## RESULTS

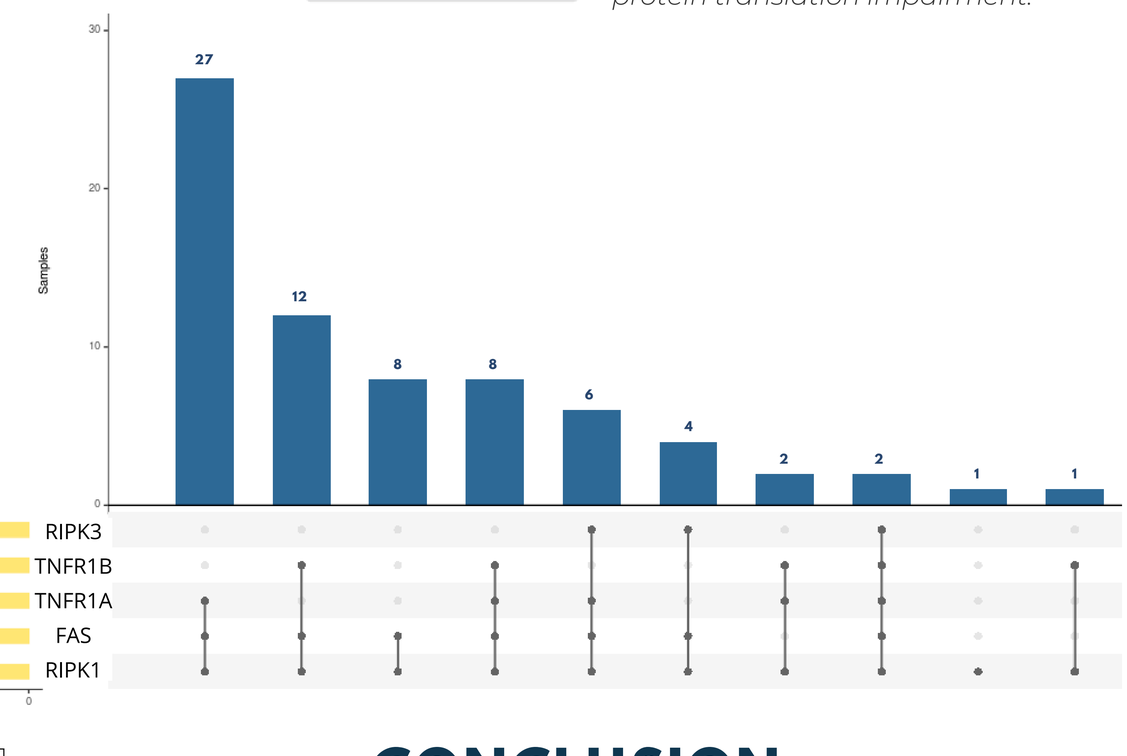


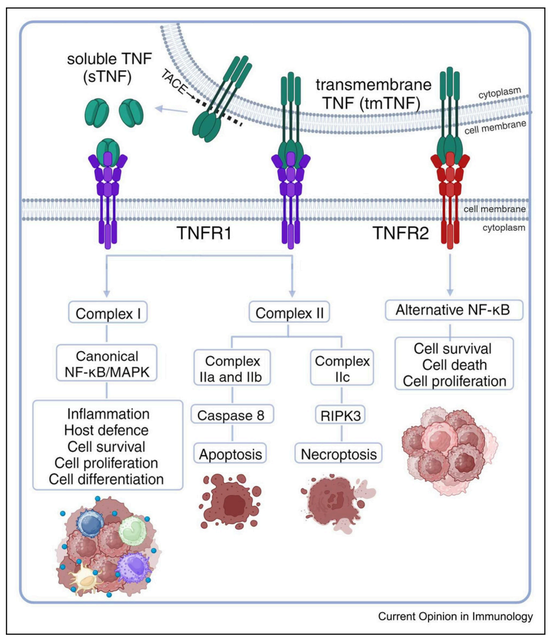

***Mutations:*** *upstream, 5'UTR, and fewer 3'UTR, along with non-frameshift substitutions, were identified across all gene samples.*




*The most frequent mutation types were non-synonymous and stop codon, which are critical for protein translation impairment.*




## CONCLUSION

*TNFRSF1B (TNFR2) emerged as the most frequently mutated receptor in this cohort. Specifically, 57.5% of the mutations were characterized as Stop Gain variants, while the remaining were Non-synonymous SNVs. These mutations were identified in 35.21% of samples, frequently co-occurring with mutations in RIPK1.*

*All TNFR2 variants detected are classified as VUS according to ClinVar. This finding underscores the potential for additional investigation to clarify the biological and clinical impact in breast cancer pathology.*

## ACKNOWLEDGMENT