INTRODUCTION

- Liquid-liquid phase separation (LLPS) is the natural process where a liquid separates into two distinct phases which forms membrane-less organelles in cells.
- Condensates can form from high expression, intrinsically disordered regions, multiple folded domains, and other physical properties of proteins.

METHODS

ClinVar presents data on human variation and phenotype and the Human Gene Mutation Database (HGMD) presents mutations associated with human disease.

LLPS Protein Databases (DB)

- Using the LLPS UniProt IDs, generate a list of gene names
- Pathogenic clinical significance
- Single nucleotide variants
- LLPS Vs. Non-LLPS
- Edgotype Distribution
- Significantly Enriched Diseases
- GOF/LOF

CONCLUSION

Overall, we observe significant data associating cancer and neurodegenerative diseases with mutations in LLPS related diseases. Joining the structural analyses with our current data and performing the same analyses on cancer mutation databases like The Cancer Genome Atlas will allow us to better understand the roles of mutations in LLPS related diseases.

Acknowledgements

I would like to thank Dr. Yi and Dr. Sahni for welcoming me into their project and giving me the chance to work with very supportive people. I thank Sueda Cetinkaya for her patience and guidance and always being ready to help. Finally, I would like to thank the department of Women and Minority Faculty Inclusion for allowing me to participate in the Partnerships for Careers in Cancer Science and Medicine program again.

FUTURE DIRECTIONS

We acquired promising data in our systematic analyses for further investigating the impact of mutations on protein structure and properties to understand the roles of LLPS in diseases. Joining the structural analyses with our current data and performing the same analyses on cancer mutation databases like The Cancer Genome Atlas will allow us to better understand the roles of mutations in LLPS related diseases.

REFERENCES

1) Sahni, Yi et al. Widespread Macromolecular Interaction Perturbations in Human Genetic Disorders 2015;161:647-660
2) Bouchard et al. Cancer Mutations of the tumor suppressor SPOP disrupt the formation of active, phase-separated compartments 2019; 72:19-36
4) Tsang et al. Phase Separation as Missing Mechanism for Interpretation of Disease Mutations 2020;183:1742-1756