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CMPSC/BIO
Lab 6 Proposal

# Genetic Screening

For our final project, our work will be centered around the idea of genetic screening (particularly of infants at birth). We are going to make an attempt to query an on-line database (disgenet.org) from our Python program that produces results based on user input. Following the query, the database will search for genes commonly associated with the input disease. We will take either one or a few of the top related genes associated with the disease (rating is determined by the reliability of the gene – disease base pair, meaning the % of cases where the gene is related to the disease) and use the link(s) provided by disgenet that offers a link to the NCBI database where we are able to obtain the FASTA file of the non-mutated gene. Once that information is gathered, we will compare it to a sample sequence of the gene that we generate and look for mutations. If any mutations are present, we will then take the necessary following steps to determine the type of the mutation which could factor into the development of the disease and output the information to the user, along with an explanation of our analysis.

According to [1] The U.S. Department of Health and Services recommends a screening of infants for 31 core disorders and 26 secondary disorders separated into the following categories:

- Metabolic disorders
- Endocrine disorders
- Hemoglobin disorders
- Other disorders (ex: cystic fibrosis)

We are looking to expand this idea so that someone who is able to obtain the necessary genetic information can search for a things that may not be included in the recommended list which may run in a family's genetic history.

This tool could also be used in the protection of infants as well. As suggested by the New England Journal of Medicine [3], which looked into neuroblastoma in infants and how research done into early screening could reduce the number of child deaths from the tumor.

# Bibliography

[1] https://labtestsonline.org/understanding/wellness/a-newborn-1/a-newborn-2/

[2] Beck, C. L., & Rubarth, L. B. (2014). Genetic Testing in Infants. *Neonatal Network*, *33*(4), 217-220.

[3]Woods, W. G., Gao, R. N., Shuster, J. J., Robison, L. L., Bernstein, M., Weitzman, S., ... & Tuchman, M. (2002). Screening of infants and mortality due to neuroblastoma. *New England Journal of Medicine*, *346*(14), 1041-1046. Obtained at: http://www.nejm.org/doi/pdf/10.1056/NEJMoa012387