

Neighbor FoxP1

Mutation variants attributor

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Problems we are tackling

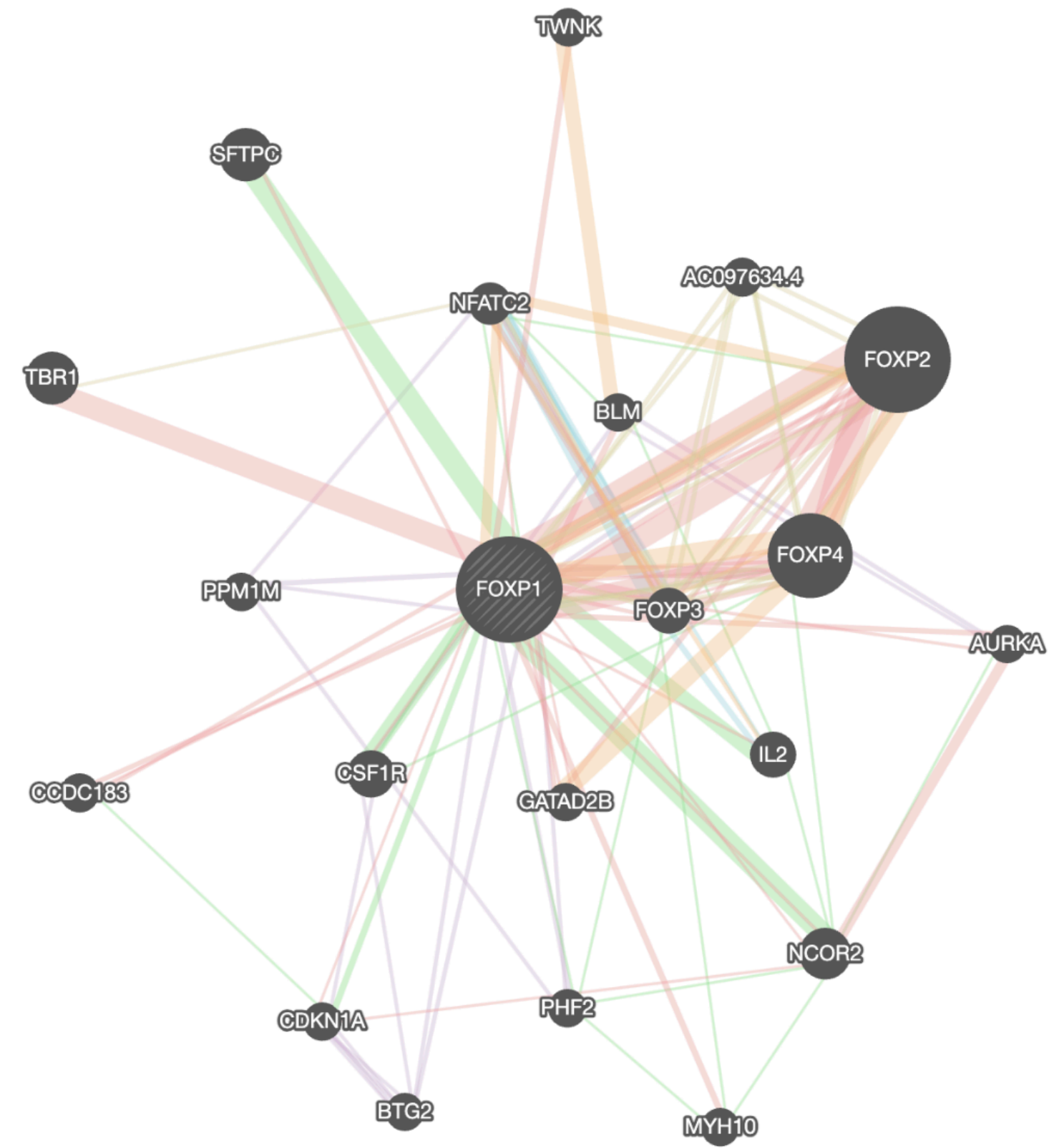
- The importance of understanding genetic variants in FoxP1 gene and their implications for rare diseases.
- A visual representation of all FoxP1 mutation variants which will help as a useful tool for doctors.
- We faced many challenges in interpreting FoxP1 variants due to limited available data and the need for computational tools to aid in variant analysis.
- We also faced the challenge of uncertainty in determining which gene exactly causes FoxP1.
- We decided to develop a tool that models different variants of FoxP1 and gives a correlation with many attributes that relates to genes it interacts with.

Our Approach

- The tool models the given variant provided as input in HGVS format by reading the Fasta values of the existing FoxP1 and mutating upon it.
- The mutation sequence model is further used to compare with the genes that have affiliation towards FoxP1.
- Various quantifiable attributes are derived between these models.
- The derived attributes are presented to visualize alongside the limited dataset associated with FoxP1 variant and possible patterns that could be observed.
- This approach allows us to map the limited dataset to abundant data of genes which are different and provide high correlation to understand its role in treatment of rare disease.

Data we have worked with

- We used the limited data sets which was available from the paper
- Develop Med Child Neuro-2021-Braden- Severe speech impairment is a distinguishing feature of Foxp1 related disorder.
- Link- <https://onlinelibrary.wiley.com/doi/epdf/10.1111/dmcn.14955>
- We also worked with <https://genemania.org/search/homo-sapiens/foxp1/>



Universe of proteins that interact with FoxP1

Results

- The results obtained from the analysis of FoxP1 variants using our approach.



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c.532C>T p.(Gln178*)

Neighbors	DNA Affinity	Alignment Score	Percentage Identity	GC Content (Comparing - Mutation)	Transition Traversal Ratio	Genetic Distance	Nucleotide Diversity	Probable mutation
IL2	21.00	3780.00	71.92	9.52	0.47	0.00	0.75	Insertion
GATAD2B	21.00	6808.00	9.69	0.60	0.51	0.16	0.07	Deletion
FOXP1	21.00	0.00	0.00	0.37	0.49	0.02	0.01	Deletion
FOXP4	21.00	6808.00	12.16	-13.24	0.51	0.20	0.09	Deletion
CSF1R	21.00	6808.00	11.33	-6.62	0.51	0.19	0.08	Deletion
BLM	21.00	6808.00	6.89	0.75	0.51	0.11	0.05	Deletion

Related Conditions

Severity: Mild

Action Items

- Assess need for: Community or online resources such as Parent to Parent; Social work involvement for parental support; Home nursing referral; Early intervention referral; Case management support referral.
- To inform affected persons & their families re nature, MOI, & implications of FOXP1 syndrome to facilitate medical & personal decision making
- To screen for behavior concerns incl ADHD, impulsivity, anxiety, sleep disturbances, &/or findings suggestive of ASD 1
- Evaluate speech production & receptive/expressive language in all persons, regardless of age.

Future things

- Researchers can use it and generate more data and understand the relation.
- Doctors can use it to take next steps and the severity of mutation.

Further implementation

- Accommodate more attributes as a function of relations between the gene.
- Build the protein structure with the mutations and correlate it better.
- Build more prediction algorithms.