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A bioinformatics analysis of A168D Substitution in the DMD Gene in Duchenne muscular dystrophy

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Background

- **Background**
- Duchenne muscular dystrophy is a progressive neuromuscular disease that leads to difficulties in movement and premature death. This disease is an X-linked recessive disorder. DMD disease is caused by mutations in DMD gene (encoding dystrophin) that destroy the production of dystrophin in muscle. Muscles without dystrophin are sensitive to damage, that cause progressive loss of muscle tissue and function . In this study, a single nucleotide polymorphism (SNP) in NCBI was selected in the DMD gene for investigation.

Materials and Methods

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- In (rs128626236 G>T), the effect of conversion of alanine, a hydrophobic amino acid to aspartic acid, a hydrophilic amino acid on protein structure was assessed using SIFT, PROVEAN and I-Mutant databases.

Results

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- In this study, the SIFT database showed that Substitution at pos 168 from A to D is predicted to AFFECT PROTEIN FUNCTION with a score of 0.01.The variant was then examined in the PROVEAN database. According to the PROVEAN database, the A168D variant with provean score = -4.513 (cutoff= -2.5) is predicted to be a DELETERIOUS variant. Finally, the variant was assessed using the I-MUTANT database [this database is Predictor of Protein Stability Changes upon Mutations] and DDG = -0.52 was obtained (DDG less than 0: Decrease Stability).

Conclusion

- **Conclusion**
- These results suggest that the A168D variant in the rs128626236 region of the DMD gene probably is a Deleterious variant and affects protein function. However, more investigation is needed to clarify this.

References

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