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# Analysis of IL11RA IN Association with Craniosynostosis

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craniosynostosis. This study's purpose is to identify the effects of the IL11RA gene and missense variants. Simple ClinVar analysis was used to identify variants associated with this disease. Computational analysis tools, such as SIFT and PolyPhen2, were used to predict the effects of the Arg296Trp mutation. It was concluded that the Arg296Trp mutation is damaging and will impact protein function. The objective of this research is to furnish the medical community with additional insights, thereby advancing the quest for further breakthroughs in treatments.

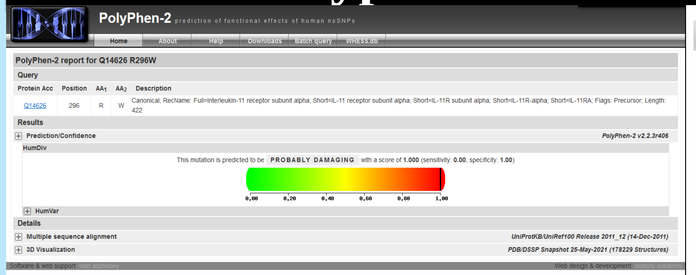
# INTRODUCTION

Craniosynostosis is a disorder in which fibrous joints go between the bones of a baby's skull and close prematurely. For example, babies skull are like puzzle pieces, the puzzle pieces are made up of several bones. When babies are born they have borders called cranial sutures that are not firmly connected yet. Which leads the skull to expand as the babies are growing. However, when one of the sutures closes too soon, this can cause many issues. If it's not treated, the increased intracranial pressure can lead to developmental problems, headaches, brain damage, or blindness. Researches have found many genes that are mutated linked to craniosynostosis.

# METHODS

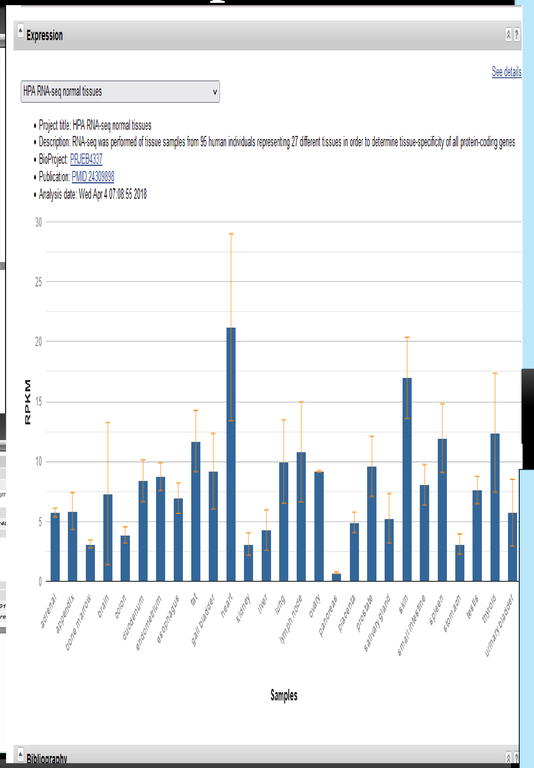
- Simple-Clinvar was used in this study to find gene mutations IL11RA in the presence of craniosynostosis
- Swiss modeling was used to obtain automated protein homology
- Modeling of protein methods was used in this study to find amino acid substitution that will affect protein function
- SIFT programming was used to select the variants that affect protein function.

## Polyphen 2

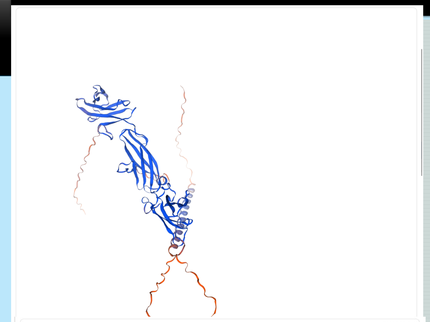


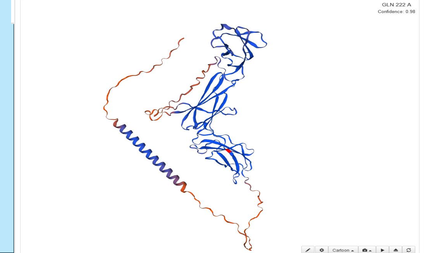


## Gene Expression

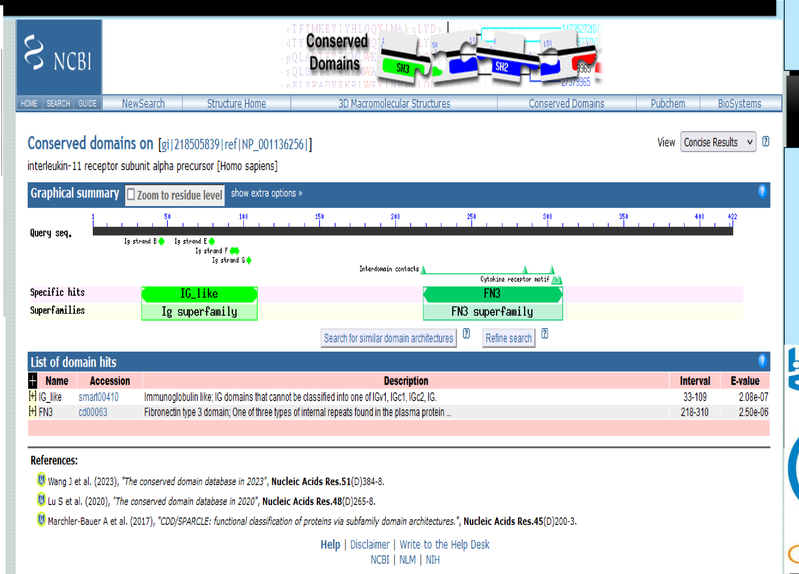



## SWISS Pro Modeling





## Conserved Domain

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CODE

Characterizing Our DNA Exceptions