Oakwood University

Student Posters

Student Creative Works

4-2024

Analysis of IL11RA IN Association with Craniosynostosis

Elaine Vanterpool Oakwood University, evanterpool@oakwood.edu

Sukari Daphnis Oakwood University

Follow this and additional works at: https://ouscholars.oakwood.edu/student-posters

Recommended Citation

Vanterpool, Elaine and Daphnis, Sukari, "Analysis of IL11RA IN Association with Craniosynostosis" (2024). *Student Posters*. 50.

https://ouscholars.oakwood.edu/student-posters/50

This Poster is brought to you for free and open access by the Student Creative Works at OUScholars. It has been accepted for inclusion in Student Posters by an authorized administrator of OUScholars.

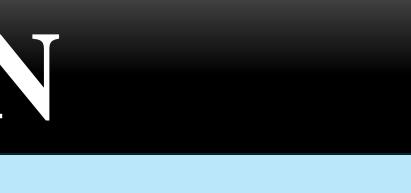
cramosynosiosis. This sludy s purpose is to identify the effects of the IL11RA gene and missense variants. Simple ClinVar analysis was used to identify variants associated with this disease. Computational analysis tools, such as SIFT and PolyPhen2, were used to predict the effects of the Arg296Trp mutation. 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.

NETHODS

- 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.

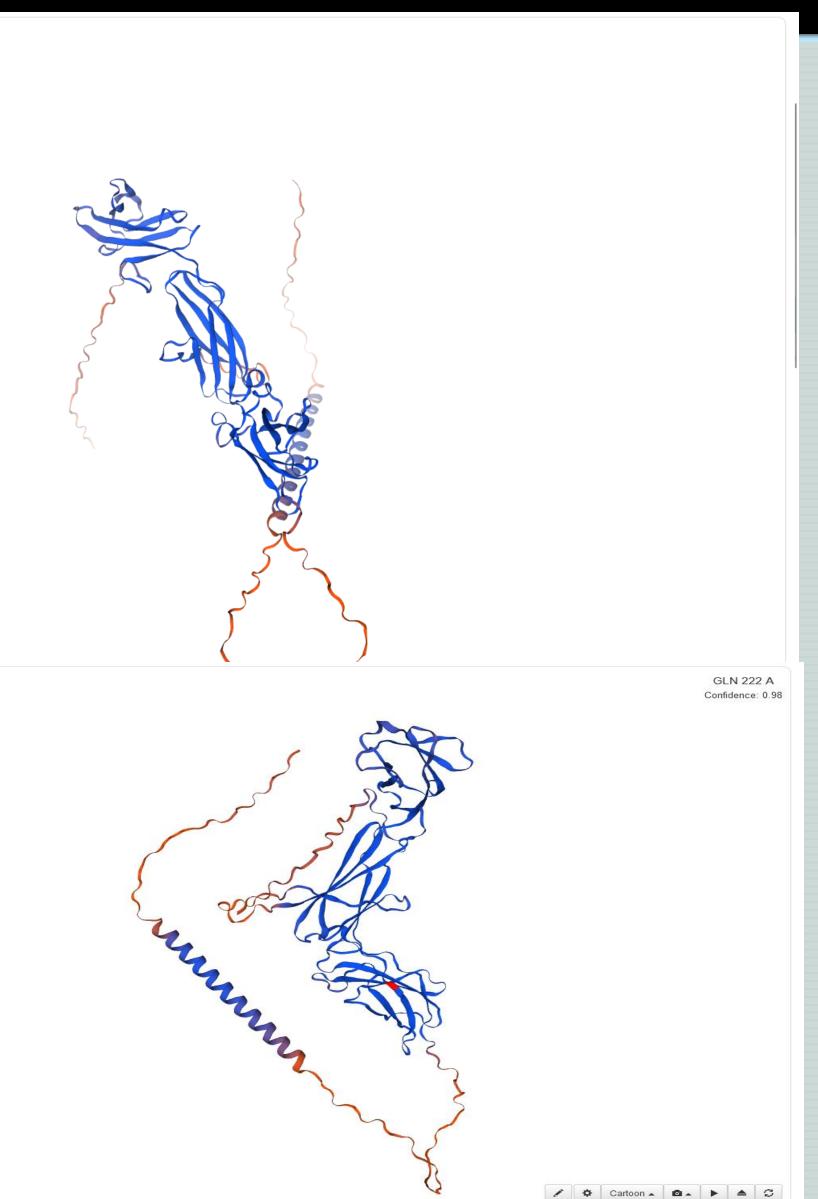


		-	Home	About	t Heip	Downie	oads Batch	query WH	ESS.db	
PolyPhen-2	report fo	or Q1	4626	P221R						
Query										
Protein Acc	Position	AA ₁	AA ₂	Description						
<u>Q14626</u>	221	Р	R	Canonical; RecN 422	Name: Full=Interleu	kin-11 recepto	r subunit alpha;	Short=IL-11 rec	eptor subur	nit alph
Results										
+ Prediction	/Confidenc	e								
HumDiv										
					This mutation is p	edicted to be	PROBABLY	DAMAGING	with a sco	ore of 1
						0.00	0,20	0,40	0.60	0.8
+ HumVa	r									
Details										
+ Multiple s	equence al	ignme	ent							
+ 3D Visuali	tation									

PolyPhen-2

Prediction/Confidence

SWISS Pro Modeling Conserved Domain



Expression IPA RNA-seg normal tissues Project title: HPA RNA-seg normal tissue PolyPhen-2 v2.2.3r40 Description: RNA-sed was performed of tissue samples from 95 human individuals representing 27 different tissues in order to determine tissue-specificity of all protein-coding gener BioProject: PRJEB4337 Publication: PMID 24309898 Analysis date: Wed Apr 4 07:08:55 201 PolyPhen-2 v2.2.3r niProtKB/UniRef100 Release 2011 12 (14-Dec-20

	S NCBI	k eTFTMKEVIYHLGQYIMAkqLYDe k dTY Conserved Q pQLADTEWWKMLWK sQLGDOMAINS/WA s SLSRADWYKRIWEVINH LOISENSULATION SH2 s SLSRADWYKRIWEVINH SH2 S SLSRADWYKRIWE									
	HOME SEARCH GUIDE	NewSearch	Structure Home	3D Macromolecular Structu	ires	Conserved Domains	Pu				
	Conserved domains on [gi 218505839 ref NP_001136256] interleukin-11 receptor subunit alpha precursor [Homo sapiens]										
Graphical summary Coom to residue level show extra options »											
	Query seq.	50	100	150 200 	250 	300	350 I I I I I				
	400 J 00 J+ —	Ig strand B 🌞 Ig	strand E 🔶 Ig strand F 年 Ia strand G 🌰								

Cytokine receptor motif Specific hits FN3 -IG like Superfamilies Ig superfamily FN3 superfamily Search for similar domain architectures 🔹 Refine search List of domain hits Name Accession Interval E-value Description Immunoglobulin like; IG domains that cannot be classified into one of IGv1, IGc1, IGc2, IG. 33-109 2.08e-0 [+] IG_like 218-310 Fibronectin type 3 domain; One of three types of internal repeats found in the plasma protein 2.50e-06

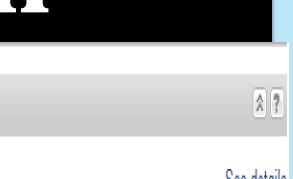
Interdomain contacts 🥼

References:

Wang J et al. (2023), "The conserved domain database in 2023", Nucleic Acids Res.51(D)384-8.

💹 Lu S et al. (2020), "The conserved domain database in 2020", Nucleic Acids Res.48(D)265-8

- 💹 Marchler-Bauer A et al. (2017), "CDD/SPARCLE: functional classification of proteins via subfamily domain architectures.", Nucleic Acids Res.45(D)200-3



ubchem BioSystems

View Concise Results

in the gene IL11RA predict that the missense mutations are probably damaging and affect protein function. Craniosynostosis is very important to study because in many cases a mutation change in certain genes can lead to the child developing craniosynostosis. This can lead to long-term complications. With further

research, we can be able to identify more genes that may have an impact on genetics.

References

. Mayo Foundation for Medical Education and Research. (2022, September 15). Craniosynostosis. Mayo Clinic. https://www.mayoclinic.org/diseasesconditions/craniosynostosis/symptoms-causes/syc-20354513

2. Centers for Disease Control and Prevention. (2023, June 28). Facts about craniosynostosis. Centers for **Disease Control and Prevention.**

https://www.cdc.gov/ncbddd/birthdefects/craniosyno stosis.html#:~:text=What%20is%20Craniosynostosis %3F-

,Craniosynostosis%20is%20a%20birth%20defect%20 in%20which%20the%20bones%20in,flexible%20mat erial%20and%20called%20sutures.

3. Craniosynostosis. Johns Hopkins Medicine. (n.d.). https://www.hopkinsmedicine.org/health/conditionsand-diseases/craniosynostosis

Acknowledgment

This project was supported by Oakwood's undergraduate research program. Special thanks to Dr. Vanterpool, PhD

Characterizing Our DNA Exceptions

HUDSONALPHA