International Journal of Advanced Research in Biological Sciences
ISSN: 2348-8069
www.ijarbs.com
(A Peer Reviewed, Referred, Indexed and Open Access Journal)
DOI: 10.22192/ijarbs Coden: IJARQG (USA) Volume 9, Issue 5 -2022

Research Article

Special Issue on Potential Applications of Bioinformatics in Biological Sciences - PABBS 2022

DOI: http://dx.doi.org/10.22192/ijarbs.2022.09.05.01.012

Single nucleotide polymorphism and structure analysis of cleft palate

R. Priya[1]*, M.Vinoth[2], Mahendran Radha[3]
Department of Bioinformatics,
Vels institute of Science and Technology & Advanced Studies, Chennai.
*Corresponding e-mail : *priya.sls@velsuniv.ac.in*

Abstract

Cleft lip and cleft palate are openings or splits in the upper lip, the roof of the mouth (palate) or both. Cleft lip and cleft palate result when facial structures that are developing in an unborn baby don't close completely. Birth defects arise from the interplay of multiple genetic and environmental factors. Although such complex traits are characterized by familial aggregation, recurrence rates within families are relatively low; the risk that an affected child will have a sibling who is also affected is typically less than 5 percent. The main objective if this project is to predict the mutant structure of a gene which has no structure. And the mutant position of a protein (G0Z349_HUMAN) by using bio informatics tools.so i identified the gene that encodes interferon regulatory factor 6 (*IRF6*) as a candidate gene on the basis of its involvement in an autosomal dominant form of cleft lip and palate. A single-nucleotide polymorphism in this gene results at several amino acid position eg: 2(A2V) A replaced by V at 2nd position. The found structure.In future if anyone research about cleft palate the below result may useful.

Keywords: IRF6-Interferon regulatory factor 6- mutant model-protein_protein interaction

Introduction

Cleft lip and cleft palate are among the most common birth defects. They most commonly occur as isolated birth defects but are also associated with many inherited genetic conditions or syndromes.

Having a baby born with a cleft can be upsetting, but cleft lip and cleft palate can be corrected. In most babies, a series of surgeries can restore normal function and achieve a more normal appearance with minimal scarring.

Materials and Methods

Materials:

NCBI:

NCBI is database which is used to charged with creating automated systems for storing and analyzing knowledge about molecular biology, biochemistry, and genetics.NCBI is now a leading source for public biomedical databases, software tools for analyzing molecular and genomic data, and research in computational biology

DbSNP

The Single Nucleotide Polymorphism Database is a free public archive for genetic variation within and across different species developed and hosted by the National Center for Biotechnology Information in collaboration with the National Human Genome Research Institute

UNIPROT

UNIPROT also called as SWISSPROT It provides an up-to-date, comprehensive body of **protein** information at a single site.It aids scientific discovery by collecting, interpreting and organising this information so that it is easy to access and use.

SIFT-The **scale-invariant feature transform** (**SIFT**) is a feature detection algorithm in computer vision to detect and describe local features in images. It was published by David Lowe in 1999.[1] Applications include object recognition, robotic mapping and navigation, image stitching, 3D modeling, gesture recognition, video tracking, individual identification of wildlife and match moving.

POLYPHEN2

PolyPhen-2 (**Poly**morphism **Phen**otyping v**2**) is a tool which predicts possible impact of an amino acid substitution on the structure and function of a human protein using straightforward physical and comparative considerations. Please, use the form below to submit your query.

PROVEAN

PROVEAN (**Pro**tein **V**ariation **E**ffect **An**alyzer) is a software tool which predicts whether an amino acid substitution or indel has an impact on the biological function of a protein.

PROVEAN is useful for filtering sequence variants to identify nonsynonymous or indel variants that are predicted to be functionally important.

STRING

In molecular biology, STRING is a biological database and web resource of known and predicted protein–protein interactions. The STRING database contains information from numerous sources, including experimental data, computational prediction methods and public text collections

HOPE

is an easy-to-use web service that analyses the structural effects of a point mutation in a protein sequence. Input your protein sequence and the mutation and HOPE will collect and combine available information from a series of web services and databases and will produce a report, complete with results, figures and animations.

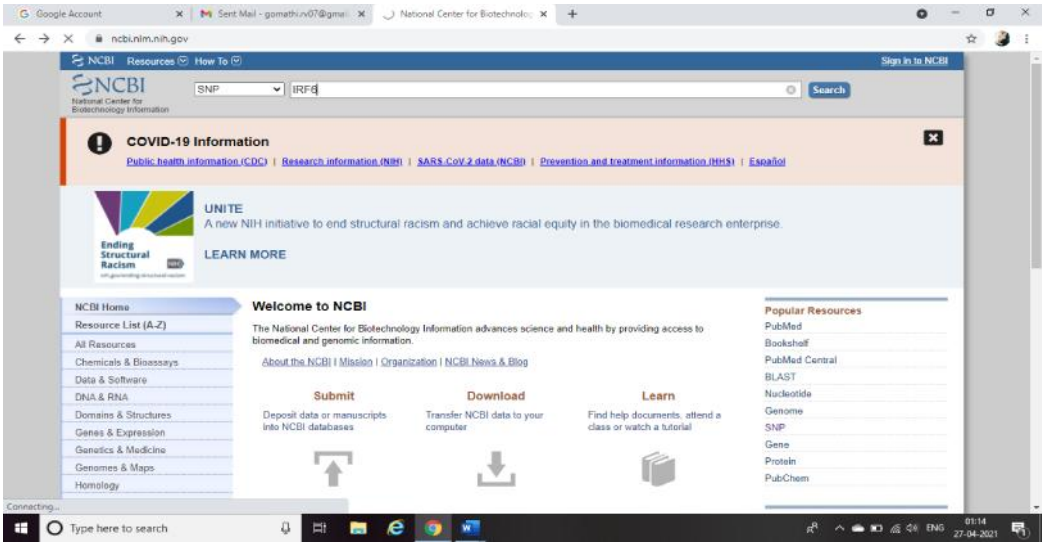
SWISSMODEL

SWISSMODEL is the online web server dedicated to homology modeling of 3D protein structures. Homology modeling is currently the most accurate method to generate reliable three-dimensional protein structure models and is routinely used in many practical applications.

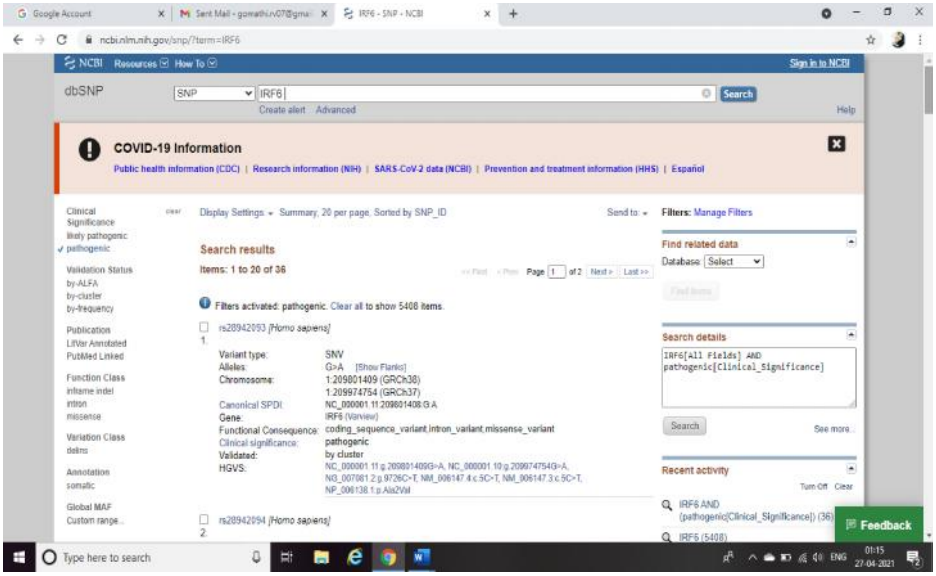
Methods:

1. Target Selection:

Target protein is identified in NCBI database which shows

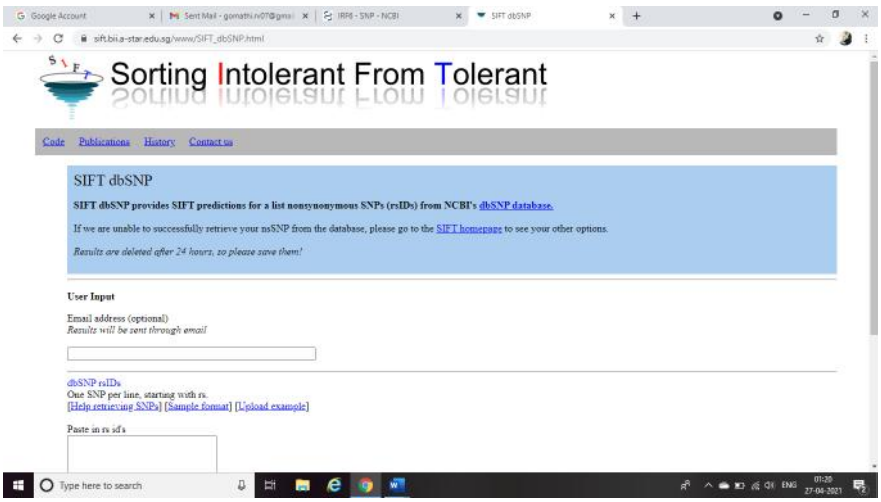


2. dSNP database Selection

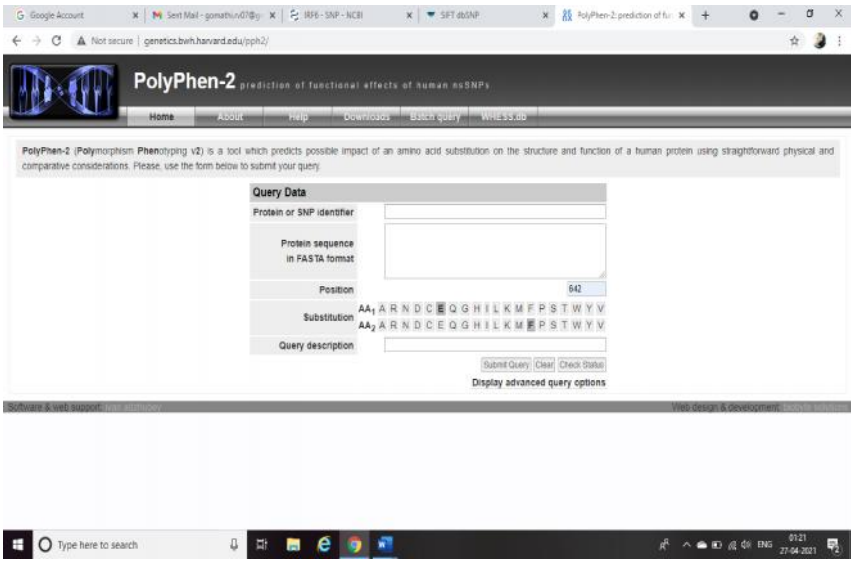


3. Single Nucleotide Polymorphism Analysis

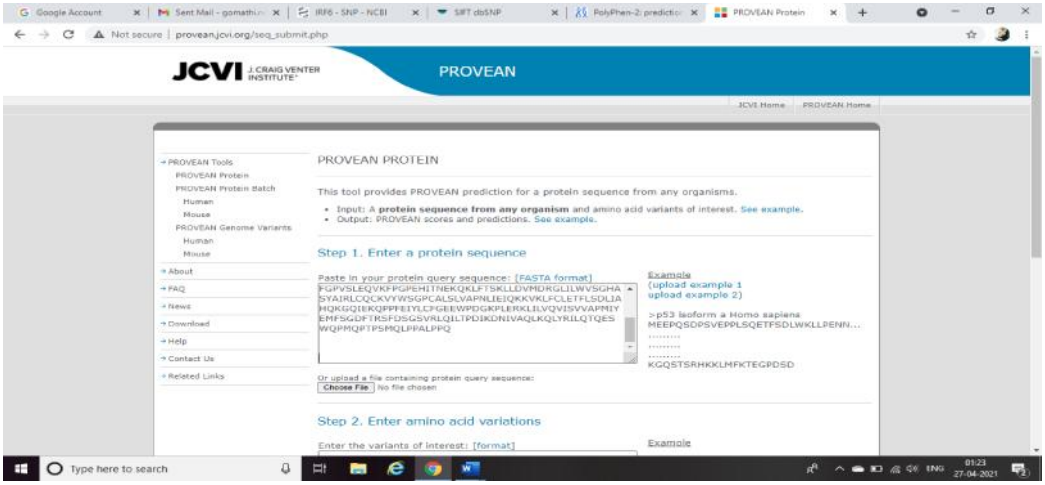
a. SIFT-Aminoacid Changes



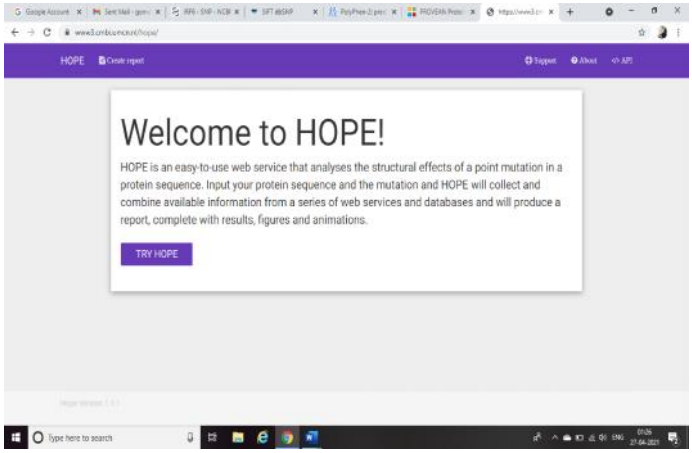
b. POLYPHEN – Mutation identification



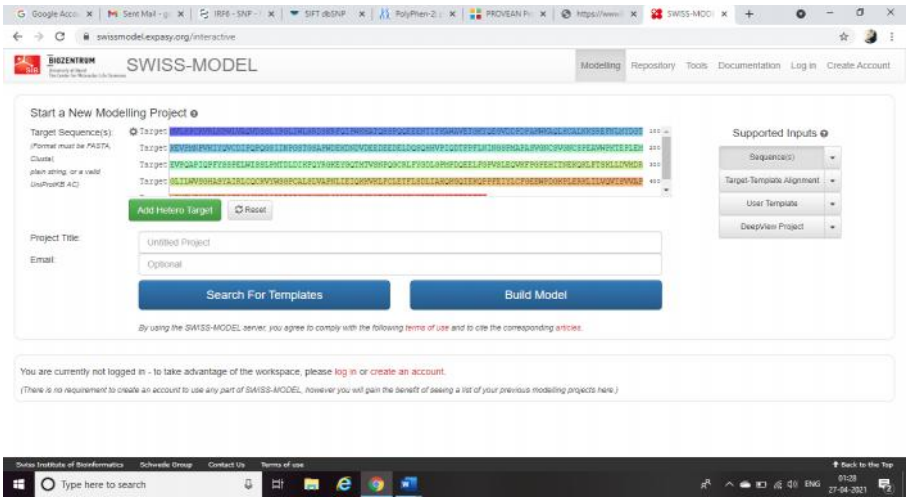
c.PROVEAN- Identification of Single mutated Base



4. HOPE
5.



6. SWISS MODEL



Results and Discussion

SNP *irs id:*
rs28942093
rs28942094
rs28942095
rs121434224
rs121434225
rs121434226
rs121434227
rs121434228
rs121434229
rs121434230
rs121434231
rs200166664
rs387906967
rs387906968
rs397515434
rs587776569
rs769068305
rs886038202
rs886039388
rs886039389
rs886039390
rs886039391
rs886039570
rs886041484
rs1057520168
rs1057520569
rs1057520738
rs1060499555
rs1064797000
rs1553247595
rs1553247602
rs1553247774
rs1553248638
rs1553248641
rs1571979802
rs157198629

Sift result:

SNP	AMINO ACID CHANGE	GENE ID	TRANSCRIPT ID	PROTEIN ID	SIFT SCORE	SIFT PREDICTION
rs28942093	A2V	ENSG00000117595	ENST00000367021	ENSP00000355988	0.017	DELETERIOUS
rs28942094	R6C	ENSG00000117595	ENST00000367021	ENSP00000355988	0	DELETERIOUS
rs28942095	R305W	ENSG00000117595	ENST00000542854	ENSP00000440532	0	DELETERIOUS
rs28942095	R400W	ENSG00000117595	ENST00000367021	ENSP00000355988	0.001	DELETERIOUS
rs121434226	R84C	ENSG00000117595	ENST00000367021	ENSP00000355988	0	DELETERIOUS
rs121434227	R84L	ENSG00000117595	ENST00000367021	ENSP00000355988	0	DELETERIOUS
rs121434227	R84H	ENSG00000117595	ENST00000367021	ENSP00000355988	0	DELETERIOUS
rs121434229	R45Q	ENSG00000117595	ENST00000367021	ENSP00000355988	0.01	DELETERIOUS
rs121434230	P396S	ENSG00000117595	ENST00000367021	ENSP00000355988	0.037	DELETERIOUS
rs121434230	P301S	ENSG00000117595	ENST00000542854	ENSP00000440532	0.045	DELETERIOUS
rs121434231	R339I	ENSG00000117595	ENST00000367021	ENSP00000355988	0	DELETERIOUS
rs121434231	R244I	ENSG00000117595	ENST00000542854	ENSP00000440532	0	DELETERIOUS
rs200166664	R305P	ENSG00000117595	ENST00000542854	ENSP00000440532	0	DELETERIOUS
rs200166664	R400P	ENSG00000117595	ENST00000367021	ENSP00000355988	0.003	DELETERIOUS
rs387906968	S329L	ENSG00000117595	ENST00000542854	ENSP00000440532	0	DELETERIOUS
rs387906968	S424L	ENSG00000117595	ENST00000367021	ENSP00000355988	0.004	DELETERIOUS

Polyphen2 result:

RS id	Amino acid change	probability	Score
rs28942093	A2V	Possibly damaging	1.00
rs28942094	R6C	Possibly damanging	1.00
rs28942095	R400W	Probably damaging	0.99
rs121434226	R84C	Probably damaging	0.99
rs121434227	R84L	Possibly damaging	1.00
rs121434227	R84H	Probably damaging	0.99
rs121434229	R45Q	Possibly damaging	1.00
rs121434230	P396S	Probably damaging	0.99
rs121434231	R339I	Probably damaging	0.99
rs200166664	R400P	Probably damaging	0.99
rs387906968	S424L	Possibly damaging	1.00

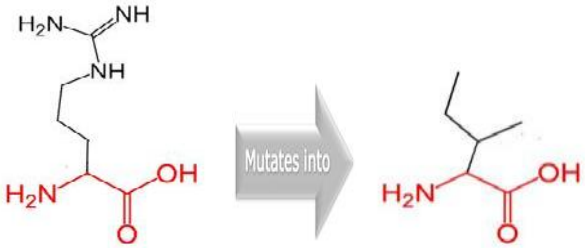
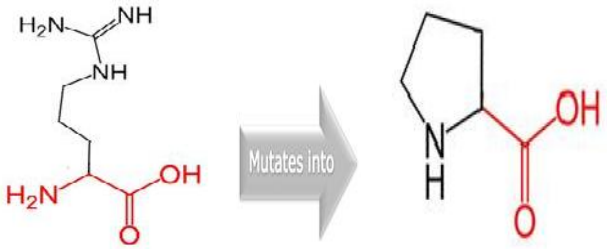
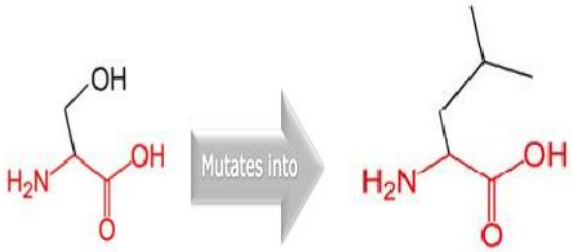
Sensitivity-0.14
Specificity-0.99

Provean result:

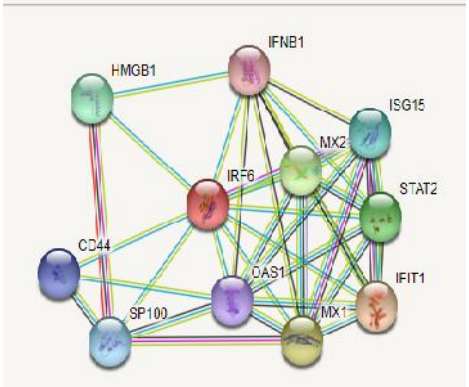
Variant	Provean score	Prediction (cutoff=-2.5)
A2V	-2.076	Neutral
R6C	-3.559	Deleterious
R400W	-6.388	Deleterious
R84C	-7.156	Deleterious
R84L	-6.229	Deleterious
R84H	-4.339	Deleterious
R45Q	-2.061	Neutral
P396S	-6.211	Deleterious
R339I	-7.329	Deleterious
R400P	-5.513	Deleterious
S424L	-3.505	Deleterious

HOPE RESULT:

Rs ID	Amino acid change	Images	Function impact
rs28942093	A2V		Highly
rs28942093	R6C		High
rs28942093	R400W		Medium
rs121434226	R84C		High

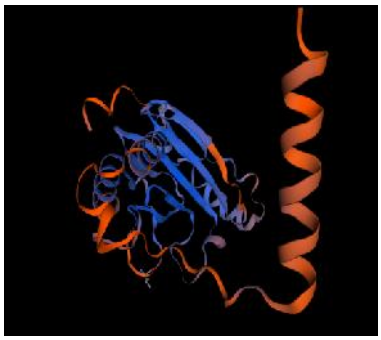
rs121434227	R84L		Low
rs121434227	R84H		Low
rs121434229	R45Q		Medium
rs121434230	P396S		High
rs121434231	R339I		Medium
rs200166664	R400P		High
rs387906968	S424L		Low

String result:

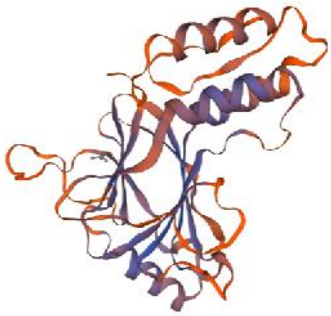


number of nodes:11
number of edges:37
average node degree:6.73
avg. local clustering coefficient:0.833
expected number of edges:12
PPI enrichment p-value:3.07e-09

Swiss model result:



Normal pdb structure



mutant pdb structure

Conclusion

In this study I analyzed snp and predicted the mutant structure of gene (IRF6) and the postion where it get mutated by using above tools. In future if anyone predict about cleft palate snp analysis this may useful.

References

1. Shi B, Deng D, Wang H. An experimental study of the growth pattern and mechanisms of surgically induced cleft palate and palatoplasty on maxillary growth in dogs. *West China J. Stomatol.* 1997;**15**:151–155.
2. Sarnat BG. Palatal and facial growth in macaca rhesus monkeys with surgically produced palatal clefts. *Plast. Reconstr. Surg.* 1958;**22**:29–41. doi: 10.1097/00006534-195807000-00003
3. Weinzweig J, et al. The fetal cleft palate: I. Characterization of a congxenital model. *Plast. Reconstr. Surg.* 1999;**103**:419–428. doi: 10.1097/00006534-199902000-00009
4. Martinez SE, et al. A new technique for feeding dogs with a congenital cleft palate for surgical research. *Lab. Anim.* 2011;**45**:70–80. doi: 10.1258/la.2010.010044

5. Bardach J, Kelly KM, Sarn. Does interference with mucoperiosteum and palatal bone affect craniofacial growth? An experimental study in beagles. *Plast. Reconstr. Surg.* 1990;**86**:1101–1102. doi: 10.1097/00006534-199012000-00008
6. Longaker MT, Dodson TB, Kaban LB. A rabbit model for fetal cleft lip repair. *J. Oral Maxillofac. Surg.* 1990;**48**:714–719. doi: 10.1016/0278-2391(90)90056-8
7. Djasim UM, Hekking-Weijma JM, Wolvius EB, van Neck JW, van der Wal KG. Rabbits as a model for research into craniofacial distraction osteogenesis. *Br. J. Oral Maxillofac. Surg.* 2008;**46**:620–624. doi: 10.1016/j.bjoms.2008.04.002]
8. Bardach J, Kelly KM. Role of animal models in experimental studies of craniofacial growth following cleft lip and palate repair. *Cleft Palate J.* 1988;**25**:103
9. Koo SH, Cunningham MC, Arabshahi B, Gruss JS, Grant JH. The transforming growth factor-β3 knock-out mouse: an animal model for cleft palate. *Plast. Reconstr. Surg.* 2001;**108**:938–951. doi: 10.1097/00006534-200109150-00018
10. Kouichi Y, et al. Central distribution of neuronal cell bodies innervating the levator veli palatini muscle and associated pattern of myosin heavy chain isoform expression in rat. *Brain Res.* 2003;**968**:80–88. doi: 10.1016/S0006-8993(02)04273-7.
11. Vinoth, M., and Natarajan, B., "Isolation, characterization, antibiological and anticancer activity of Tetrahydrobenzochromone glycosides from leaf of Melochia corchorifolia." Rasayan Journal of Chemistry, 14(2), pp. 768-777, 2021.
12. Vinoth, M., and Natarajan, B., "Isolation, structural elucidation of flavonolignan derivative from Melochia corchorifolia - Evaluation of their biological, anticancer activity, and molecular docking studies." Rasayan Journal of Chemistry, 14(3), pp. 166-1679, 2021.
13. Vinoth, M., and Natarajan, B., and ShanmugaSundaram, C., "Characterization and evaluation ethyl acetate extract of Melochia corchorifolia leaf- anticancer antibiological and molecular docking studies on breast cancer estrogen receptor." Indian Journal of Chemical Technology, 28(2), pp. 188-196, 2021.
14. Vinoth, M., and Natarajan, B., "Evaluation of phytochemical, anthelmintic activity and Antidiabetic activity-Melochia corchorifolia ethanol extract." International Journal of Research in Pharmaceutical Sciences, 12(1), pp. 1-6, 2021.

Access this Article in Online

Website: www.ijarbs.com

Subject: Bioinformatics

Quick Response Code

DOI:10.22192/ijarbs.2022.09.05.01.012

How to cite this article:
R. Priya, M.Vinoth, Mahendran Radha. (2022). Single nucleotide polymorphism and structure analysis of cleft palate. Int. J. Adv. Res. Biol. Sci. 9(5): Special Issue 1: 108-118.
DOI: http://dx.doi.org/10.22192/ijarbs.2022.09.05.01.012