



a collaborative rare
disease report
generator
empowering
clinicians and
patients

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Introduction

- We introduce **RareInsight**, an **open-source**, interactive **dashboard** designed for clinicians and patients to address the knowledge gaps in **rare diseases**.
- With a focus on **collaborative** research, RareInsight aims to transform the **interpretation** of genetic variant data into **detailed reports**, fostering improved diagnostic and therapeutic outcomes in the challenging landscape of rare disease diagnosis and research.

Methods

- Customizable and interactive reports from **nf-core/raredisease VCF files**
- VCF files from whole **genome** or whole **exome** sequencing,
- Advanced filtering and diverse export **options**.
- Developed using **Shinydashboard** and tested with data from **dbGaP**
- Clinical relevance, genetic implications, and ACMG classification.
- **Resources** for patients and clinicians
- **Open-source**

Conclusion

RareInsight stands at the forefront of reshaping the landscape of rare disease diagnosis and research, fostering **collaboration** and **innovation** to elevate healthcare outcomes within the framework of an **open-source** platform.

The dashboard

Dashboard home page

Aim

By promoting knowledge exchange, RareInsight aims to :

- 1- Simplify the complex process of diagnosing rare diseases and improving therapeutic outcomes.
- 2- Processes genetic variant data into customizable, interactive reports.
- 3- Ensures accuracy and reliability.
- 4- Facilitating seamless sharing and collaboration on reports.
- 5- Enrich the collective understanding of rare diseases
- 6- Enhance healthcare outcomes in an open-source format.
- 7- Revolutionize rare disease diagnosis and research by leveraging collaboration and innovation, ultimately reshaping the landscape of healthcare.

Search Panel

Search Type: Gene

Enter Search Term: COL1A1

Search

Show 10 entries

X.AlleleID	Type	Name	GenelD	GeneSymbol	HGNC_ID	ClinicalSignificance	ClinSigSin
31454	single nucleotide variant	NM_000088.4(COL1A1):c.824G>A (p.Gly275Asp)	1277	COL1A1	HGNC:2197	Pathogenic	
31455	single nucleotide variant	NM_000088.4(COL1A1):c.824G>A (p.Gly275Asp)					

VCF Panel

Upload VCF File

Browse... justhusky_clinical_snv.ann_filter.vcf

Upload complete

Results Graphs

CHROM	POS	REF	ALT	QUAL	FILTER	AF	AQ
21	35273121	C	T	1	NA	0.333333	1
21	35273125	A	T	0	NA	0.333333	T[downstream_gene_vari
MT	596	T	A	NA	base_qual;clustered_events;strand_bias	.	.
MT	605	T	A	NA	base_qual;clustered_events;strand_bias;weak_evidence	.	.
MT	610	T	A	NA	base_qual;clustered_events;strand_bias;weak_evidence	.	.

Patient Support

This is the content for Patient Support.

Diagnostic report panel