



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.
- Open-source**
- Data **privacy** and security ensured

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

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About us

Services

Input User Information

Search Panel

VCF analysis

Diagnostic report

Acknowledgement

Summary

RareInsight is a project aimed at tackling the challenges posed by rare diseases through collaborative efforts in research. Developed as an open-source, interactive dashboard tailored for both clinicians and patients, RareInsight focuses on transforming the interpretation of genetic variant data into customizable, interactive reports, ideally generated by nf-core's raredisease pipeline. These reports are derived from whole genome or whole exome sequencing data and offer advanced filtering options, statistical analysis capabilities, and diverse export formats. Developed using Shiny and tested with data from respected sources such as the Undiagnosed Disease Program and NHGRI GREGOR Consortium datasets found in dbGaP, RareInsight ensures accuracy and reliability. RareInsight aids informed decision-making for clinicians and patients alike and fosters collaboration among researchers and clinicians, facilitating seamless sharing and collaboration on reports. By promoting knowledge exchange, RareInsight aims to enrich the collective understanding of rare diseases and enhance healthcare outcomes in an open-source format. In summary, RareInsight aims to revolutionize rare disease diagnosis and research by leveraging collaboration and innovation, ultimately reshaping the landscape of healthcare.

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.

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VCF Panel

Upload VCF File

Browse... justusky_clinical_snv.ann_filter

Upload complete

Variant Information

CHROM	POS	ID	REF	ALT	QUAL	FIL
21	35273121	21_35273121_C_T	C	T	1	NA
21	35273125	21_35273125_A_T	A	T	0	NA
MT	596	NA	T	A	NA	ba
MT	605	NA	T	A	NA	ba
MT	610	NA	T	A	NA	ba
MT	750	NA	A	G	NA	PA
MT	1189	NA	T	C	NA	PA
MT	1438	NA	A	G	NA	PA
MT	1811	NA	A	G	NA	PA

User Data Inputs

Name

Surname

Ethnicity

Doctor

Date of Birth

2024-02-20

Clinical Diagnosis (OMIM)

Phenotype (HPO Terms)

Test Performed

WES

Download User Info

Search Panel

Search Type:

Gene

Enter Search Term:

COL1A1

Search

Show 10 entries

Search:

PhenotypeList	Origin	OriginSimple	Assembly	ChromosomeAccession	Chromosome	Start	Stop
Osteogenesis imperfecta, recessive perinatal lethal	germline	germline	GRCh37	NC_000017.10	17	48274012	482740
Osteogenesis imperfecta, recessive perinatal lethal	germline	germline	GRCh38	NC_000017.11	17	50196651	501966