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