# a collaborative rare disease report generator empowering clinicians and patients

**KC Coetzer1, OI Awe2,3**

1 Division of Molecular Biology and Human Genetics, Department of Biomedical Sciences, Faculty of Medicine and Health Sciences, Stellenbosch University, Cape Town, South Africa
2 African Society for Bioinformatics and Computational Biology, Cape Town, South Africa
3 Department of Computer Science, University of Ibadan, Ibadan, Oyo State, Nigeria

## 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