Zetta Genomics

Case Study: The MRC Human Genetics Unit at the University of Edinburgh

THE UNIVERSITY of EDINBURGH

Future proofing our genomic data needs.

At a glance

The MRC Human Genetics Unit - part of the Institute of Genetics and Cancer - at the University of Edinburgh aims to transform genomic research and ultimately, clinical outcomes by identifying genetic variants in rare diseases. The MRC Human Genetics Unit currently hold around 1,000 whole genome sequences, but as their project progresses this could rise significantly to >50,000. Their aim is to not only discover which genes cause rare disease but also how the mechanism of these genes cause disease.

Customer Feedback

"Using XetaBase, our potentially weeks-long process will be reduced to minutes – allowing us to store, analyse and constantly re-interrogate genomic data at scale and speed. Research groups can now define their own needs and refine results using the self-service web browser. It reduces time, resource, cost and – critically – gets the results of analysis to clinicians more quickly than ever before."

Senior Research Fellow & Bioinformatics Analysis Core Manager, MRC Human Genetics Unit, University of Edinburgh

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Find out more here

CHALLENGES

Existing genomic data management and interpretation systems aren't built with the needs of genomic data in mind. Mostly file based, genomic data is stored as a static artefact – It's difficult to access, annotate, re-analyse and share – so the potential for additional insight and discovery is wasted.

The MRC Human Genetics Unit wanted to be able to liberate this genomic data and to create dynamic and trusted research environments. Essentially, they needed a genome-optimised data management solution; one that is designed to store, easily access, analyse and constantly re-interrogate data at scale and speed.

SOLUTIONS

XetaBase

XetaBase - A pioneering genomic-native data management system that's built on proven technology, easily interrogated by bioinformaticians and non-bioinformaticians alike, copes with large volumes of data at speed – and makes this data securely accessible. Adopting a single, interoperable platform allows the ability to plug in a wide variety of information sources

De-slio and access genomic data at scale and in real-time

Analyse variants within and between studies to cross compare cohorts

Data use is smarter - Now dynamic rather than an artefact

BENEFITS

1 Scale

XetaBase is built on a proven technology platform, designed specifically for genomic data management at scale. As the WGS data need increases the MRC Human genetics unit now has confidence that the data interpretation platform can scale as required to support research needs

2 Data access

The MRC Human genetics unit can now de-silo and access huge volumes of currently under-utilised genomic data - Customisable APIs mean they can access this data from virtually anywhere

3 Clinical insight and re-analysis

Pioneering indexing architecture within the platform means they can rapidly access data and automatically synchronise variant annotation – removing time intensive duplication of data annotation across multiple projects