RD Connect

Join RD-Connect and solve your rare disease cases

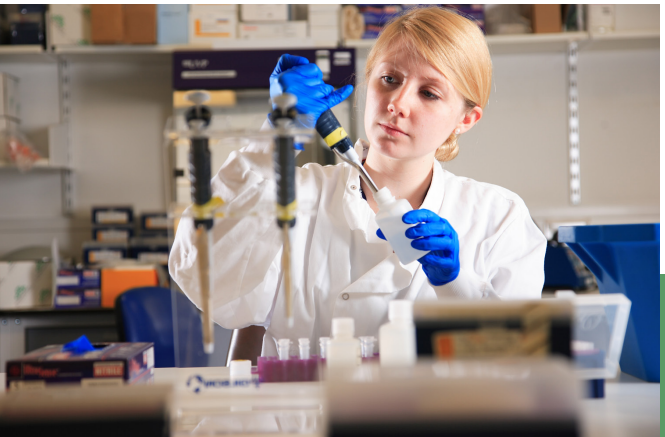
The RD-Connect Genome-Phenome Analysis Platform is free of charge and open to any rare disease researchers willing to share their sequencing and phenotypic data.

RD-Connect provides both a centralised data repository and a user-friendly online analysis system to registered users.

Whole-genome, exome and gene panel data are securely shared within the platform, and transferred to the ***European Genome-Phenome Archive*** (EGA) for archival. Raw genomic data is fully reprocessed through a standardised analysis pipeline, while associated phenotypic descriptions are recorded in ***PhenoTips*** using ***HPO***, ***OMIM*** and ***ORDO***, permitting machine-readable querying. Results are made available through the genomics analysis interface, which enables filtering and prioritisation of variants by inheritance mode, location/gene(s) of interest, variant impact, pathogenicity prediction, control population frequencies, and phenotype-gene associations using ***Exomiser***/***OMIM***.

This allows users to perform entire primary genomic analyses of their own patients online, compare findings with other submitted cohorts, and find similar cases using ***Matchmaker Exchange*** (MME, matchmakerexchange.org).

The platform already includes several thousand cases from partner projects such as ***NeurOmics*** (rd-neuromics.eu) and ***BBMRI-LPC*** (bbmri-lpc.org).



info site

platform site

Funded by the European Union

What do you need to join?

- Rare disease cases/families for which the Informed Consent explicitly allows sharing for research purposes
- Raw NGS sequencing data available to be shared (FastQ, BAM, CRAM formats)
- Be prepared to input the associated phenotypic description for your samples into the easy-to-use RD-Connect PhenoTips instance

What are the benefits of joining?

- A full (re)analysis of your data using the RD-Connect standard analysis pipeline
- Full access to the RD-Connect Genome-Phenome Analysis Platform, allowing you to perform your own variant analysis and prioritisation, once your samples have been processed and uploaded, including an optional six-month embargo period if requested
- Access to genotypic and phenotypic information from thousands of other samples within the platform, which may provide a match for your own unsolved rare disease cases
- Access to MME to allow you to look for matches across the globe

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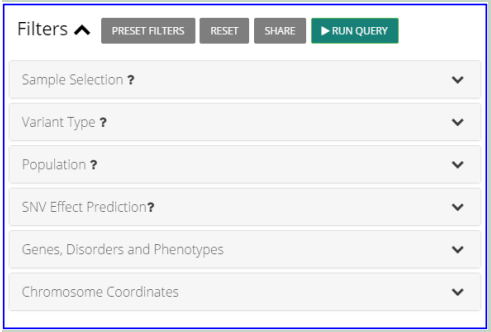
Servers at CNAG

Further information

- You can test the genomics analysis interface using 50x WGS data from a HapMap trio, processed using by going to ***platform.rd-connect.eu*** and selecting “Try out the Genomics Platform”
- If you have any further questions regarding participation, please send an e-mail to ***platform@rd-connect.eu***



RD-Connect User Interface

rdconnect

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