Congenica®

Enabling genomic medicine

Clinical decision support software + Clinical consulting services

Enabling comprehensive, sample-to-report carrier screening

Carrier screening for inherited disorders is often performed for the mother only and is often restricted to a limited set of individual disease-causing variants known to have an elevated carrier rate in a particular population. This approach gives an incomplete picture of carrier risk and has the potential to provide a false sense of confidence for couples planning for a family – particularly in geographical areas with a dense population of individuals with shared ancestry.

Using NGS technology enables enhanced screening, in both the mother's and the father's genome, for the presence of disease-causing variants. Working independently, or in collaboration with our clinical services team, the resulting data is efficiently analysed and interpretated using Congenica's CE marked IVD software*. The result is a comprehensive carrier screening report that provides greater actionable knowledge and higher family planning confidence for the couple.

> "Congenica is an absolute game changer
>
> **Dr Tessa Homfray**
> Medical Genetics Consultant, NHS

Full support from sample to report

We want your lab to be successful now as well as in the future. We work with you to provide as much or as little support as you need to meet demand and scale your operations.

Sequencing
Securely upload your own sequenced data or let us facilitate access to clinical grade sequencing through our accredited and certified partners

Secondary analysis
Upload FASTQ files directly and we'll perform variant calling. Or use your own in-house or 3rd party pipeline for variant calling and upload the VCF output

Tertiary analysis
Our clinical services team creates computational models for the couples potential offspring and uploads the resulting VCF files

Interpretation
Interrogate the data directly or leverage our clinical services team to perform a first pass check of the data or to create a final report

Reporting
Generate a customizable report for export and presentation of the results to the ordering clinician

The couple's genetic data is used as input. Principles of autosomal recessive and X-linked inheritance are used to computationally construct genetic models of the couple's potential offspring.

Data for both of the parents as well as the modeled data for their potential offspring are uploaded to the Congenica clinical decision support platform for interpretation.

*CE marked under IVDD 98/79/ec and available for IVD use where this is recognized

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www.congenica.com

Why choose the Congenica platform?

Enabling best-in-class analysis, interpretation and reporting of NGS data, the CE marked IVD platform* provides:

- **Standardized clinically-approved workflows**
  Ensure consistent application of ACMG and ACGS recommended SOPs
- **Support for pre-set virtual gene panels and curated variant lists**
  Make your analysis as broad or as specific as you want
- **Automated interpretation of known variants**
  Interpret variants you've seen before in as little as 5 minutes
- **Auto-ACMG classification of unknown variants**
  Save significant time interpreting variants you haven't previously seen
- **Full audit trail**
  Easily maintain complete records for proof of CAP and CLIA compliance

Why Congenica's clinical services team?

Our team has worked with organizations throughout the UK and Middle East including the UK's National Health Service, Royal College of Surgeons Ireland, Sidra Medical and Research Center, Viafet Genomics Laboratory, and National Genetic Centre at Royal Hospital Oman.

Skilled UK team

Our team is staffed with UK-based, HCPC-registered Clinical Scientists ready to help with support and training

Industry leading experts

We have extensive rare disease expertise having interpreted thousands of exome and genome cases, including for the Genomics England 100K genome project

Quality assurance

Our work has been independently assessed by GenQA and awarded full marks every year since 2016, ensuring the highest quality genomic data analysis

> "We have utilised the knowledge and skills of Congenica and their Clinical team to implement new innovative genetics services in the NHS. Using Congenica software and working collaboratively with their experienced Clinical team has significantly improved the services we provide to patients. We cannot recommend Congenica highly enough.

Professor Sahar Mansour
St George's University Hospital, London

Partner with Congenica

Discover how easy it is to provide comprehensive carrier screening when you partner with Congenica. Talk to a clinical expert today.

www.congenica.com/cis
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Request a consultaton
www.congenica.com/consult

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