Congenica®

Rare Disease

Resolving the interpretation bottleneck

Rapid diagnoses to improve patient outcomes

With the increasing understanding that most rare diseases are caused by genetic mutations, next generation sequencing (NGS) has become the standard method for diagnosis. As a result, the demand for clinical interpretation has drastically increased putting significant pressure on healthcare services to deliver comprehensive clinical reports in a timely manner.

To support clinicians and healthcare providers in providing life-changing answers fast and with the highest efficiency, accuracy & confidence, Congenica has developed a state-of-the-art, AI-powered Clinical Decision Support Platform for rapid analysis and interpretation of often complex sequencing data.

- ✓ Deliver life-changing answers fast
- ✓ Increase case throughput and diagnostic yield
- ✓ Reduce workload, analysis times and costs
- ✓ Comply with regulatory and clinical best practice

“Congenica’s combination of flexible workflows and data visualisation tools have helped provide a diagnosis where other approaches have failed. This is a powerful tool that has helped a family bring eight years of turmoil to a close.”

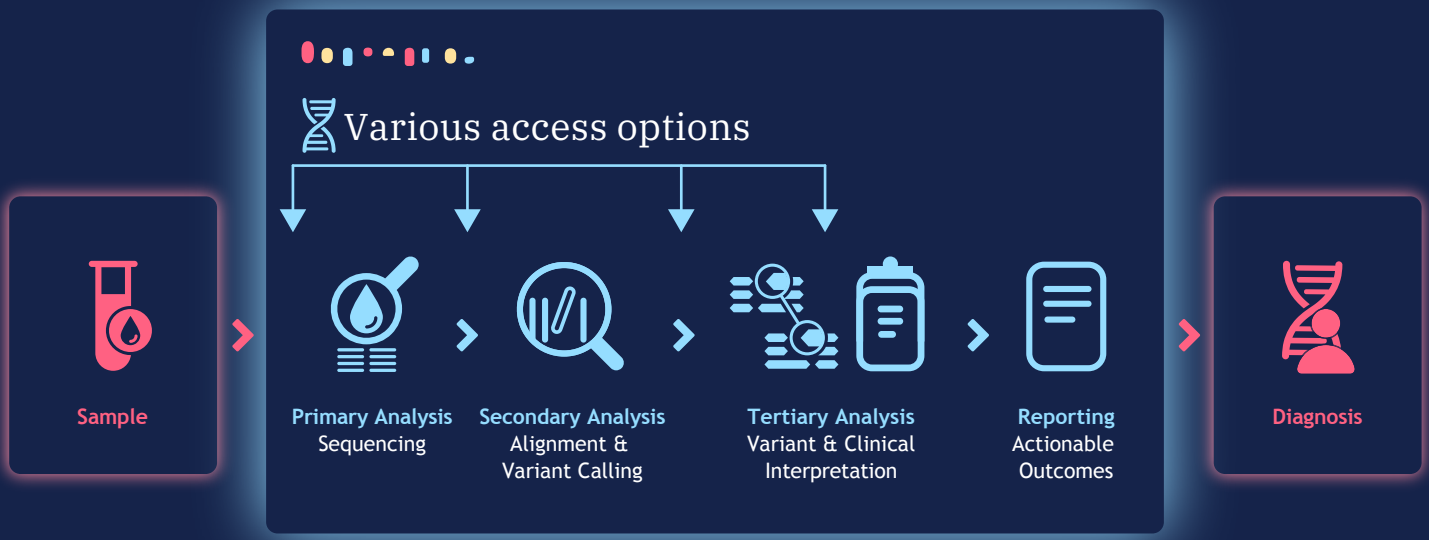
Professor Sahar Mansour
Consultant Clinical Geneticist and Physician, St George’s University Hospitals NHS Foundation Trust

Transforming sequencing data into actionable insights fast

Congenica's highly flexible, scalable and sequencer-agnostic CE-marked solution enables the analysis of NGS panels, exomes and whole genomes to the highest international standards.

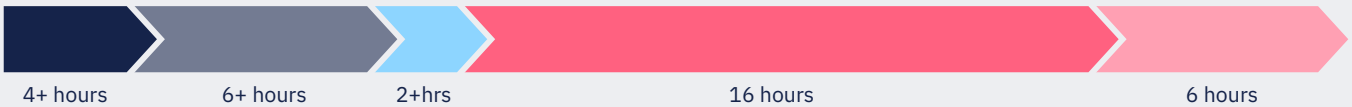
Processes and workflows can be fully automated for instant interpretation of known causal variants, semi-automated to include novel investigations for selected variants of interest, or executed as deep-dive reviews for complex cases.

Whether starting from FASTQ files, or uploading BAM and VCF files from existing secondary and tertiary pipelines into the platform, Congenica offers a broad range of high-quality analysis and variant interpretation options to deliver life-changing answers fast.



Without Congenica - Standard workflow: **22 hours** to complete tertiary analysis & reporting

Average times from 400 whole genome samples in Oxford University Hospital laboratory [1]



Congenica

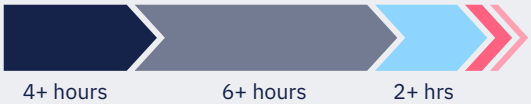
Average times with Congenica based on 2,000 whole genome samples

Congenica

30 minutes
from data to report

Congenica Express (known variants)

Average times with Congenica based on 4,000 whole genome samples



Congenica

5 minutes
from data to report

- DNA Sample
- Library prep
- Sequencing
- Tertiary analysis
- Reporting

1. Schwarze, K. et al,. 2020. Genet Med 22, 85–94

Features & benefits

Rapid turnaround time

Automated processes for fast and accurate decision-making in as little as 5 minutes

High diagnostic yield

30% higher diagnostic yield compared to our competitors due to expertly curated reference databases, automated variant prioritisation and AI driven variant identification

Exceptional flexibility

Support of gene panels, exomes and whole genomes, short or long-read sequencing, bespoke platform integrations or on-premise solutions to meet individual requirements

CE-IVD certified

Validated, accurate and secure platform for high confidence in diagnostic outcomes

Delivering results at scale

Genomics England has been successful in delivering the groundbreaking 100,000 Genomes Project and establishing the world's first national health service to offer whole genome sequencing. As partner to Genomics England and the exclusive Clinicial Decision Support partner for the NHS Genomic Medicine Service, we have adapted our platform to drive the analysis of whole genomes at national scale.

>200
whole genomes per day

50%
increase in diagnostic yield

20 fold
reduction in analysis times

95%
less manual processing time

Enabling world-class genomic medicine services

“Congenica has been able to process a huge number of samples for the 100,000 Genomes Project and routinely process thousands of samples for Genomics England every month. Working with Congenica we’ve been able to provide high quality variant interpretation of genome sequences to the NHS, helping deliver benefits to patients at scale.”

Dr Augusto Rendon
Director of Bioinformatics of Genomics England

Get in touch to learn more

www.congenica.com/contact-us
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Congenica is available as a CE Marked IVD clinical decision support platform for clinical diagnostic use for inherited genetic disorders in the UK and EU, Iceland, Lichtenstein, Norway, Switzerland and Turkey. In all other countries, ensuring compliance with relevant local, national and international clinical laboratory regulations is the responsibility of the laboratory.