Congenica®

Rare Disease

Resolving the interpretation bottleneck

Rapid analysis to improve actionable outcomes

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

To support 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 platform for the rapid analysis and interpretation of often complex sequencing data.

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

“At Congenica, we understand how to transform complex genetic data into actionable insights to drive precision medicine at scale.”

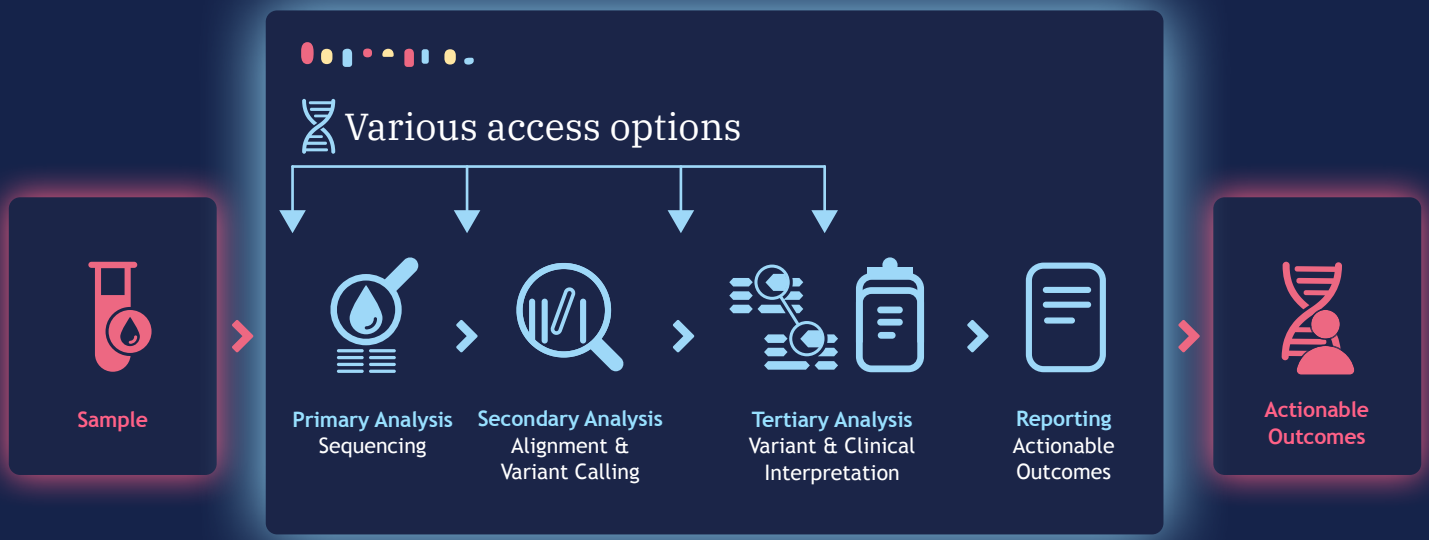
Robert Denison,
CEO at Congenica

Transforming sequencing data into actionable insights fast

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

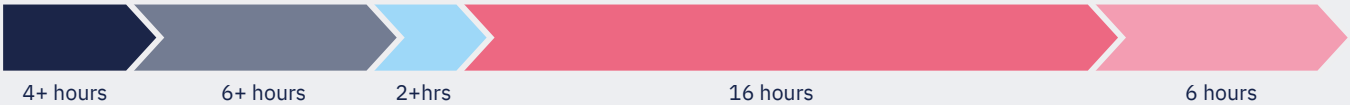
Processes and workflows can be fully automated for instant interpretation of known causal variants, semi-automated to include novel investigations for selected variants of interest, or executed as deep-dive reviews for complex cases.

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 help healthcare providers deliver life-changing answers fast.



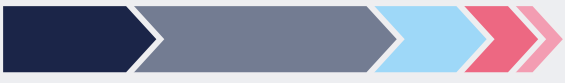
Without Congenica - Standard workflow: **22 hours** to complete tertiary analysis & reporting

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

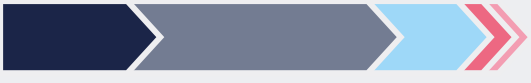


Congenica

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 to aid fast and accurate decision-making in as little as 5 minutes

High analytical yield

30% higher analytical 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

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 analysis 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 analytical yield

20 fold
reduction in analysis times

95%
less manual processing time

Get in touch to learn more

www.congenica.com/contact-us
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@congenica

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.