From Data to Diagnosis - Delivering Results on a National Scale with Genomics England

Enabling a national program to process over 2,700 whole genomes per week and increase diagnostic yield by 50%

Congenica

Enabling genomic medicine

Executive Summary

Establishing the world's first national genomic medicine program was always going to be a step into the unknown and pose significant challenges. Recognizing this, the UK Government's Department of Health and Social Care set up Genomics England. It successfully completed the ambitious 100,000 Genomes Project thanks to partnerships with a range of genomics tools providers including Congenica.

Genomics England is now enabling genomic testing in the UK to be provided through a single national testing network, the NHS Genomic Medicine Service. The service aims to sequence 500,000 whole genomes in its first five years. Following a rigorous evaluation by an independent panel of experts, the Congenica platform was selected as the exclusive clinical decision support solution for the service.

The Congenica clinical decision support platform rapidly transforms whole genome, whole exome and gene panel data into actionable information, enabling clinicians to improve diagnostic yield and case throughput, maximize workflow efficiency and increase confidence in diagnoses.

Data from the 100,000 Genomes Project and subsequent evaluations showed the Congenica platform helped the NHS to increase their diagnostic yield by 50%, while improving analysis times 20-fold.

With Congenica at its core, the NHS Genomic Medicine Service now has the potential to transform lives by enabling a quicker diagnosis for patients with rare diseases and bring an end to their 'diagnostic odysseys'.

Congenica: the exclusive Clinical Decision Support partner for the NHS Genomic Medicine Service

Increase diagnostic yield by **50%**

Improve analysis times **20-fold**

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The Challenge

Genomics England was established to enable the NHS to harness the power of genomic technology and science, in order to improve the health of the UK population.

Genomics England's first project was the 100,000 Genomes Project; a world-leading government initiative launched in 2012 to sequence the genomes of 100,000 individuals with a rare disease or cancer. The project aimed to transform patient care and quality of life by bringing advanced diagnosis and personalized treatment to those in need, with the resulting genomic data providing an invaluable resource to further our understanding of the biology of human disease.

This was an unprecedented number of whole genomes to sequence and analyze and presented a huge challenge for Genomics England.

Genomics england

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Embarking on a World First

Integrating genomic testing into routine diagnostic practice at a national level had never been done before and required extensive planning, collaboration and expertise. In order to deliver this ambitious project and maximize benefits to patients, Genomics England faced a number of significant challenges, including:

- Developing national protocols and infrastructure to enable enrolment and consent of patients, and collection, storage and transport of patient samples
- Achieving high throughput whole genome sequencing (WGS)
- Providing rapid, reliable and high-volume data analysis, clinical interpretation and reporting

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By 2026 interpretation of data will account for **56%** of the cost of the clinical use of next generation sequencing

According to market research published by BIS Research, by 2026 interpretation of data will account for 56% of the cost of the clinical use of next generation sequencing and will become a significant bottleneck in its clinical utility without the use of advanced software solutions[1]. It was therefore essential for Genomics England to be able to provide efficient, fast and quality interpretation of genomic data to ensure the project's success, and to reduce future costs to the NHS.

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1. Global NGS Informatics and Clinical Genomics Market - Analysis and Forecast, 2018-2028 BIS

Selection of a Clinical Decision Support Platform

Genomics England selected Congenica as the exclusive clinical decision support solution for the UK NHS Genomic Medicine Service following an extensive and competitive tender process involving the leading global providers of genomic data analysis solutions.

In the initial stages of the tender process, 16 clinical decision support solutions were challenged to identify the causal variants in patients affected by rare diseases. Of the 16 reports Genomics England received, only four companies were able to solve all of the cases – with Congenica leading the field.

The final stage of the evaluation process involved the testing of each solution against a robust set of criteria that included scalability, usability, clinical accuracy, case throughput and commercial value.

The final stage of the evaluation process involved the testing of each solution against a robust set of criteria:

- Scalability
- Usability
- Clinical accuracy
- Case throughput
- Commercial value

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Congenica: the exclusive Clinical Decision Support partner for the NHS Genomic Medicine Service

Best in Class

An independent panel of experts evaluated the various options and rated the Congenica platform the highest overall across these criteria leading to the Congenica platform being selected as the exclusive clinical decision support solution for the NHS Genomic Medicine Service.

In order to maximize the utility of the platform, Congenica's technical and clinical teams worked collaboratively with Genomics England and the Genomic Laboratory Hub network. This allowed all stakeholders to fully understand the specific technical and functional requirements needed to deliver the NHS Genomic Medicine Service and become seamlessly integrated into the UK's national health system.

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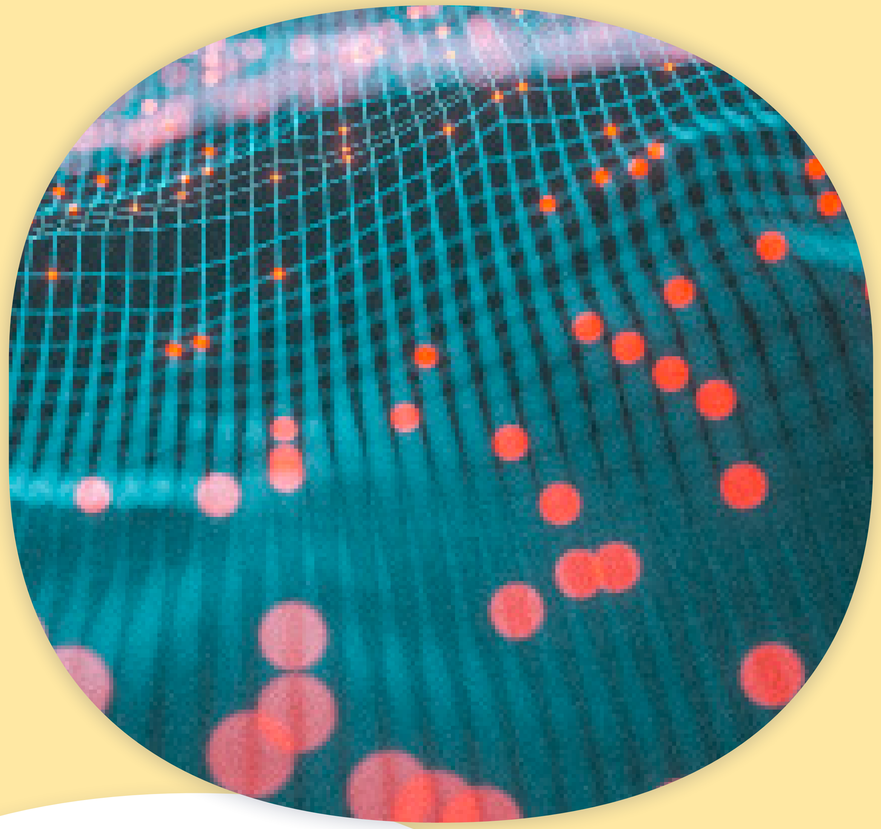
The Outcome

Genomics England has embedded Congenica at the core of the Genomic Medicine Service. Each of the Genomic Laboratory Hubs will use Congenica for clinical interpretation and reporting, reducing infrastructure overheads and simplifying clinical workflows.

The platform also enables clinicians and Clinical Scientists to dive deeper into variant analysis and significantly increase their case throughput at a national level.

The Congenica platform has empowered Genomics England to be able to process an unprecedented 2,700 whole genomes per week. These data are seamlessly and immediately delivered to the Genomic Laboratory Hubs for rapid variant interpretation to support clinical reporting, all within one solution.



Genomics England can now process an unprecedented

2,700 genomes per week

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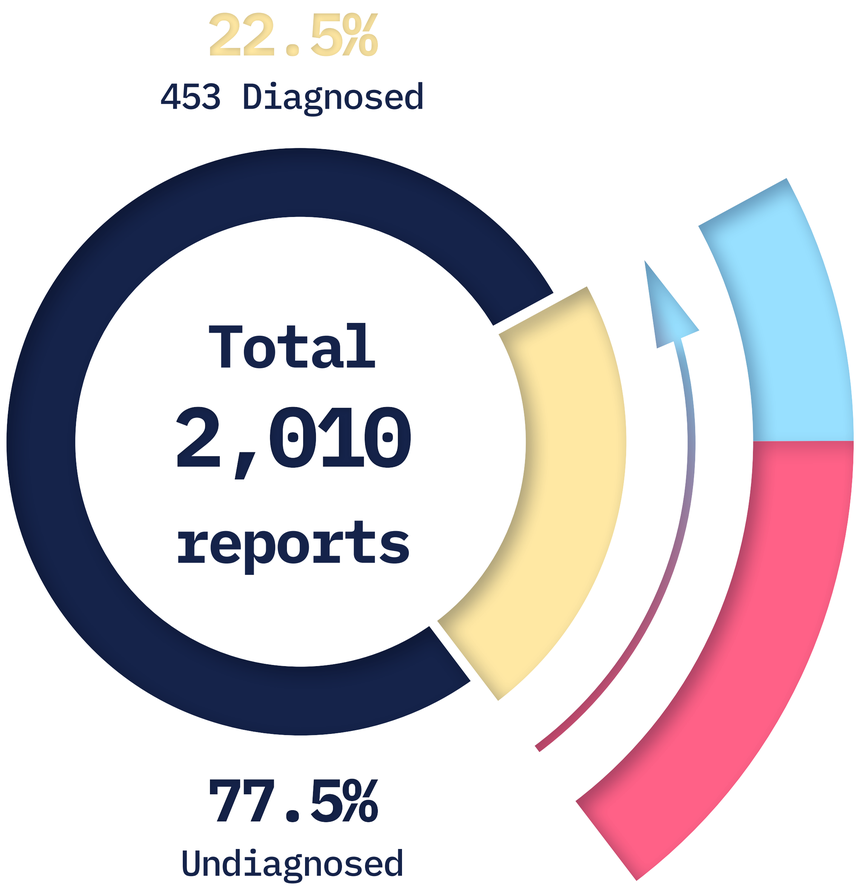
Increase Diagnostic Yield by >50%

In a pilot study during the 100,000 Genomes Project, Congenica identified additional variants that had previously been undetected, helping to increase the diagnostic yield of patients by 50%. In total, one third of patient diagnoses that were identified by the Congenica platform involved variants outside of Genomics England's initial selection of candidate variants.

The Genomic Laboratory Hubs in the UK are required to report patient results within six weeks to meet contracted turnaround times - using Congenica's scalable processing pipeline enables them to process 200+ samples every 24-hours.



35%
Additional variants identified by Congenica
135 Unique genes

65%
Variants identified in Genomics England Bioinformatic Pipeline
225 Unique genes

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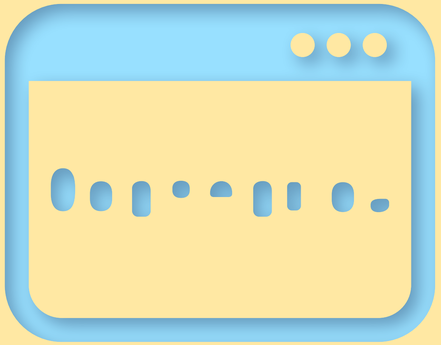
Furthermore, analysis of more than 2,000 complex cases from the 100,000 Genomes Project pilot study shows the use of the Congenica platform enables analyses to be conducted 20X faster while reducing the amount of manual processing time by 95%.

Secure data sharing functionality within the Congenica platform enables the Genomic Laboratory Hubs to share variant curations and previously siloed local knowledge bases across hospital networks. The knowledge-sharing capability also allows non-expert centers to benefit from the work of specialist labs and facilitate faster diagnoses to their patients with more confidence.

Congenica and Genomics England continue to work together to ensure the evolving needs of healthcare professionals in the NHS Genomic Medicine Service are met. This close collaboration and commitment to delivering excellence is leading to shorter time to diagnosis, greater lab efficiency and improved patient outcomes across the UK.

See our whitepaper here for more details

Reduce manual data processing by **95%**



Reduce analysis time by **20X**

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Congenica supports Genomics England to processes thousands of whole genome samples every month. Working with Congenica we're able to provide high quality variant interpretation to the NHS, helping deliver benefits to patients at scale.

Dr Augusto Rendon

Director of Bioinformatics, Genomics England

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Summary

Genomics England has been successful in delivering the 100,000 Genomes Project and establishing the world's first national genomic medicine program with Congenica at the core of both.

In partnership with Congenica, Genomics England has been able to achieve:

- 95% reduction in manual data processing
- 20-fold reduction in analysis times
- 50% increase in diagnostic yield
- Scale of 2,700 whole genomes processed per week

These results have led to an ongoing partnership and Genomics England selecting Congenica as the exclusive clinical decision support platform for the interpretation of all rare disease cases in the UK's groundbreaking NHS Genomic Medicine Service.

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NGS lab efficiency whitepaper

Click here to learn how to “Analyze, interpret and report NGS data faster than ever before”.

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congenica.com

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Enabling genomic medicine