**Cite as:** Nick Lench, Congenica: using genomic data to provide actionable insight
Innovation & Impact, 2021
**URL:** iai.digital/2021/Congenica

# Congenica: using genomic data to provide actionable insight

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Abstract: Congenica is a digital health company enabling the rapid analysis and interpretation of genomic data, empowering researchers to provide life-changing answers that improve wellbeing and disease management. Congenica's world-leading software enables rapid genomic data analysis at scale, performing 20x faster than industry averages and providing a 30% higher analytical yield, reducing genomic interpretation costs by up to 95%. Born out of pioneering research from the Wellcome Sanger Institute and the NHS, Congenica has a global footprint supporting leading international laboratories, academic medical centres and biopharmaceutical companies and is the exclusive Clinical Decision Support partner for the NHS Genomic Medicine Service.

Keywords: Congenica, genomics, data, Wellcome Sanger, Cambridge, NHS

## 1. The success story

Congenica is a pioneer in the analysis of complex genetic and genomic data [1]. Its clinical decision support software enables the rapid analysis and interpretation of DNA sequence data, empowering users currently to provide both life-changing answers for patients with rare genetic diseases and inherited cancer and clinical interventions that can improve wellbeing and disease management. Based in Cambridge, UK, Congenica was born out of ground-breaking clinical genomics research from the Wellcome Sanger Institute and the NHS. Congenica's world-leading software enables rapid genomic data analysis at scale, bringing together DNA sequence data and information from clinical practice. It is this unique combination and the deep understanding of how the clinical scientist's mind works that has made Congenica and its platform such a success.

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A recognised global leader in the genomic analysis of rare diseases and inherited cancer, Congenica has established a diverse, global customer-base of national programmes, hospitals, diagnostic laboratories, academic medical centres and pharmaceutical companies. The exclusive Clinical Decision Support partner for the UK NHS Genomic Medicine Service, Congenica's success has expanded internationally with a customer network spanning over 20 countries.

At the centre of Congenica's story are patients. Congenica's technology is able to provide a definitive molecular diagnosis more accurately and in a much shorter time than was previously possible, reducing the "diagnostic odyssey" from many years to as little as a few days. For clinicians, patients, and their families this is invaluable; it means that the clinician can provide the most effective management of a patient's condition. As medicine continues to rapidly evolve, the timely and accurate diagnosis of genetic conditions can ensure that patients can access potentially life-saving new therapies as they become available.

## 2. How did we start

In 2012 Nick Lench was working at Great Ormond Street Hospital (GOSH) for Children in London where, as the Director of Genetics Services, he led a regional genetics service providing DNA diagnostic testing to a population of over 4.5 million in the north Thames region. The world of diagnostics had undergone a paradigm shift in the preceding years with the invention of next-generation sequencing by Solexa (acquired by Illumina in 2007), a breakthrough which has subsequently enabled the sequencing of an entire human genome in less than 24 hours. Working alongside Nick, Philip Beales, a consultant clinical geneticist at GOSH, was conducting whole exome sequencing on his cohort of rare disease patients. Together they realised that the way forward was to be in a position to offer whole exome sequencing to every child referred to GOSH who had presented with a suspected inherited or acquired genetic disease. It became clear to Nick and Phil that there were two barriers to achieving this: the sequencing capacity to achieve this at scale and the ability to analyse the data, reviewing thousands of genes and variants and performing bioinformatics to achieve relevant findings.

At the same time, the Wellcome Trust Sanger Institute in Cambridge was running two large scale cohort-based exome sequencing projects, the UK10K Project and the Deciphering Developmental Disorders (DDD) Project, led by Congenica co-founders Richard Durbin and Matthew Hurles respectively [2]. These two pioneering projects paved the way for the 100,000 Genomes Project (UK100K). The DDD study was the first nationwide genomics project involving all of the regional UK NHS genetics services, recruiting approximately 14,000 parent/offspring trios with severe undiagnosed developmental disorders. As part of this, whole exome trio sequencing was performed to investigate the genetic causes of abnormal development.

It was against this backdrop that the idea of founding a commercial entity to deliver genomic analysis at scale was born. The catalyst for this was Tom Weaver, a genomics entrepreneur who was founding CEO of Geneservice, a genomics services company (now Source Bioscience). Tom believed that the time was right to combine clinical and diagnostics expertise with cutting edge

bioinformatics and data analytics led by a team of international key opinion leaders. In October 2012 Congenica was formed.

At the same time, the UK prime minister, David Cameron, announced the launch of the UK100KGP creating Genomics England to deliver the sequencing of 100,000 genomes from NHS patients with rare genetic disease or cancer [3]. At the time no other country in the world was attempting such an ambitious undertaking and its launch was a key milestone in the UK becoming a global leader in genomic medicine.

## 3. Our technology

Congenica develops and provides technology and data that take a "clinical first" approach to making the diagnosis of rare disease and other genetic disorders as quick and as easy as possible. The technology is centred on providing the highest possible diagnostic yield, increasing the proportion of patients for whom a diagnosis is achieved. By this method the technology achieves a definitive molecular diagnosis and significantly shortens the time to diagnosis.

The Congenica platform uses a complete diagnostic pipeline and comprehensive database, alongside its user interface and reporting engine. Through this integration Congenica can provide clinicians with everything they need to make a molecular diagnosis and, by making this process automated, the technology removes much of the need for human intervention. Congenica also incorporates artificial intelligence (AI) to further accelerate accurate diagnosis and remove the need for specialist intervention in the process. To support the development of this AI Congenica is building a unique data set, which also serves to improve the quality of the features and capabilities of the software platform. The platform is fully cloud based but also retains appropriate security features with the ability to hold data within different sovereign territories.

Innovation is key to successful technology development; Congenica has a team dedicated to ideation in translational research – moving something from a basic idea and translating it to clinical practice. Innovation takes place across levels of the business, from systems architecture to bioinformatics and finding new ways for the clinical interpretation of DNA sequence variants.

Congenica has designed its own proprietary exome capture assay to improve the efficiency of next generation DNA sequencing of genes associated with inherited and *de novo* genetic disorders. A key advantage of this design is the ability to call copy number and structural variations along with standard calling of single nucleotide variants. Congenica is also developing new assays for the non-invasive prenatal diagnosis of single gene disorders.

One of the things that sets Congenica apart from its competitors is the ability to scale whole genome analysis, interpretation, and reporting. In fact, Congenica is one of the few organisations in the world that can operate on genome scale data as opposed to exome scale and the platform can handle hundreds to thousands of genomes per week. This is something that has been really challenging to solve but is a key achievement for the company. As more national genome programmes are initiated along with the integration of whole genome

sequencing into healthcare systems, the ability to provide a scalable and stable platform becomes increasingly important.

## 4. The journey so far

Congenica has grown rapidly from its beginnings as a start-up and is now supported by a diverse, global investor base. It is a real success story for both the UK genomics sector and the Sanger Institute and highlights the power of the Cambridge technology ecosystem.

Nick Lench is now Congenica's Chief Scientific Officer, having left GOSH in 2014 to move into the company full-time. Since then, the Congenica management team and Board has moved from strength to strength, including the appointment of David Atkins as CEO in 2018. David has been instrumental in the ongoing global expansion of the business and its platform. On the bioinformatics side the addition of Rob Denison as Chief Information Officer has brought significant experience of growing and developing disruptive technology. Congenica's founders have maintained their academic and clinical posts, ensuring the company remains at the forefront of academia and clinical innovation.

Throughout the years Congenica has continued to punch above its weight and its position as the clinical decision support service partner for Genomics England and the NHS England Genomics Medicine Service is testament to this. Congenica's long standing partnership with Genomics England began in 2012, when the company was successful in the initial Genomics England Bake Off that invited 28 companies to analyse, interpret, diagnose and report on whole genome sequences from a cohort of rare disease patients. Further rigorous technical and clinical evaluation led to Congenica providing over 2000 patient reports as part of the UK100KGP pilot study and in 2018 after further competitive tendering, Congenica was appointed exclusive provider of clinical decision support services to the NHS England GMS [4].

A key activity for the company is engagement with patients and patient advocacy groups and this has resulted in the formation of a Patient Advisory and Engagement Board. The Board is comprised of international opinion leaders and influencers representing rare diseases groups, charities, and individuals personally affected by genetic disorders. The link between commerce and patient advocacy groups is becoming increasingly important to drive research and development in the discovery of new medicines and the repurposing of existing medicines. Through their own experiences, patients are best placed to provide valuable information on self-reported outcomes that describe the natural history of their disease and can therefore inform the choice of clinical endpoints used in clinical trial design.

Congenica is funded to drive future success and continue to accelerate its global expansion. The company successfully raised £40 million ($50 million) in November 2020 which will drive further commercialisation of its rare disease, oncology and personalised health platforms. Congenica is now present in over 20 countries with a significant focus on commercial growth and an increasing number of partnerships in strategically important territories.

## 5. Looking to the future

Congenica's overall goal is to deliver a future where clinical genomics is fully integrated into healthcare, becoming a routine component of clinical care. We envisage full automation of the processes involved and eventually even automated diagnosis.

Our ambition is to internationalise further, developing our global customer base and making our platform the gold standard software throughout the world. Building upon our success in rare diseases and inherited cancer we will move further into somatic cancer and into new indications such as screening and precision health, enabling people to live longer and healthier lives. We also plan to work with pharma partners to uncover new research and findings in genetic disease treatment.

The "omics" revolution continues at pace and we are very excited about the potential to revolutionise medicine. We see a future where individuals can have their genome sequenced at birth and are then monitored at different points in their lifetime, looking for risk factors for various conditions and enabling the best possible care. With this, the question is not "if" but "when" and our view is that this will happen soon.

### Acknowledgments

I would like to thank all of the co-founders, Tom Weaver, Richard Durbin, Phil Beales and Matt Hurles and especially Tom for his endless energy and optimism and Andy Richards for his enthusiastic encouragement.

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## The company

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**Nick Lench**, prior to founding Congenica, was previously Director of Genetics Services at Great Ormond Street Hospital for Children, London, with responsibility for the strategic and operational management of a genetics service that provides DNA diagnostic testing and services to a population of approximately 4.5M people. Nick is an honorary Reader at the UCL Institute of Child Health and has over 25 years' of academic, healthcare and commercial experience in medical genetics and genomics. He was awarded a personal chair in Medical Genetics at Cardiff University in 2005 and was a founding CEO of London Genetics Ltd and Programme Director at Oxagen Ltd.