Congenica®

A SeqOne Company

Service Definition Document: Congenica Rare Disease

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Summary

The purpose of this document is to provide documentary evidence to ensure customer confidence in the Congenica Clinical Decision Platform and Clinical Interpretation Services.

Scope

A high-level overview of service, compliance conformity and certification, demonstrating Congenica's continuous growth and development in the genomics field.

Congenica Rare Disease - Service Description

Congenica RD software is a CE Marked In Vitro Diagnostic (IVDR) stand-alone clinical decision support platform, for clinical diagnostic use in inherited genetic disorders for the analysis and interpretation of patient genomic data. The software requires sequence data (in the form of FASTQ, BAM, VCF genetic data files), to enable the analysis of and reporting on variants considered to be causal in relation to the subject's genetic disease. Identification of causal variants aids diagnosis, in relation to genetic disease using public, commercial and customer internal data sources. The platform provides features for recording variant assessments and the drafting of variant analysis reports and is used for clinical decision support by trained Medical Staff, Clinical Scientists, and Genetic Scientists.

Key Features & Benefits

Congenica Rare Disease key features and benefits are summarised as follows:

Features:

1. CE Marked (IVDR), ISO13485, ISO27001, DCB0129, GDPR certified
2. Includes Secondary and Tertiary Analysis, Interpretation and reporting
3. Auditable 2-check workflow designed to follow clinical best practice
4. Includes AI and Machine Learning or simpler, faster analysis
5. Highly flexible to align with customer working practices
6. Can be fully automated from data to report
7. Over 40 Reference datasets to facilitate analysis
8. Fully integrated via API for data ingestion and export
9. Scalable to process hundreds of Whole Genomes per day
10. Browser-based, no local software needed

Benefits

1. Compliance (CE/IVDR): Highest standards of clinical quality and safety.
2. End-to-End Analysis: Streamline your entire diagnostic workflow in one place.
3. Auditable Workflow: Maintain clinical best practices with verifiable, multi-step checks.
4. AI & Machine Learning: Accelerate analysis with intelligent, automated analysis.
5. High Flexibility: Align with your organisational Standard Operating Procedures.
6. Full Automation: Reduce manual effort from hours to minutes.
7. Reference Datasets: Enhance diagnostic accuracy using wide-ranging datasets for analysis.
8. API Integration: Connect your existing tools for seamless data sharing.
9. Scalable Processing: Handle massive genomic workloads without compromising on speed.
10. Security & Encryption: Protect sensitive patient data.

Customer Support

Congenica's Customer Support Team is comprised of technical and scientific experts fully versed in the workings of the Congenica platform and associated APIs and workflows. As part of the onboarding process, a dedicated representative works with each customer to set up appropriate workflows based on Best Practice Guidelines and Good Scientific Practice and to provide training on usage of the platform for variant interpretation and reporting.

Optional Professional Services are available to address custom needs. Such services fall outside the scope of the certifications discussed in this document.

Technical Overview

The following information may be useful for the assessment of the Congenica Rare Disease application:

Customer Onboarding:

Customers are consulted throughout the evaluation and onboarding processes to understand their specific needs and ensure a smooth transition into live operation.
A dedicated Field Application Scientist (FAS) will go through their data, workflow, reporting, integration, user permissions and user interface requirements to ensure a clear understanding.
All aspects of the service are all set up by the FAS and Customer Support in continuous consultation with the customer, who will sign off that the implementation aligns with their expectations.
Customers are given full product training, which can be carried out online or on-site as required.
Full user documentation is provided, which includes a technical system overview and user documentation.

Following completion of the onboarding process, the customer then receives a three-month ‘hyper care’ period where they have continued access to their FAS, who also runs weekly clinics for the customer to address any issues, uncertainties or concerns.
At the end of the hyper care period, the customer is transitioned to a ‘business as usual’ state where ongoing support is provided by the Congenica Customer Support team.
Customers can continue to request a FAS support call and any time if required.

Data import/loading and export:
Data is loaded and exported using the user interface, an SfTP site or using a command line client, depending in the data type and the data in question.
A fully documented, version API protocol is also available for integrated data import and export.
Formats and methods will be agreed with the customer in accordance with the data type and requirements as part of the onboarding process.

Access requirements
The platform requires only a web browser for access can be controlled by Multi-Factor Authentication (MFA) or User Name and Password, depending on customer preference.

Data security
In addition to access controls, data is encrypted in-transit using TLS 1.2 level encryption and at rest using AES-256 encryption.
Congenica is ISO27001:2022, GDPR and Cyber Essentials certified.

Audit
Full Audit Trails are available for all cases within the user interface, which can be accessed and downloaded on demand.
System Audit Logs are fully captured and downloads can be provided automatically via API or on demand through Customer support.

Regulatory Compliance

Congenica prides itself in upholding high standards, with a strong focus on ISO International Standards, nationally recognised Best Practice Guidance, Professional Body membership, Continued Professional Development, with up-to-date compliance and certifications in regulatory standards. In doing so we maintain the provision of high-quality services across all teams within the company.

Quality Management System (QMS)

The QMS is continuously maintained and improved to ensure its effectiveness and its compliance with the requirements of the company Quality Manual and the relevant international standards. Congenica is compliant with EN ISO 13485:2016 - certification can be viewed in appendix 1.

Congenica's Senior Quality Assurance and Regulatory Affairs management staff ensure appropriate Risk assessment and Hazard identification, Incident management, Quality Assurance and Clinical Safety, conforms to recommended standards and polices according to relevant UK, EU and other international legislation. The next sections highlight key elements of Congenica's software and service compliance.

CE-Marked Medical Device

In 2022, EU Regulation 2017/746 on in vitro diagnostic medical devices (IVDR) introduced a major update to the regulatory framework for *in vitro* Diagnostic Medical Devices (IVD's), replacing the IVDD (IVD Directive). Under the new IVDR, all labs performing Laboratory Developed Tests (LDTs) in the European Union must use a CE Marked IVD device or justify why they are not doing so for regulators.

Congenica software for the interpretation of NGS data is a CE Marked In Vitro Diagnostic (IVD) platform for clinical diagnostic use in inherited genetic disorders. Using Congenica allows full integration with existing medical records and laboratory management systems, with confident diagnosis of rare diseases and inherited genetic disorders as part of routine medical care.

Regulatory compliance is essential for patient safety and the effective application of genomic medicine for clinical use. Congenica is certified to the EU IVDR, and therefore can be placed on both the GB and Northern Ireland markets under the transitionary provisions introduced into the UK Medical Device Regulation (UK MDR). The Declaration of Conformity can be viewed in Appendix 1.

The software IVD continues to conform with the requirements of the European IVD Directive 98/79/EC, as required by the UK MDR, and therefore can be UK CA marked if required.

Clinical Risk Management System

The Clinical Safety Officer has devised a Clinical Safety Case Report to provide assurance that the Congenica Rare Disease (RD) Platform is compliant with Clinical Safety Standard DCB 0129: Clinical Risk Management. A Clinical Risk Management Plan (CRMP) describing how Congenica Ltd. conducts clinical risk management to ensure patient safety with respect to services provided and the interrelated and interactive activities that will occur also attests that the Congenica RD Platform meets the requirements of specific Safety standards set out below:

- DCB0129: Clinical Risk Management: its Application in the Manufacture of Health IT Systems

ISO Standards and Conformity

Conformity to International Standards ensures Congenica adheres to rules and guidelines required to achieve the optimum performance in testing strategies, codes of practice, guideline standards and management systems standards.

The following standards on their latest edition were applied in full or in part as found appropriate to the device. Relevant certifications can be viewed in Appendix I.

Standard Title and Relevant Sections:

- **EN ISO 13485:2016+A11:2021** Medical devices - Quality management system - Requirements for regulatory purposes
- **EN ISO 14971:2019+A11:2021** Medical devices - Application of risk management to medical devices

- **EN ISO 15233-1:2021** Medical devices - Symbols to be used with information to be supplied by the manufacturer.
- **EN ISO 18113-3:2022** In vitro diagnostic medical devices - Information to be supplied by the manufacturer (labelling)
- **EN ISO/IEC 27001**:2022 Information security, cybersecurity and privacy protection - Information security management systems - Requirements
- **BS EN ISO 62304:2006 + A1:2015** Medical devices software - Software lifecycle processes

GDPR Compliance and Data Security

Congenica is compliant with the UK General Data Protection Regulation regulation (GDPR) and the Data Protection Act 2018 and certified to ISO 27001:2022. As such Congenica has appropriate and documented technical and organisational measures in accordance with Article 32 of the GDPR.

Congenica has measures in place to process patient data, company employee data and customer data. This includes use of contracts outlining what data we hold, its use and where it will be stored and the duration.

All relevant data received by Congenica from a controller is in a pseudonymised state containing only reference identifiers that can only be linked to actual patient records by the uploading party. Congenica does not receive or store any identifying information directly.

Congenica has an appointed Data Protection Officer and is registered with the Information Commissioners Office (ICO). Security techniques and information security management systems ensure Congenica has mature data and information security management processes. Further compliance assessments have been made to ensure all other GDPR requirements are satisfied, including Data Audit and Data Flow Mapping; GDPR Staff Training; Information Commissioner’s Office.

Third Party Providers and Partners

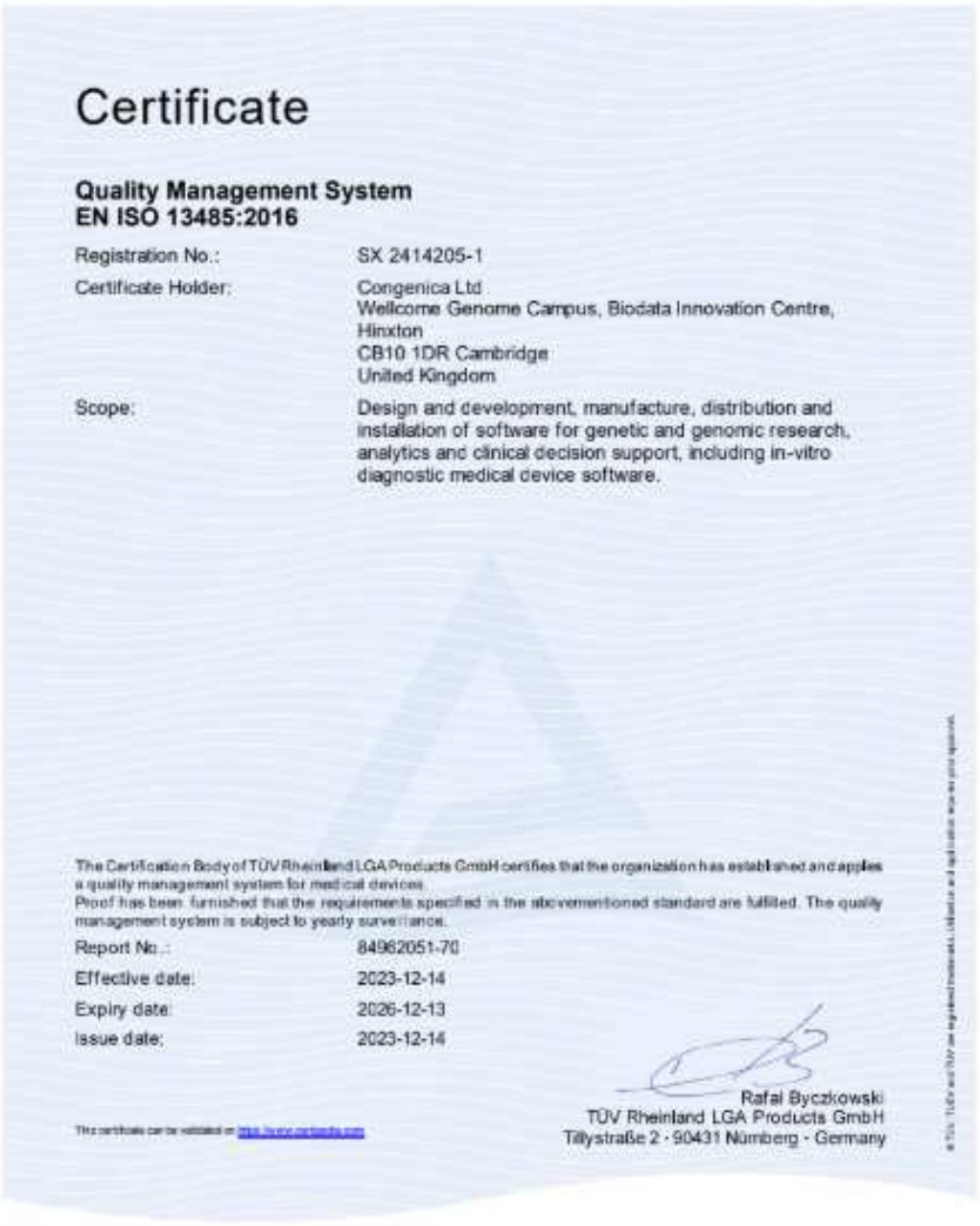
Congenica ensures high quality provision of third party services through detailed evaluations along with application and conformity of ISO standards. Assessing potential risk to product quality, patients and end users, data/information security, and any risk or impact to our Quality Management Systems is essential.

Detailed supplier questionnaires are required from potential partners should risk be calculated as medium or high. Regulatory Certifications and documents of conformity are an essential part of the approval process to ensure appropriate accreditation levels or equivalence where appropriate e.g., ISO15189/CAP/CLIA etc.

Utilising Best Practice validation and verification guidance for sequencing providers ensures accuracy of quality control processes, sensitivity, specificity, and DNA sequencing coverage parameters. Congenica currently have approved UK, European, US and SE Asian affiliates which conform to the highest quality standards.

Clear Non-Disclosure Agreements (NDAs), Statements of Work (SoW) and Service Level Agreements are drawn up to ensure clarity in any work to be undertaken. Independent Quality Management Team approval is also mandatory, with Supplier review processes performed annually as required as part of routine practice to ensure continuous confidence in our providers.

Appendix I - Certifications



Certificate

Quality Management System
EN ISO 13485:2016

Registration No.:	SX 2414205-1
Certificate Holder:	Congenica Ltd Wellcome Genome Campus, Biodata Innovation Centre, Hinxton CB10 1DR Cambridge United Kingdom
Scope:	Design and development, manufacture, distribution and installation of software for genetic and genomic research, analytics and clinical decision support, including in-vitro diagnostic medical device software.

The Certification Body of TÜV Rheinland LGA Products GmbH certifies that the organization has established and applies a quality management system for medical devices.
Proof has been furnished that the requirements specified in the above-mentioned standard are fulfilled. The quality management system is subject to yearly surveillance.

Report No.:	84962051-70
Effective date:	2023-12-14
Expiry date:	2026-12-13
Issue date:	2023-12-14

This certificate can be validated at [illegible]

Rafal Byczkowski
TÜV Rheinland LGA Products GmbH
Tillystraße 2 · 90431 Nürnberg · Germany

DAkkS
TÜVRheinland®
Precisely Right.

British Assessment Bureau
Part of the Amtivo group

CERTIFICATE OF REGISTRATION

The management system of certificate number **205723**

Congenica Ltd

Wellcome Genome Campus, Biodata Innovation Centre, Hinxton, Cambridge, CB10 1DR, United Kingdom

has been assessed and certified as meeting the requirements of:

ISO/IEC 27001:2022

Provision of genetic and genomic clinical decision support and research products and services, Research into inherited and acquired genetic disease, The development and licensing of software for genetic and genomic research, analytics, and clinical decision support, A knowledgebase containing genetic, genomic, and clinical information, A software platform using databases and computer systems and Identification of external risks including functions that are outsourced with client worldwide.

This is in accordance with the Statement of Applicability **version 2.0, last amended on 29 May 2024.**

Further clarifications regarding the scope of this certificate and the applicability of requirements may be obtained by consulting the certifier.

UKAS MANAGEMENT SYSTEMS
8289

IAF

Valid from:
Initial certification: 15 June 2015
Latest issue: 22 July 2025
Expiry date: 14 June 2027
Subject to annual assessments.

Authorised by

Mike Tims
Chief Executive Officer

british-assessment.co.uk

Certificate issued by Amtivo Group Limited T/A British Assessment Bureau Ltd.
Certification is conditional on maintaining the required performance standards throughout the certified period of registration.
Amtivo Group Limited, 30 Tower View, Kings Hill, Kent, ME19 4UY.

Congenica
Enabling precision medicine

EU Declaration of Conformity
Congenica Rare Disease 101

This declaration of conformity is issued under the sole responsibility of the manufacturer, Congenica Ltd.

Item	Details
Manufacturer	Congenica Ltd, Wellcome Genome Campus, Biodata Innovation Centre Hinxton, Cambridge, CB10 1DR, UK
Manufacturer SRN	GB-MF-000026168
EU Authorised Representative	Acorn Regulatory Consultancy Service Ltd, Knockmorris ER21 R766, Co. Tipperary, Ireland
EU Authorised Representative SRN	IE-AR-000000592
Product and Trade Name	Congenica Rare Disease
Intended Purpose	Congenica Rare Disease enables analysis of genomic data to facilitate the identification of causal variants, to aid diagnosis of patients with a suspected germline genetic disease, worldwide using public, commercial and customer internal data sources. It provides features for recording variant assessments and the drafting of variant analysis reports. The software will be used for clinical decision support by trained medical staff, clinical scientists, and genetic scientists. The software is semi-automatic and semi-quantitative.
Product Code(s) / Catalogue Number(s)	101
Basic UDI-DI	506100780CONGIDS101DX
Device Classification	Class C per EU IVD Regulation 2017/746 Annex VIII, Rule 3(i)
Applicable Common Specifications (CS)	N/A
Notified Body	TÜV Rheinland LGA Products GmbH, Tillystraße 2, 90431 Nürnberg, Germany
Notified Body Number	NB 0197
Conformity Assessment Route	Regulation (EU) 2017/746 Annex IX
CE Certificate No.	84962051-70

Congenica
Enabling precision medicine

EU Declaration of Conformity
Congenica Rare Disease 101

On behalf of Congenica Ltd, I hereby confirm that the device(s) identified above conform(s) with:

Regulation (EU) 2017/746 of the European Parliament and of the Council of 5 April 2017 on *in vitro* diagnostic medical devices and repealing Directive 98/79/EC and Commission Decision 2010/227/EU

Signed on behalf of Congenica Ltd on 19 Dec 2025 at Hinxton CB10 1DR

Name of signatory: Dr Eve Hutchinson

Function of signatory: QA/RA Manager

Congenica
Enabling precision medicine

EU Declaration of Conformity

We

Company Name	Congenica Ltd
Postal Address	Biodata Innovation Centre Wellcome Genome Campus Hinxton Cambridgeshire
Postcode	CB10 1DR
Country	United Kingdom
Telephone Number	01223 499965
Email Address	quality@congenica.com

Declare that this DoC is issued under our sole responsibility for the following product

Product Name	Congenica Rare Disease
Product Type	Software Clinical Decision Support Platform
Software Version	Version 3.1 and higher

The product above is in conformity with the relevant European Union harmonisation legislation

IVD Directive	Directive 98/79/EC of the European Parliament and of the Council of 27 October 1998 on in vitro diagnostic medical devices
Category	Annex III General Class IVD. Congenica software is not for self-testing and does not appear in Annex II List A or List B

The following harmonised standards and technical specification have been applied

Title and date of standard applied
EN ISO13485:2016 Medical devices – Quality management systems – Requirements for regulatory purposes
EN ISO 14971:2012 Medical devices – Application of risk management to medical devices
EN62304:2006/AC:2008 Medical device software – Software life-cycle processes
EN ISO 15223-1:2016 Medical devices-Symbols to be used with medical device labels, labelling and information to be supplied – Part 1: Terms, definitions and general requirements
EN ISO 18113-1:2011 In vitro diagnostic medical devices – Information supplied by the manufacturer (labelling) – Part 1: Terms, definitions and general requirements
EN ISO 18113-3:2011 In vitro diagnostic medical devices – Information supplied by the manufacturer (labelling) – Part 3: In vitro diagnostic instruments for professional use

Congenica
Enabling precision medicine

EU Declaration of Conformity

EU Authorised Representative

Company Name	Acorn Regulatory Consultancy Services Ltd
Postal Address	Knockmorris Cahir Co. Tipperary
Postcode	E21 R766
Country	Republic of Ireland

Signed on behalf of Congenica Ltd.

Name	Eve Hutchinson
Position	QA/RA Manager
Signature	 Eve Hutchinson (Mar 11, 2025 13:35 GMT)
Date	11 Mar 2025
Issued	Biodata Innovation Centre, Wellcome Genome Campus, Hinxton, Cambridgeshire, CB10 1DR, United Kingdom

EU Certificate

Quality Management System
REGULATION (EU) 2017/746 on In Vitro Diagnostic Medical Devices
Annex IX Chapter I, Section 2 and 3 and Chapter III

Registration No.:	HX 2414205-1
Manufacturer:	Congenica Ltd Wellcome Genome Campus, Biodata Innovation Centre Hinxton, Cambridge CB10 1DR United Kingdom
EUDAMED Single Registration No.:	GB-MF-000026168
Products:	Products of class C: Nucleic Acid Testing Instruments IVR 0401 Devices intended to be used in screening/confirmation of congenital/inherited disorders W02050192 – NUCLEIC ACID TESTING INSTRUMENTS EXCEPT MICRO-ARRAYS - IVD MEDICAL DEVICE SOFTWARE

The Notified Body hereby declares that the requirements of Annex IX, Chapter I, Section 2 and 3 of the REGULATION (EU) 2017/746 have been met for the listed products. The above named manufacturer has established and applies a quality management system, which is subject to periodic surveillance, defined by Annex IX, Chapter I, Section 3 of the aforementioned regulation. The requirements of Annex IX, Chapter III are fulfilled.
If class D devices are covered by this certificate an EU technical documentation assessment certificate according to Chapter II, Section 4 is required before placing them on the market.
If class B, C or D devices for self-testing or near-patient testing are covered by this certificate an EU technical documentation assessment certificate according to Chapter II, Section 5.1 is required before placing them on the market.
If companion diagnostics are covered by this certificate an EU technical documentation assessment certificate according to Chapter II, Section 5.2 is required before placing them on the market.

Report No.:	84962051-70
Effective date:	2025-12-04
Expiry date:	2030-12-03
Issue date:	2025-12-04

This certificate can be validated on https://www.certipedia.com

Rafał Byczkowski
TÜV Rheinland LGA Products GmbH
Tillystraße 2 · 90431 Nürnberg · Germany

TÜV Rheinland LGA Products GmbH is a Notified Body according to REGULATION (EU) 2017/746 concerning medical devices with the identification number 0197.

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EU Certificate

Quality Management System
REGULATION (EU) 2017/746 on In Vitro Diagnostic Medical Devices
Annex IX Chapter I, Section 2 and 3 and Chapter III

Registration No.:	HX 2414205-1
Manufacturer:	Congenica Ltd Wellcome Genome Campus, Biodata Innovation Centre Hinxton, Cambridge CB10 1DR United Kingdom
EUDAMED Single Registration No.:	GB-MF-000026168
Authorized representative(s):	Acorn Regulatory Consultancy Services Ltd Knockmorris, Cahir Co. Tipperary E21 R766 Ireland

Certificate history		
Revision:	Description:	Issue date:
0	Initial certification	2025-12-04

Report No.:	84962051-70
Effective date:	2025-12-04
Expiry date:	2030-12-03
Issue date:	2025-12-04

This certificate can be validated on https://www.certipedia.com

Rafal Byczkowski
TÜV Rheinland LGA Products GmbH
Tillystraße 2 · 90431 Nürnberg · Germany

TÜV Rheinland LGA Products GmbH is a Notified Body according to REGULATION (EU) 2017/746 concerning medical devices with the identification number 0197.

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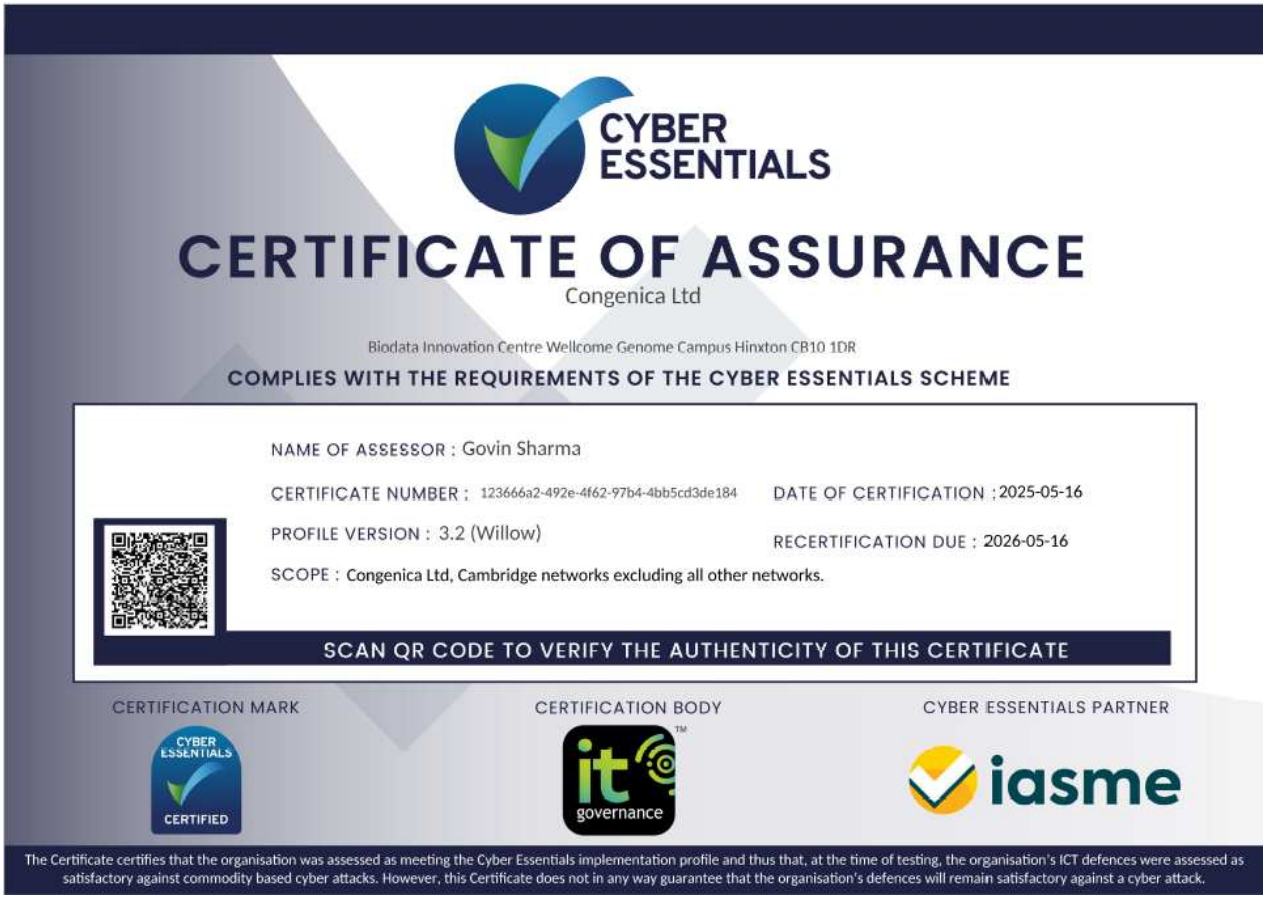

CYBER ESSENTIALS

CERTIFICATE OF ASSURANCE

Congenica Ltd

Biodata Innovation Centre Wellcome Genome Campus Hinxton CB10 1DR

COMPLIES WITH THE REQUIREMENTS OF THE CYBER ESSENTIALS SCHEME

NAME OF ASSESSOR : Govin Sharma

CERTIFICATE NUMBER : 123666a2-492e-4f62-97b4-4bb5cd3de184

DATE OF CERTIFICATION : 2025-05-16

PROFILE VERSION : 3.2 (Willow)

RECERTIFICATION DUE : 2026-05-16

SCOPE : Congenica Ltd, Cambridge networks excluding all other networks.

SCAN QR CODE TO VERIFY THE AUTHENTICITY OF THIS CERTIFICATE

CERTIFICATION MARK

CERTIFICATION BODY

CYBER ESSENTIALS PARTNER

The Certificate certifies that the organisation was assessed as meeting the Cyber Essentials implementation profile and thus that, at the time of testing, the organisation's ICT defences were assessed as satisfactory against commodity based cyber attacks. However, this Certificate does not in any way guarantee that the organisation's defences will remain satisfactory against a cyber attack.

Data Security and Protection Toolkit

NHS Digital

2024-25 (version 7)

CONGENICA LTD

51 Arbury Road, Cambridge, Cambridgeshire, England, CB4 2JB

Standards met

Date of publication: **30 June 2025 (valid to: 30 June 2026)**

This organisation has completed a Data Security and Protection Toolkit self-assessment to demonstrate it is practising good data security and that personal information is handled correctly.

www.dsptoolkit.nhs.uk

Data Protection Registration Certificate

CONGENICA LTD

Bic Wellcome Genome Campus
Hinxton
Cambridge
Cambridgeshire, CB10 1DR

Registration reference: ZA066945
Date registered: 22 July 2014
Registration expires: 21 July 2026

Data Protection Officer

Data Privacy Simplified

31 Rooksmead
Bedford
MK41 7QX

Email: dpo@dataprivacysimplified.co.uk
Telephone: 07384780865

Issued by: Information Commissioner's Office,
Wycliffe House, Water Lane, Wilmslow, Cheshire
SK9 5AF

Telephone: 0303 123 1113
Website: ico.org.uk

Appendix II - Congenica Testimonials

Dr Tessa Homfray
Consultant in Medical Genetics, NHS
“Achieving a rapid prenatal exome result with Congenica allowed us to consider potentially life-saving interventions in pregnancy that would not otherwise have been possible. This result allows us to tailor monitoring of the baby and mother both before and after birth and provide personalised care as appropriate"

Dr Meriel McEntagart
Clinical Geneticist, St George’s University Hospitals NHS Foundation Trust
“Congenica has demonstrated its diagnostic utility even in cases where the low levels of the variant could have led to a missed diagnosis. It provides reassurance to our clinical team that such patients will not be missed in the future.”

David A. Pearce, PhD
President of Innovation, Research and World Clinics, Sanford Health
“This partnership between Congenica and Sanford has the potential to have a transformational impact on clinical outcomes for rare disease patients. We selected Congenica for this project as its platform has the capability of generating rapid and accurate analysis for patients who are currently the hardest to diagnose. The scalability of Congenica also offers scope for widespread integration across our network as we make clinical genomics and personalized medicine more accessible.”

Parent (William’s case study)
"We are not only lucky, but also very grateful to be able to find out about our son’s genetic disorder. Not only does it help with understanding his developmental delays, it has also helped us understand the risks with future pregnancies. It has brought us more opportunities to support our son and his additional needs, as well as getting support for future pregnancies. As a family, we would like to thank you.”

Parent (Arvin’s case)
“It was the rapid exome sequencing that allowed us to discover the variant gene in our son. Having an answer has helped us to access the support that is out there and a huge weight has been take off our shoulders”

Professor Sahar Mansour
Consultant Clinical Geneticist and Physician, St George’s University Hospitals NHS Foundation Trust
“Congenica helped us to provide a diagnosis where other approaches had failed – all within a 20-day window. This is a powerful tool that has helped this family bring 8-years of turmoil to a close”

Dr Anne-Karin Kahlert
Institute of Immunology and Genetics in Kaiserslautern
“Congenica represents an outstanding technology for the interpretation of genetic variants. This helps us to make a genetic diagnosis in a very short time for targeted and therapy-

relevant questions. Especially in prenatal genetic diagnostics as well as in unclear syndromal cases, Congenica supports us in the evaluation in a unique way. Thanks to Congenica, we can now achieve incredibly short turnaround times, which will be of great benefit to our patients."

Gianpiero Cavalleri Ph.D.
Professor of Human Genetics at Royal College of Surgeons Ireland (RCSI), Deputy Director of the SFI FutureNeuro Research Centre and Director of the Human Genetic Variation Research Group
"Using Congenica we significantly increased our diagnostic yield and lab efficiency."

Dr Neil Morgan
Institute of Cardiovascular Sciences, University of Birmingham
"Congenica software is a more robust tool to analyse WES as it provides a higher variant detection rate compared to other bioinformatic tools."

Dr Xue Gao
R&D Scientist, Pangenia
"The expected causal variants are ranked number one by Congenica, which saves a massive amount of time in case interpretation."

Dr Katherine Benson
Postdoctoral Researcher, Royal College of Surgeons Ireland (RCSI)
"By utilising Congenica we increased our diagnostic yield and delivered a molecular diagnosis to patients who would not have received one using our standard bioinformatic pipeline."

Dr Gholson Lyon, M.D., Ph.D.
Principal Investigator and Clinician, Institute for Basic Research in Developmental Disabilities, New York
"Cogenica's clinical team is a unique differentiator. As practicing genomic interpretation scientists they understand the clinical questions we are trying to address and work with us to develop flexible solutions. Their knowledge and experience has been key in making our variant interpretation services a success."

Dr Tessa Homfray
Consultant in Medical Genetics, NHS
"Congenica is an absolute game changer, enabling us to be certain about our diagnoses."

Dr Sahar Mansour
Professor in Clinical Genetics, St George's University Hospitals NHS
"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."

Dr Augusto Rendon
Director of Bioinformatics of Genomics England
"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."

Professor Norman Delanty
Associate Professor at the FutureNeuro Research Centre and a Consultant Neurologist
"In the evolving new era of genomic medicine, Congenica Neuro is an exciting and powerful tool to facilitate the rapid molecular diagnosis of young children with a variety of important neurodevelopment disorders"

Dr Simon Ramsden, BSC MSc PhD FRCPath
Consultant Clinical Scientist, Manchester Centre for Genomic Medicine
"Congenica has allowed us to scale our operation to accommodate the explosion in genomic analysis. It enables us to collate genomic information, do the analysis, and then make decisions within a single piece of software - making our entire service far more efficient.

"In the past a baby with neurological problems would first see a paediatrician, then a neurologist, and if there's ophthalmic problems they'll see the ophthalmologist. Now on day one we can look at a panel of genes that cause cataracts and not worry about all the other biochemical tests that take a long time and cost the NHS a lot of money. It has radically changed the experience for patients.

"A lot of pathologies are progressive, so when they first appear they're not typical. It may be difficult for a clinician to make an accurate diagnosis until it has progressed to the full-blown disease. Whole exome analysis allows us to make that diagnosis much earlier.

"What Congenica does is give us a format: it is a web-based tool where we can take our data, compare it to what's out there in the world, and support the clinicians to come up with a diagnosis.

"We now find ourselves in a multidisciplinary team (MDT) sitting in a room with surgeons, geneticists and scientists. The technology does not presume a particular diagnosis so we look at the genetics and the clinician comes to us with a range of symptoms, then we piece together a diagnosis. Congenica has dramatically changed the way we operate."

Lucy Jenkins
Director North East Thames Genetics Laboratories and joint head of Clinical Service for Genetics
"We are very excited about using Congenica. The platform is very intuitive to use and will speed up diagnosis. It also brings together all the known information about gene-disease associations in one convenient place. Some diseases are so rare a consultant would only see a condition once in his or her career; now consultants will be able to cross-reference

their patients with the knowledge base in Congenica. This ultimately benefits the patient and the family by shortening the time to diagnosis"

Martin Armstrong
Senior Director of Molecular Genetics, UCB Pharma
"Generating sequencing data is not the challenge, making sense of it is. In order to realise our aims we need to be able to analyse and interpret NGS data in the correct biological context and in a manner that allows us to make informed decisions on what might, or might not be, a relevant target or mechanism to pursue.
"The competitive advantage will be achieved by doing this in an efficient and objective manner, integrating supporting data from multiple sources and making use of the latest analytical tools and platforms, both those developed within UCB and those developed by expert companies working in the field.
"This arrangement [with Congenica] will provide access to a cutting-edge genome sequence analysis and decision support environment that will help us effectively resolve the genetic origins of rare disease."

Dr Pieter van Rooyen
Chief Executive Officer, Edico Genome
"As genomics marches towards the clinic, we recognize clinicians and researchers need easy to use, all-in-one solutions that enable genomic data to be analysed and shared quickly, easily, accurately and cost effectively.
"Congenica has first-hand perspective of the needs of the clinical genomics community from its extensive work with the NHS, including the Genomics England initiative, and through this new collaboration we're able to create an all-in-one, easy-to-use offering that significantly accelerates the ability of hospitals and clinical labs to move from the sequencing of a sample to a clinical diagnosis."

Dr Weike Mo
VP, Digital China Health
"Congenica has demonstrated how, with the right investment and leadership, the UK can build world-leading technology companies.

"Our business partnership reflects Congenica's proven abilities to analyse and interpret big data and underlines our confidence that its technology platform will contribute to the new era of genomic medicine internationally."

Appendix III - Logos in Use