Congenica®

A SeqOne Company

Compliance Confidence: Congenica Genomic Analysis Platform

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1. Service Overview

1.1 Service Name

Genomic Analysis Platform

1.2 Service Description

The Genomic Analysis Platform is a cloud-based, CE-IVDR Class C certified bioinformatics Software-as-a-Service (SaaS) solution designed for clinical genomic analysis in healthcare settings. The platform provides end-to-end capabilities for the detection, annotation, interpretation, and reporting of genetic variants from next-generation sequencing (NGS) data, supporting both germline analysis for rare disease and hereditary cancer diagnostics, and somatic analysis for oncology applications including solid tumours, haematological malignancies, and liquid biopsy.

The service accepts raw sequencing data in standard formats (FASTQ, BAM, VCF) from all major NGS instruments including Illumina, MGI, and Element Biosciences platforms. Users can configure custom analysis pipelines or utilise validated, ready-to-use workflows. The platform performs secondary analysis (read alignment, variant calling) and tertiary analysis (variant annotation, filtering, prioritisation, classification, and clinical reporting) within an integrated, browser-based interface.

Key Clinical Applications

Germline Analysis (GermVar): Complete variant analysis for rare diseases and hereditary cancer syndromes, supporting singleton cases, family-based analysis (trios, quads, extended pedigrees), and reanalysis workflows. The platform detects single nucleotide variants (SNVs), multi-nucleotide variants (MNVs), copy number variants (CNVs), short and mid-size insertions/deletions (indels), and Alu element insertions across targeted panels (1–800 genes), clinical exomes, whole exomes (WES), and whole genomes (WGS).

Somatic Analysis (SomaVar, SomaCGP, SomaHemato): Tumour-only analysis for solid tumours and haematological malignancies, with detection of SNVs, indels, copy number alterations (CNAs), gene fusions, microsatellite instability (MSI), tumour mutational burden (TMB), and loss of heterozygosity (LOH). Deep sequencing mode supports detection of low-frequency variants at coverage depths up to 5,000x with unique molecular identifier (UMI) support.

Homologous Recombination Deficiency (SomaHRD): Clinically validated Genomic Instability Score (GiS) calculation from shallow whole-genome sequencing, concordant with PAOLA clinical trial methodology for identifying patients likely to benefit from PARP inhibitor therapy.

RNA Analysis (SomaRNA): Targeted RNA panel analysis for detection of gene fusions (known and novel) and splice variants (e.g. METΔ14), alterations relevant to treatment selection.

Liquid Biopsy (SomaLBx): Highly sensitive circulating tumour DNA (ctDNA) analysis from plasma samples, with variant allele frequency (VAF) detection down to 0.1% and demonstrated 100% sensitivity for SNVs and amplifications at 0.5% VAF.

DNA Methylation (SomaMethyl): Analysis of differential methylation regions (DMRs) with advanced visualisation, cohort comparison capabilities, and CpG island coverage assessment.

DiagAI and Variant Interpretation

The platform incorporates **DiagAI**, our proprietary explainable artificial intelligence engine, which automates variant prioritisation by correlating genomic findings with patient phenotypes (Human Phenotype Ontology terms), inheritance patterns, and the clinical evidence base. DiagAI provides transparency into its prioritisation reasoning, enabling clinical scientists to understand and validate AI-assisted recommendations.

Variant interpretation is supported by comprehensive annotation from curated databases including ClinVar, COSMIC, CKB Boost (actionable oncology knowledge), OMIM, GnomAD population frequencies, SpliceAI

splice predictions, and CADD pathogenicity scores. The platform provides automated classification according to ACMG/AMP guidelines for germline variants and AMP/ASCO/CAP tiering for somatic variants, with full traceability of classification criteria and evidence.

GenomeAlert provides continuous reanalysis capabilities, automatically re-evaluating previously negative cases against monthly knowledge base updates to identify newly classified pathogenic variants, ensuring patients benefit from evolving scientific understanding without manual intervention.

The **Flow** automation tool enables integration with laboratory information management systems (LIMS), and third-party systems through configurable workflows for data import, sample tracking, result export, and notification triggering. Flow workflows are implemented using the platform's secure REST API, enabling event-driven automation, external system orchestration, and reproducible pipeline execution with support for API, file, or email triggers.

The platform is designed for use by NHS Genomic Laboratory Hubs (GLHs), clinical genetics laboratories, molecular pathology departments, and reference laboratories requiring validated, auditable genomic analysis workflows that meet regulatory requirements for clinical diagnostic use.

1.3 Features and Benefits

Features

#	Feature
1	CE-IVDR Class C Medical Device Certification
2	Comprehensive Germline Variant Analysis
3	Advanced Somatic Oncology Analysis
4	Clinically Validated HRD Scoring
5	DiagAI Explainable Artificial Intelligence
6	Premium Annotation and Knowledge Bases
7	Automated Variant Classification
8	Flow Workflow Automation
9	GenomeAlert Continuous Reanalysis
10	Complete Audit Trail and Traceability
11	Sequencer and Wetlab Agnostic Design
12	Customisable Report Templates
13	Virtual Panel Management
14	Long-Read Sequencing Support
15	Multi-Language Interface
16	REST API

Benefits

#	Benefit
1	Reduced Variant Interpretation Time
2	Elimination of Capital Expenditure
3	Certified Secure Health Data Hosting
4	High Availability and Resilience
5	Preservation of Existing Infrastructure Investment
6	UK and EEA Data Residency
7	Continuous Platform Improvement
8	Dedicated NHS and Public Sector Support
9	Proven NHS Deployment Track Record
10	Regulatory Compliance Assurance
11	Improved Diagnostic Yield
12	Standardised Interpretation Workflows

2. Operational and Technical Details

2.1 Onboarding and Offboarding

Onboarding Process

We provide a comprehensive, structured onboarding programme designed to bring laboratories to full operational readiness within 4–6 weeks of contract signature depending on deployment complexity. Simpler deployments may complete in as few as 4 weeks, while complex integrations (LIMS, SSO, extensive validation) typically require longer. The onboarding process is included in the standard subscription at no additional cost.

Phase 1: Project Initiation (Days 1–5)

Upon contract execution, we assign a dedicated Customer Success Manager and initiate the onboarding project with a kick-off call involving key laboratory stakeholders. This session establishes scope of initial deployment, technical requirements and integration points, user account structure and permission requirements, training schedule, method validation requirements, and success criteria with go-live date targets.

Phase 2: Platform Provisioning (Days 2–7)

Technical provisioning activities include dedicated customer environment creation on ISO 27001/HDS certified infrastructure, user account provisioning with role-based access control (RBAC) configuration, data transfer channel establishment (secure web upload, SFTP, S3 bucket integration), single sign-on (SSO) configuration if required (SAML 2.0 compatible), IP allowlisting and network security configuration, and sandbox/training environment provisioning.

Phase 3: Configuration and Customisation (Days 5–14)

Platform configuration is tailored to laboratory-specific requirements including analysis pipeline selection, virtual gene panel creation, variant filter profile configuration, report template customisation, quality control threshold configuration, and Flow automation workflow configuration.

Phase 4: Training (Days 10–21)

Comprehensive training includes Administrator Training (2–4 hours), User Training (4–8 hours), and Bioinformatics Training (2–4 hours). Training materials provided include user guides in PDF format (English, French, Spanish), quick reference cards, video tutorials, and access to online Knowledge Base.

Phase 5: Validation Support (Days 14–35)

We provide method validation support to assist laboratories with accreditation requirements including validation protocol templates aligned with ISO 15189 requirements, test data sets, technical consultation on validation design and acceptance criteria, review and interpretation of validation results, and documentation support for accreditation submissions.

Phase 6: Go-Live and Hypercare (Days 28–42)

Following successful validation and user training: formal go-live approval and production environment activation, hypercare period with enhanced support response times, daily check-in calls during first week of production use, rapid issue resolution and configuration adjustment, post-go-live training reinforcement as needed, and transition to standard support model following stabilisation.

Offboarding Services

Services Included in Contract Price:

- Full platform access until contract end date
- Self-service data export in standard formats throughout notice period
- Guidance on data extraction procedures and format options
- 90-day post-contract grace period for data retrieval (read-only)
- Written confirmation of data deletion upon request

2.2 Data Management

Data Importing

The platform supports flexible data import methods including Web Upload (drag-and-drop with resumable uploads), SFTP Transfer (automated, scheduled transfer with SSH key authentication), S3 Integration (high-throughput transfer with server-side encryption), and API Upload (programmatic file upload via REST API).

Supported Upload Formats:

- FASTQ (raw sequencing reads from Illumina, MGI, Element Biosciences)
- BAM (aligned reads from third-party pipelines)
- VCF (variant calls for tertiary analysis only)
- PED (pedigree files for family analysis)
- BED (target region definitions)
- HPO (Human Phenotype Ontology term files)
- XLS (aCGH data)

Data Exporting

Export Formats: VCF, TSV/CSV, JSON, PDF, XLSX, HTML, BAM, FASTQ

Export Methods: Web Download, SFTP Transfer, S3 Transfer, API Retrieval

All API interactions are authenticated via API keys, logged in the audit trail, and subject to the same role-based access control (RBAC) policies as the web interface. The REST API provides programmatic access to projects, samples, analyses, files, and results, enabling automated data ingestion, workflow orchestration, and

system-to-system integration with LIMS, EHRs, and research platforms. Additionally, the platform provides specialised DiagAI APIs for phenotype-driven analysis, including the Text2HPO API for automated extraction of Human Phenotype Ontology terms from clinical notes, and the PhenoGenius API for diagnostic gene ranking based on phenotype data.

Data Retention Periods

Data Type	Retention Period
Active project data	Retained indefinitely while contract is active
Deleted project data	Soft-delete retention (configurable, default 30 days)
Post-contract data	90-day grace period, then permanent deletion
Audit logs	Retained for minimum 12 months (anonymised after data deletion)
Backup retention	30 days rolling, then overwritten

2.3 Security

Availability

Metric	Value
Contractual SLA	99.0% monthly availability
Target Availability	99.9%
Measured Performance	99.9% consistently achieved
Planned Maintenance Window	Outside business hours (18:00–06:00 UK time)
Maintenance Notification	Minimum 5 working days advance notice
Typical Maintenance Duration	Maximum 4 hours per quarter

Resilience

Infrastructure Resilience: Multi-Availability Zone Deployment with active-active configuration across multiple AWS Availability Zones within the primary region (Paris). Load balancing, auto-scaling, and PostgreSQL databases with Multi-AZ standby replicas providing automatic failover.

Data Resilience: Amazon S3 provides 99.999999999% (eleven nines) durability. Full database backups performed daily with 30-day rolling retention.

Disaster Recovery: Secondary environment in AWS Europe (Ireland) region. RPO: Maximum 24 hours. RTO: Target 4 hours. DR procedures tested quarterly.

Operational Security

Access Control: Role-Based Access Control (RBAC), mandatory Multi-Factor Authentication (MFA), strong password policy, automatic session timeout, and optional IP allowlisting.

Encryption: All data encrypted at rest using AES-256 with AWS KMS. All data transmission protected by HTTPS with TLS 1.2 minimum.

Multi-Tenancy Isolation: Logical separation at application, database, and storage levels. Customer data tagged with tenant identifiers with cross-tenant data access technically prevented by architecture.

Security Certifications

Certification	Status	Scope
ISO/IEC 27001:2022	Certified	Information security management system
HDS (Hébergeur de Données de Santé)	Certified	French health data hosting certification
ISO 13485	Certified	Quality management system for medical devices
Cyber Essentials	Compliant	UK government cybersecurity baseline
CSA STAR	Compliant	Cloud security certification
HIPAA	Compliant	US health data protection
OWASP Top 10 (2021)	Compliant	Web application security
SOC 2 Type II	In progress	Service organisation controls

2.4 User Support

Support Channels

Email and Ticketing System: Primary support channel via support@seqone.com or integrated support portal.

Telephone Support: UK Support Number provided upon contract signature. Hours: 08:00–17:00 UK time, Monday–Friday.

On-Site Support: Available for complex implementation issues, quoted separately at Professional Services rates.

Response Time Commitments

Severity	Initial Response	Target Resolution
Severity 1 (Critical)	4 business hours	2 business days
Severity 2 (High)	1 business day	3 business days
Severity 3 (Medium)	1 business day	5 business days
Severity 4 (Low)	1 business day	5 business days

2.5 Technical Requirements

Browser Interface

Supported Browsers: Google Chrome, Mozilla Firefox, Microsoft Edge (current stable releases).

Browser Requirements: JavaScript enabled, cookies enabled, TLS 1.2 or higher support, minimum screen resolution 1280 × 720 pixels.

Network Requirements: Stable internet connectivity with minimum 10 Mbps bandwidth, HTTPS (port 443) access, WebSocket connectivity for real-time updates.

Compatibility

Sequencing Platform Compatibility: Illumina (NovaSeq, NextSeq, MiSeq, HiSeq, iSeq), MGI (DNBSEQ-G400, DNBSEQ-T7), Element Biosciences (AVITI), PacBio (Revio, Sequel II), Oxford Nanopore

(PromethION, MinION).

Reference Genome Support: GRCh37/hg19, GRCh38/hg38, with lift-over between assemblies supported.

3. Audit and Standards

3.1 Audit Information for Users

The Genomic Analysis Platform maintains audit capabilities to support clinical governance, regulatory compliance, and accreditation requirements. The platform provides full audit trail and traceability for regulatory compliance with tracking of user activities, timestamps, and user identification for actions.

Audit Log Retention

Log Type	Retention Period
System logs	At least 12 months

External Audit Support

- **Right to Audit:** Contract provisions for customer audit rights
- **Audit Questionnaire Response:** Timely response to security and compliance questionnaires
- **Evidence Provision:** Certification documentation and penetration test summaries
- **Audit Facilitation:** Coordination with customer audit teams for information requests

3.2 Energy Efficiency and Sustainability

AWS Infrastructure Sustainability: AWS committed to powering operations with 100% renewable energy. European regions (Paris, Ireland) powered by renewable energy sources. Purpose-built data centres designed for energy efficiency with advanced cooling technologies.

SaaS Model Environmental Benefits: Multi-tenant cloud model enables resource sharing across customers with 65-80% server utilisation (vs. 10-20% for typical on-premises), continuous AWS investment in efficient hardware, and purpose-built data centre cooling.

3.3 Regulatory and Quality Standards

CE-IVDR Class C Certification

The Genomic Analysis Platform is certified as a Class C in vitro diagnostic medical device under the European In Vitro Diagnostic Regulation (EU) 2017/746, with classification as high individual risk, managed by a third-party Notified Body, for the intended purpose of clinical genomic analysis for diagnosis of genetic conditions.

Security Governance

Our security posture is built upon the ISO 27001 framework and is designed to comply with GDPR requirements. The governance structure includes a Chief Information Security Officer (CISO) responsible for leading the overall security strategy and a Security Committee comprising the CISO, CTO, and Compliance that convenes quarterly.

Core Policies: Information Security Policy, Acceptable Usage Policy, Access Control, Data Encryption, Vulnerability Management, Incident Management, Risk Assessment. Policies are reviewed annually and updated for new threats or architectural changes.

Data Protection

Control	Implementation
Encryption at Rest	AES-256
Key Management	AWS KMS with FIPS 140-2 Level 3 validated HSMs
Logical Isolation	Customer-specific encryption keys in dedicated isolated software modules
Data Location	UK, EEA (Primary: AWS Paris; Backups: AWS Ireland)
In-Transit Encryption	TLS 1.2 minimum

Compliance Standards Summary

Standard	Status	Application
EU IVDR 2017/746	Certified (Class C)	Medical device regulation
ISO/IEC 27001:2022	Certified	Information security management
ISO/IEC 27035-1:2023	Compliant	Incident management
GDPR	Compliant	Data protection

Penetration Testing

Aspect	Detail
Frequency	At least once a year
Provider	CREST-accredited external testers
Scope	Platform infrastructure and prior to major releases

Audit and Inspection Readiness

- **Internal Audits:** Regular internal audit programme
- **External Audits:** Annual independent external audits
- **Customer Audits:** Right to audit provisions in contracts; audit facilitation upon request
- **Regulatory Inspections:** Cooperation with competent authority inspections as required

4. Customer Testimonials

The Genomic Analysis Platform is trusted by leading healthcare organisations across Europe. Below are testimonials from customers demonstrating the platform's impact on clinical genomics operations.

North West Genomics Laboratory Hub (UK NHS)

"The big advantage of using SeqOne for us is how transparent the process is: we can get high-resolution information on each sample and understand in detail how the genomic instability score is calculated. This gives us additional confidence when reporting clinical results to the doctors and patients we serve."
— Joe Shaw, Clinical Scientist, North West GLH

Uppsala University Hospital (Sweden)

"With SeqOne, we don't just have a provider; we have a dedicated partner. The team is constantly communicating, rapidly solving issues, and evolving the platform with us. That level of collaboration is invaluable for a lab like ours."
— Hans Matsson, Clinical Laboratory Geneticist, Uppsala University Hospital

Assistance Publique - Hôpitaux de Marseille (France)

"The automation of exome sequencing marks a pivotal moment for our laboratory. With SeqOne's support, we've dramatically increased our sample analysis capacity and precision, significantly improving our ability to diagnose rare genetic diseases."
— Prof. Martin Krahn, Director of the Molecular Genetics Laboratory, AP-HM

Cerba Internacional (Spain)

"The dashboard provides a wealth of information, the AI facilitates accurate variant prioritisation, and genes where compound heterozygosity is suspected are flagged. In essence, SeqOne delivers all the critical information needed for correct variant classification in a user-friendly and straightforward manner."
— Óscar Rodríguez, MSc., Deputy Head of Molecular Biology, Cerba Internacional

Cytogenomic Medical Laboratory (Romania)

"We were wholly enchanted with the genetic interpretation platform – a truly game-changing and powerful tool vital for efficient and accurate analysis. Furthermore, the brilliant and friendly technical support team was one of the most important arguments in favour of continuing our collaboration."
— Andreea Tutulan-Cunita, Geneticist, Cytogenomic Lab