

G-Cloud 15: PhenoTips Service Definition Document

1. Service Overview: Transforming Genomic Health Record Management for the NHS

NHS trusts and public sector healthcare organisations across the UK are facing growing challenges in managing their genomic information systems. Many services rely on outdated tools that limit workflow optimisation, automation, interoperability, and regulatory compliance. In line with NHS Digital Transformation goals, several trusts are evaluating modern alternatives like PhenoTips. A secure, cloud-hosted, browser-accessible Genomic Health Record and clinical genetics platform. PhenoTips addresses key limitations of legacy tools and supports streamlined genetic services across a wide range of clinical and research settings. PhenoTips offers a modern, scalable alternative to legacy systems, enabling end-to-end digitisation of genetics workflows. This includes enhanced automation, and superior data management capabilities. Its pedigree system, phenotyping tools, and risk assessment modules are fully integrated to support accurate, standardised data capture and longitudinal tracking.

PhenoTips is also purpose-built to meet UK data retention requirements. In accordance with guidance from the British Society for Genetic Medicine (BSGM), PhenoTips allows for secure, long-term storage of genetic records for at least 30 years following the last point of contact with the family, ensuring compliance across all use cases. Its clinical features are designed with precision medicine in mind. PhenoTips supports pedigree tracking, genotype recording, Human Phenotype Ontology (HPO) tagging, and OMIM integration. This ensures structured, searchable data that supports faster diagnosis, improved research, and better patient outcomes.

The platform's scalability is evident in large-scale deployments across NHS Genomic Medicine Service Alliances (GMSAs), including examples where multiple Trusts with independent EPRs are securely connected via a centralised PhenoTips deployment. This capability enables efficient, controlled data sharing between genetics services, labs, and referring clinicians. It also ensures continuity of care for patients who may be seen across different institutions within a region.

This digital transformation is driven by several core needs within NHS genetics services:

A. Replacing Legacy Systems

Many legacy tools lack the scalability, automation, and interoperability required to meet modern standards in genetic data management. PhenoTips offers a robust, future-proof solution capable of supporting evolving clinical and operational needs.

B. Workflow Optimisation and Automation

PhenoTips enhances clinical productivity by enabling configurable workflows and integrating AI-powered tools. This reduces manual data entry, minimises errors, and improves accuracy and turnaround times for patient assessments.

C. Seamless EPR Integration

Designed for interoperability, PhenoTips integrates with leading Electronic Patient Record (EPR) systems using open standards such as HL7v2 and FHIR. This ensures centralised and accessible genetic data within existing digital health infrastructures, regardless of EPR vendor.

D. Compliance with Genetic Data Retention Policies

In alignment with guidance from the British Society for Genetic Medicine (BSGM), PhenoTips supports secure long-term data retention ensuring genetic records are preserved for at least 30 years from the last family contact, as required.

E. Advanced Phenotyping and Data Interoperability

PhenoTips includes built-in support for Human Phenotype Ontology (HPO), OMIM coding, and genotype/mutation recording. These structured data features promote interoperability, improve diagnostic workflows, and enable meaningful insights across clinical and research settings.

F. Scalability and Future Readiness

The platform is designed to grow alongside NHS organisations, supporting expansion across departments such as cardiology, oncology, and rare disease services without compromising performance or security.

G. Cross-Trust Data Sharing and Regional Collaboration

PhenoTips enables secure data sharing between Trusts through role-based permissions and regional deployments. For example, its implementation in the East GMSA connects Trusts using different EPRs to a centralised PhenoTips platform. This supports regional referrals, lab coordination, and unified patient care pathways across multiple providers. Patients benefit from uninterrupted access to their genetic records, regardless of where they are seen within the GMSA region.

By addressing the limitations of legacy systems and enabling end-to-end digital transformation, PhenoTips empowers NHS organisations to deliver safer, faster, and more personalised genomic care. Its structured, interoperable, and scalable design not only meets today's clinical genetics needs but also lays the foundation for future innovation in precision medicine. As the NHS continues to advance its digital and genomic strategies, PhenoTips offers a proven, adaptable solution aligned with national objectives for improved outcomes, operational efficiency, and patient-centred care.

2. Core Capabilities & Features

The platform is built around a complete genomic health record that provides the following features:

- a. **Comprehensive Genetic Data Management:** PhenoTips provides a unified platform for pedigree tracking, OMIM coding, mutation/genotype recording, and HPO integration. This comprehensive suite of features aligns perfectly with NHS's needs, allowing for improved data quality that leverages ontologies to provide structured data capture.
- b. **Digitised Workflow Solutions:**
 - a. *The Pre-visit Patient Questionnaire*, allowing health care providers to save time by gathering valuable medical and familial history data in advance of a patient visit. The patient-entered family history is automatically converted into a pedigree in PhenoTips.
 - b. *Digital Pedigrees* to facilitate data retention in a standardized format, enable the linking of clinical and phenotypic data within the pedigree, and provide deeper insights into the patient's familial health history.

- c. *The Cancer Risk Assessment Tool*, integrated into both Pre-Visit Patient Questionnaires as well as PhenoTips' Pedigree Tool which incorporates several cancer risk models (CANRISK, GAIL, PREMM5 and IBIS) into the application to provide a standardised risk assessment based on patient details.
- c. **Seamless Integration:** PhenoTips can seamlessly integrate with any EPR, offering compatibility with HL7v2 and FHIR, ensuring a smooth transition of data between systems. Additionally, PhenoTips provides support for data migration, ensuring that any existing data gets mapped and migrated into the client instance.
- d. **Genetic Data Retention Compliance:** PhenoTips has the capability to support NHS in adhering to the BSMG's genetic data retention guidelines. It can store and manage data for the required 30-year period from the last contact with the family, ensuring compliance with regulatory standards.
- e. **Scalable Licensing Model:** NHS institutions can benefit from PhenoTips' Monthly Active User (MAU) pricing model, allowing flexibility in user expansion as needed across various departments, including cardiology and oncology.

Given the need for a streamlined genetic data management tool, PhenoTips provides the optimal solution. Its seamless integration capabilities, comprehensive genetic data management features, compliance with genetic data retention guidelines, scalable licensing model, and digitised workflow solutions make it the only tool that can address these unique requirements.

3. Data Backup, Restore & Disaster Recovery

PhenoTips operates within a highly resilient public cloud infrastructure using Amazon Web Services (AWS) and Microsoft Azure. Backups of critical systems, are encrypted and stored weekly in durable S3 buckets. In the event of a failure of critical components, restoration is possible within hours using standard recovery protocols. The platform's multi-region, multi-availability zone architecture ensures fault tolerance and service continuity. Internet and communication redundancy is supported by fallback mobile data solutions and remote access capabilities to mitigate local disruptions.

4. Onboarding & Offboarding Support

PhenoTips provides a structured onboarding process that begins with stakeholder engagement and detailed configuration workshops. During implementation, user roles are defined, phenotype libraries and form templates are configured, and integrations with existing systems are established. Clients receive comprehensive training via virtual sessions and sandbox environments, alongside ongoing access to documentation. A dedicated account manager supports the onboarding process, facilitating a smooth transition.

Upon contract conclusion, PhenoTips supports secure offboarding through structured data exports in open formats such as CSV, JSON, and HL7 FHIR. These exports include phenotypic records, pedigrees, audit trails, and configuration metadata. Data export validation and assistance are available under a Statement of Work, and sanitisation of residual data can be completed upon request.

5. Service Constraints & Maintenance

PhenoTips operates with scheduled maintenance periods that are communicated to clients in advance. Non-urgent maintenance is notified at least seven days prior, while urgent changes are communicated with at least 24 hours' notice. Maintenance typically occurs outside of core UK business hours.

Configuration options within PhenoTips allow for user-managed customisations, such as the creation of new clinical forms and phenotype templates. However, more complex modifications, including integration development, workflow redesign, or schema extensions, require a Statement of Work (SOW) and are subject to additional costs.

6. Service Levels & Support

PhenoTips guarantees 99.5% availability, excluding planned maintenance, and is backed by a responsive support structure. First response to new tickets is guaranteed within one business day. Enterprise clients receive a dedicated account manager who oversees service quality, user satisfaction, and escalations.

In the event of service degradation or outages, critical support personnel, such as DevOps Managers and Technical Leads, are activated to restore service. The business continuity approach prioritises support functions over non-essential development during disruption, ensuring that clients receive continuous access to assistance and system functionality.

7. After-Sales Support

Following initial deployment, clients benefit from continued access to updated documentation, knowledge bases, and release notes. Refresher training sessions are available upon request, and ongoing configuration support may be arranged under an SOW. Where applicable, clients are assigned a dedicated account manager who facilitates service reviews, monitors adoption, and coordinates issue resolution. Clients can escalate complex cases for technical investigation and receive updates on feature development and roadmap alignment.

8. Technical Requirements

PhenoTips is fully browser-based and requires no local software installation. It is compatible with modern browsers including Google Chrome, Mozilla Firefox, Microsoft Edge, and Safari. Access to API functionality requires valid credentials and can be integrated into hospital systems using RESTful protocols. PhenoTips supports SAML 2.0 and OpenID Connect for Single Sign-On (SSO), enabling identity federation with institutional identity providers.

9. Outage & Maintenance Management

Outages and critical incidents are communicated to clients via direct email notifications. Additional communication methods may include scheduled update calls, depending on the severity and duration of the issue. Internally, Gene42 maintains fallback communication protocols to ensure operations and updates continue during service disruptions. These include alternatives such as Zoom, personal mobile phones, and even physical mail, in line with the company's business continuity framework. Major

incidents are reviewed as frequently as necessary to evaluate ongoing impact and response effectiveness.

10. Hosting Options & Locations

PhenoTips is hosted on cloud platforms including AWS and Microsoft Azure. Both platforms offer encryption, firewall protection, intrusion detection, and monitoring services. These cloud services comply with ISO/IEC 27001 and other relevant data protection frameworks. All UK based clients are hosted in data centers located within the appropriate region.

11. Access to Data on Exit

At the end of a contract, clients can export their data in widely supported formats such as CSV, JSON, as well as database exports. The export includes complete patient records, phenotypic annotations, user-generated data, audit logs, and user-provided attachments. Data is transferred securely and in compliance with information governance protocols. Gene42 offers validation and export assistance under a Statement of Work. Residual data on the platform will be securely deleted / cryptographically sanitised.

12. Security

PhenoTips enforces robust security measures aligned with industry standards and NHS requirements. Data in transit is encrypted using TLS 1.2 or above, while data at rest is encrypted using AES-256. Role-based access controls limit user privileges, and all user actions are logged. Multi-factor authentication (MFA) is supported, and institutions can integrate with their existing SSO infrastructure using SAML 2.0 or OpenID Connect.

The system undergoes annual penetration testing by external providers, including Bramfitt Security and Bulletproof, in compliance with security frameworks such as ISO/IEC 27001 and CSA CCM v4.0. Infrastructure is continuously monitored for anomalies, and an incident management process is in place to detect and respond to security events promptly. Security posture is documented and auditable.