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BioRepository@ELIXIR-IT: a computational environment for storing and sharing human genetic data

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Abstract. Nucleic acid sequencing is becoming more accessible, opening doors for new healthcare applications like precision medicine and pharmacogenomics. These could greatly improve treatments for conditions such as cancer and genetic diseases. However, to make the most of this, we need to address complex technical, legal, and ethical issues, regarding data management. This paper introduces BioRepository, a new integrated service by ELIXIR-IT. BioRepository is designed to manage human genetic data from its collection to its storage, supporting the use of genetic information in research and healthcare

Keywords. Genomic Data, Data Management, FAIR Data principles, Elixir, Secure Data Access

1. Introduction

Biorepositories can be defined as structured services designed to collect, store, manage, and distribute biological specimens associated data and metadata for research and clinical applications. These repositories play a pivotal role in biomedical research, enabling large-scale studies in genomics and precision medicine. By collecting high-quality omics sampling data — derived from genomics and proteomics analysis and sequencing — and linking them to relevant metadata, biorepositories can promote reproducibility, interope-

rability, and data sharing.
With the widespread adoption of cost-effective sequencing technologies, volume and complexity of genetic data have increased dramatically. This trend necessitates computing infrastructures capable of ensuring secure storage, controlled access, traceability, and compliance with regulatory frameworks and laws. The sensitive nature of genetic data, in particular, requires robust mechanisms for authentication, encryption, and enforcement of access policies.

2. BioRepository@ELIXIR-IT Service Overview

In order to meet these demands, ELIXIR-IT (1) has developed an integrated service for the management of human genetic data, encompassing the entire data lifecycle from sequencing to deposition. These services are built on a computational environment based on virtual machines (VM) infrastructure, resulting in a secure, scalable and user-friendly environment that can be tailored to the needs of researchers and clinicians. The system leverages the ReCaS (2) data center in Bari (Italy), part of the Italian Computing and Data Infrastructure (ICDI) (3) and the European Open Science Cloud (EOSC) (4). The platform adheres to the FAIR principles (Findable, Accessible, Interoperable, and Reusable) and is compliant with the European General Data Protection Regulation (GDPR) (5).

3. Core Requirements

To fully support genomic data management, the BioRepository service addresses several technical needs that are critical for secure and reliable operation. These include robust data security, scalable infrastructure, and the ability to support high-quality data processing pipelines within a reproducible and transparent framework.

Data Security

Sequenced data are encrypted and digitally signed using the CRYPT4GH (6) encryption suite, while all data transfers are secured via asymmetric-encrypted SSHv2 tunnels (using public and private keys in ED25519 format), ensuring that only authorized recipients can access the data. Digital signatures verify both data integrity and source authenticity. The platform ensures that private keys remain within a secure internal environment and that all access events are logged and auditable.

Scalability

The infrastructure is designed to support projects ranging from small cohort studies to large-scale national initiatives. It uses a high-capacity redundant Parallel Storage System (DELL Isilon – PowerScale), OpenStack (7), and Proxmox Virtual Environment (8) to manage virtualized environments. An underlying Ceph infrastructure provides distributed, fault-tolerant storage across multiple nodes, supporting high availability and elastic scalability.

Processing Quality

The infrastructure ensures that all components, from data uploading to final storage support processing reliability, traceability, and compliance with quality standards.

4. System Architecture

The infrastructure consists of two main components detailed in Fig.1. Each component is designed to address the challenges of handling sensitive data while ensuring global accessibility and full compliance with data protection regulations such as the GDPR. The key parts are represented by a secure BioRepository and a virtualized analysis environment. The BioRepository, hosted at CNR-ReCaS in Bari, offers 5 PB raw space for encrypted storage with geo-redundant backups at CNR-ITB (Milan, Italy) and CNR-ICAR (Naples, Italy). Both external (such as uploads of raw omics data from sequencer workstations and download of processed data to the recipient) and internal data transfers are protected via SSH tunnels with asymmetric keys encryption. The repository also hosts curated, versioned reference datasets which can be essential for bioinformatics pipelines.

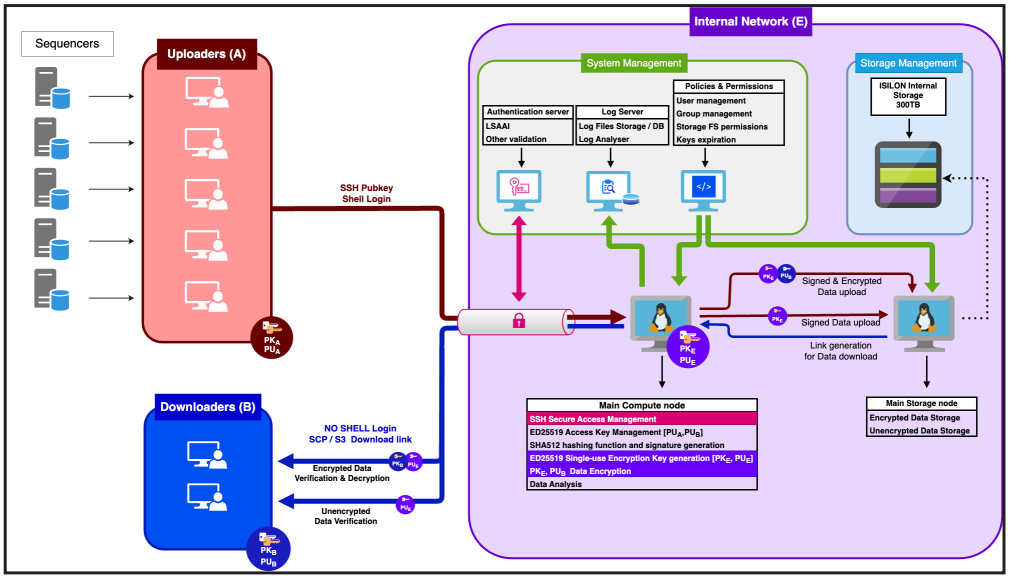

Fig. 1

BioRepository@ELIXIR-IT system architecture: visual representation of the secure data workflow across operational roles. Uploaders (A) send raw sequencing data through encrypted channels; the Internal Network (E) manages key generation, encryption, signing, logging, and storage; Downloaders (B) retrieve processed or raw data with integrity and authenticity validation. The infrastructure integrates policy enforcement, user management, and scalable encrypted storage

5. Secure Data Workflow

The system defines three operational roles: Uploaders (A), Downloaders (B), and the Internal Network (E).

- Uploaders submit raw data obtained from sequencers.
- Downloaders access processed data upon request.
- The Internal Network handles encryption, processing, and secure storage.

For each operational project, data workflow phases include:

1. Preparation: all users involved in an operational project must upload their ED25519

public keys to the system. Meanwhile, a CRYPT4GH public-private key pair PU(E) and PK(E) is created into the internal network for that operational project by the system administrator.

2. Upload: an encrypted transfer tunnel is established from the uploading workstation to the system network gateway. Data is transferred via SSH and then verified using SHA512 hash fingerprints.
3. Optionally, uploaded data can be processed using internal computing facilities, according to the specific agreement upon Institutes.
4. Hosting: sensitive data are encrypted using the download recipient public key PU(B), and digitally signed with a private key PK(E) generated into the system and unique for any different operational project, in order to grant data authenticity.
5. Download: authorized Downloaders acquire the public key PK(E) of the operational project, then they retrieve the processed data using a SSH secure tunnel. In the case of sensitive data, Downloaders can decrypt them using their own private key PK(B), while data integrity and authenticity is verified using the internal network Public Key PU(E). For non-sensitive data, the decryption part is skipped while integrity and authenticity can be verified using PU(E) as said.

Each processed dataset receives a unique identifier to support version control and traceability. All user actions are logged, and the logs are securely retained for auditing purposes. Once the download operation is complete or the intended hosting period of the operational project ceases, data and associated internal key pairs are securely deleted.

6. Infrastructure and Resources

While originally based on proprietary VMware ESXi [9], the Biorepository infrastructure currently relies on Proxmox VE, an open-source virtualization platform based on KVM/QEMU. Ceph [10] integration allows for efficient, resilient storage using OSDs, MONs, CRUSH maps, and Logical Volume Management (LVM). The system supports live migration of VMs, resource-aware scheduling, and automated failover.

7. Virtualized Analysis Environment

This environment also supports the deployment of customized, scalable bioinformatics pipelines executed on tailored VMs. VMs utilize encrypted filesystems and provide shared access to the BioRepository. High-availability configurations ensure minimal downtime, while snapshots and backup mechanisms can grant recovery and auditability of critical services.

Each VM is provisioned with:

- Up to 20 vCPUs (Intel Xeon E5/E7 with AVX-512)
- 200 GB DDR4 ECC RAM
- 500 GB local scratch storage
- 30 TB shared storage (NFS/iSCSI)

VMs can be equipped with one or two NVIDIA A100 GPUs to enable accelerated execution

of compute-intensive workflows such as Parabricks, DeepVariant, and Guppy.
Analytical environments can be replicated to support collaboration and benchmarking.

8. Software and Workflow Management

The platform supports containerized execution and environment management using:

- LXC and Docker for container isolation
- Conda and Mamba for dependency resolution and environment replication

To ensure processing consistency and traceability, bioinformatics workflows are containerized, version-controlled, and subject to rigorous quality assurance protocols. All analytical pipelines adhere to standardized sequencing data formats (e.g., FASTQ, BAM, VCF), and utilize metadata schemas aligned with the FAIR principles.
Audit trails and validation checks are embedded throughout the execution environment, ensuring reproducibility and transparency across different analyses. Containers are maintained in secure internal registries with automatic version tracking and security validation.

9. Conclusions

The BioRepository@ELIXIR-IT platform offers a secure, scalable, and standards-compliant solution for the full lifecycle management of human genetic data. By integrating open-source technologies such as Proxmox VE, Ceph, and CRYPT4GH, and by enforcing strict data protection policies, the platform ensures high levels of performance, interoperability, and trust.
Its modular and containerized architecture also supports a wide range of bioinformatics workflows, while GPU acceleration, live migration, and automated backups enhance computational efficiency and resilience. This platform serves as a forward-looking model for infrastructures supporting precision medicine, collaborative genomic research, and ethical data sharing in biomedical science.

Acknowledgments

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Angelo Sante Varvara is a PhD student within the European School of Molecular Medicine (SEMM).

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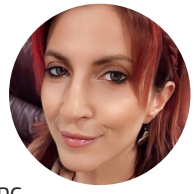

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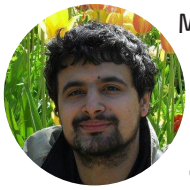

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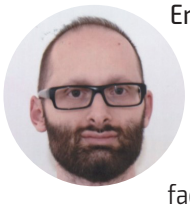

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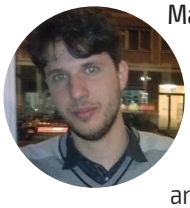

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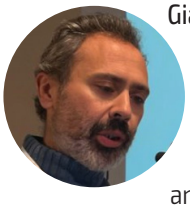

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