Achieving data FAIRification in a distributed analytics research platform for rare diseases

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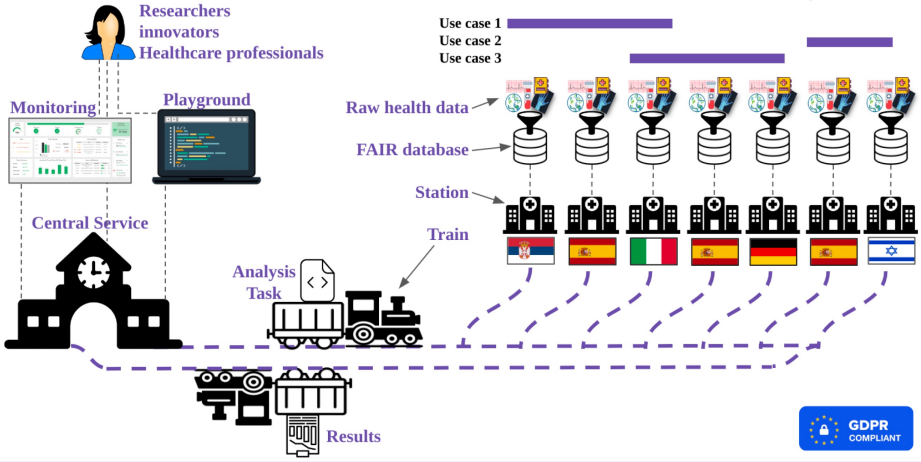
Goal

Achieving findability, accessibility, interoperability, and reusability (**FAIRification**) of data, metadata, and study results within a **network of several medical centers** participating in the BETTER Horizon Europe project, targeting the study of **rare diseases** (such as intellectual disability and inherited retinal dystrophies)

Better rEal-world healTh-daTa distributEd analytics Research platform

- Horizon Europe project started Dec. 1st, 2023 (https://www.better-health-project.eu/)
- Design and implementation of a decentralized infrastructure to exploit the full potential of large sets of multi-source health data.
- Various use cases involving 7 European medical centers providing sensitive patient data (e.g., clinical reports, medical images, genomic data, biological data, metabolic, environmental and demographic data, patient interviews, …)
- Only the secure information made available and analyzed with a GDPR-compliant mechanism via a Distributed Analytics paradigm (Personal Health Train)

Personal Health Train paradigm



- Railway system analogy that includes trains, stations, and train depots [1].
- **Train** transports goods (= **analytical tasks**).
- **Station** (= **data provider**), accessible by the Train. It executes the task, which processes the available data.
- **Depot** (= **Central Service**). It includes procedures for Train orchestration, business and operational logic, data management and discovery [2].
- Further modules for privacy and security enforcement.

Health datasets FAIRification and preprocessing

Our group is involved in overcoming cross-border barriers to health data integration, access, FAIRification, and preprocessing. Practical objectives include:

(1) Discovering and collecting datasets available at each medical centre, anticipating interoperability with external databases.

(2) Designing a unifying repository schema useful for integration (exploiting, e.g., FHIR HL7 standards) [3].

(3) Prepare ETL for processing health datasets (**reusability**) [4].

(4) Harmonizing data by employing standardized terminologies and ontologies (**interoperability**) [5].

(5) Loading aggregated information and metadata into the project's repository (**findability** and **accessibility**).

Clinical Use Cases

- Integration of genomic and phenotypic data from paediatric rare diseases to decipher pathways of intellectual disability
- Accelerate Inherited Retinal Dystrophies Diagnosis using AI
- Predicting the risk of self-harm and suicidal behaviors in patients with Autism Spectrum Disorders

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