GENOMED4ALL

Genomics and Personalized Medicine for all through Artificial Intelligence in Haematological Diseases

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Genomics for Next Generation Healthcare

Our mission

GenoMed4All is the European initiative to transform the response to Haematological Diseases by seizing the power of Artificial Intelligence

The project represents a quantum leap in advanced precision medicine, pooling genomic/ '-omics' health data through a secure and trustworthy Federated Learning platform.

Our disruptive AI models, scaled up by High-Performance Computing, will boost the processing capacity of data repositories from 10 clinical sites across Europe, empowering forward-thinking research of common and rare Haematological Diseases

Meet the Team

23 organizations from 8 EU countries

Unleashing the power of AI

Our ambition

The massive connection of -omics and clinical data repositories across Europe offers:

More **accurate** Deep Neural Networks, VE and advanced generative models using genomic and other omics information, clinical and contextual information

Optimal fusion architectures to obtain novel knowledge which cannot be made without the use of AI in a sufficient number of data, and standardization of genomics data generated from different platforms and clinical partners: defining genomics data exchange format, standardize phenotypic data using phenopackets

What are we aiming for?

Starting from 10 to reach 86+ Clinical repositories with genomics data connected across 15 EU countries

15% Accuracy improvement in specific genomic markers for prognosis and treatment

20% Increase in the adoption of open standards for -omics data per clinical site

80% Time reduction in AI analysis and model training through Supercomputing

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An open source data hub for haematological diseases

Our principles

Innovation

Using Graph Neural Networks to distributedly train deep learning algorithms

Interoperability

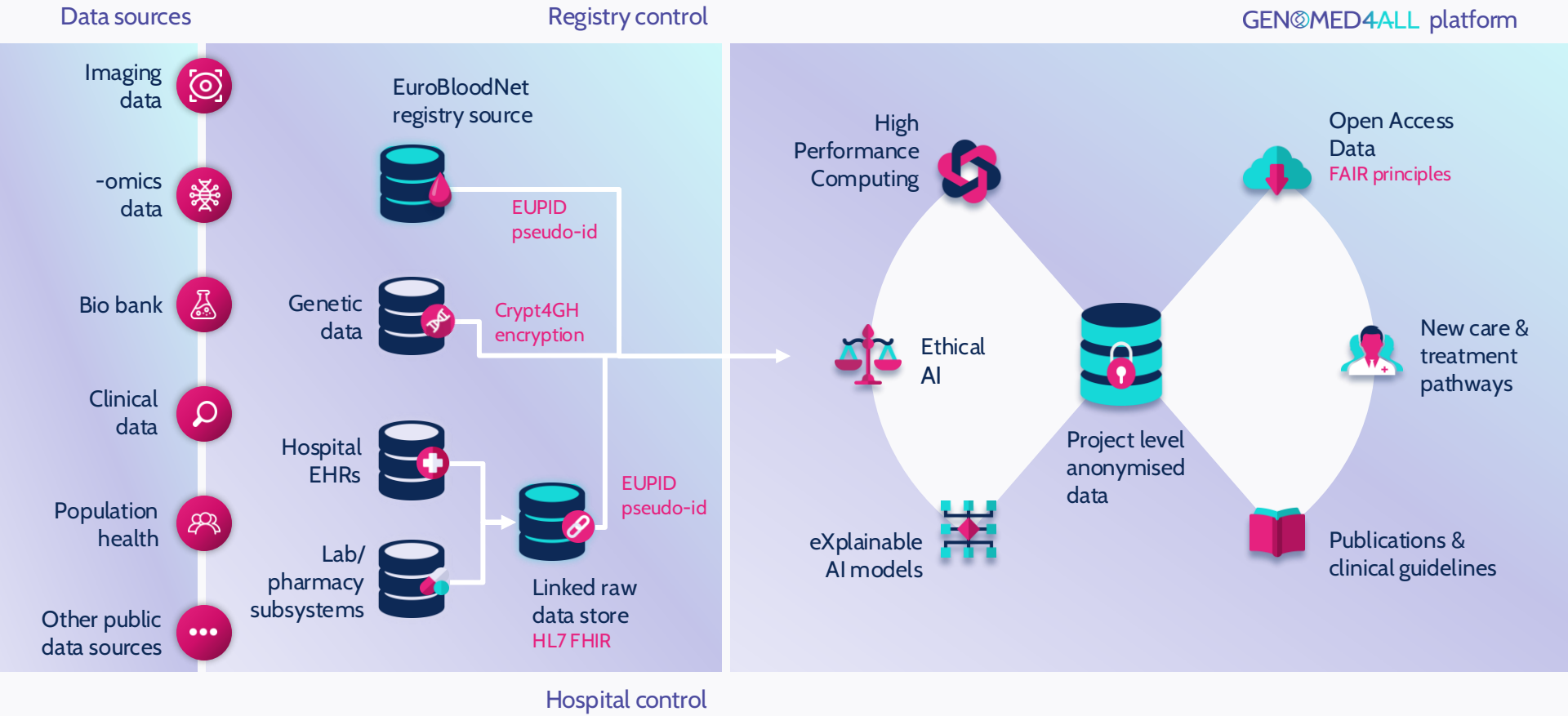
Enabling collaborative, cross-border data sharing that is standard-compliant: Phenopackets

Trust

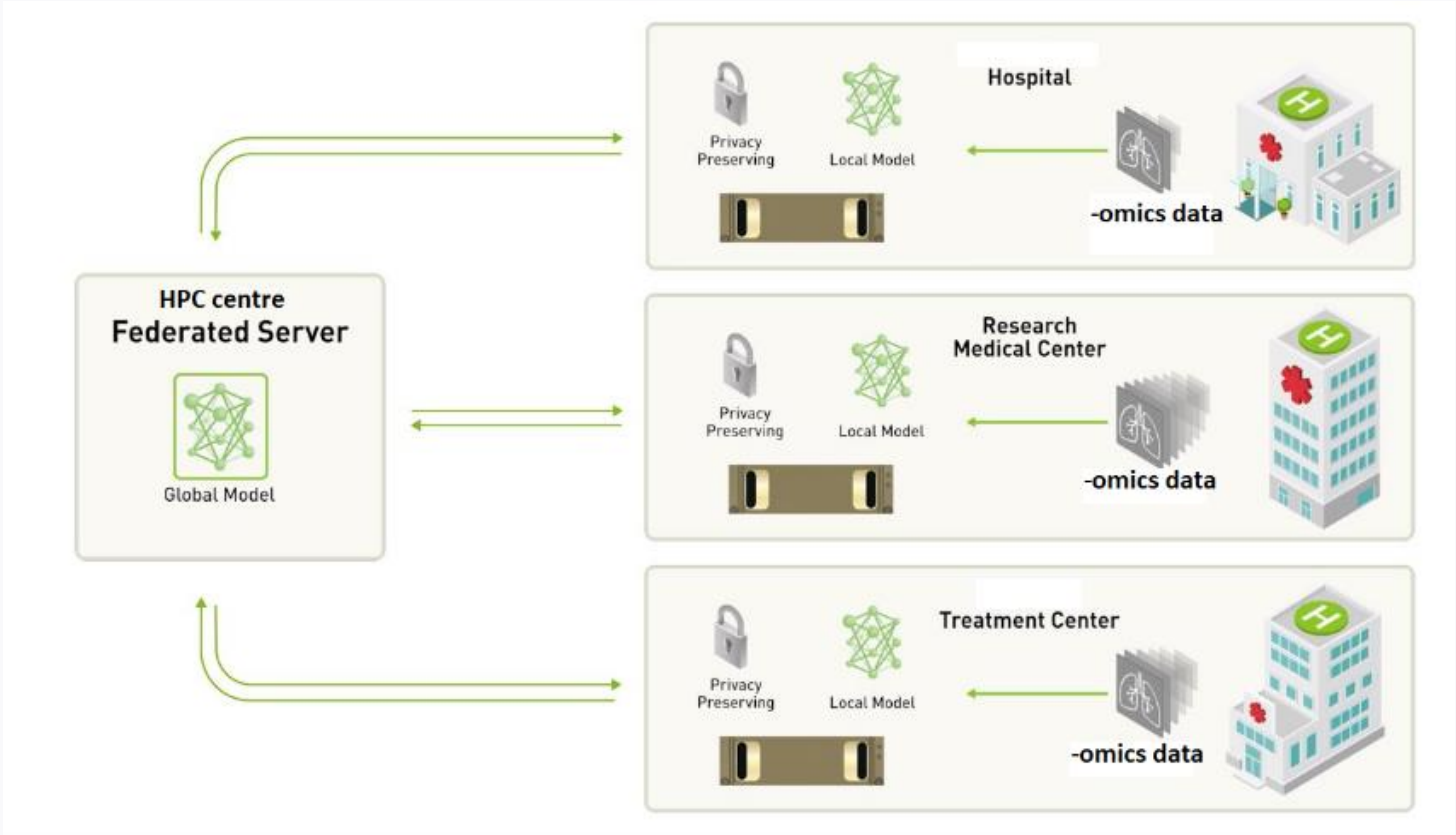
Offering security and privacy by design in all data exchanges, model training and storage

Scalability

Using containerization to build modularity and facilitate massive replicability


Data sources
Registry control
GENOMED4ALL platform
Imaging data
-omics data
Bio bank
Clinical data
Population health
Other public data sources
EuroBloodNet registry source
EUPID pseudo-id
Genetic data
Crypt4GH encryption
Hospital EHRs
Lab/ pharmacy subsystems
EUPID pseudo-id
Linked raw data store
HL7 FHIR
High Performance Computing
Open Access Data
FAIR principles
Ethical AI
Project level anonymised data
New care & treatment pathways
eXplainable AI models
Publications & clinical guidelines
Hospital control

Federated Learnign platform approach:
A privacy respectful secure machine learning framework



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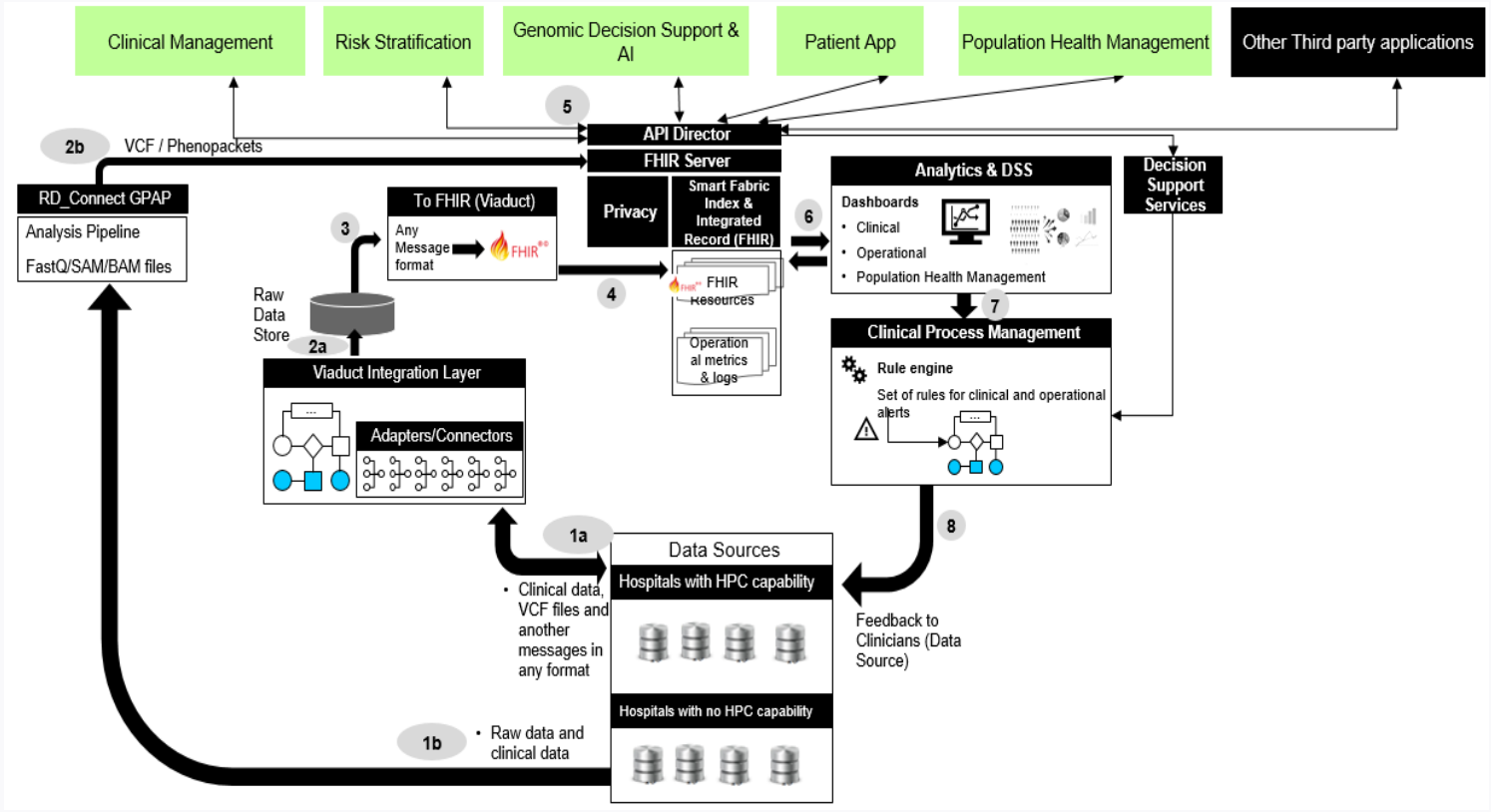
Data flow in the platform (for your files!)



The context for Haematological Diseases

Our challenges

Most have a genetic background

There are up to 450 variants (oncological and non-oncological resulting from abnormalities in blood cells, lymphoid organs and coagulation factors

They represent a growing public health challenge

Haematological malignancies account for 5% of cancers, most can cause chronic health problems and many are life-threatening conditions

EU repositories are unconnected

The number of available samples for haematological disorders remains small and there are currently no centralized big data repositories

Exploring new models in genomics for precision medicine

AI- based services for clinical support

GenoMed4All will deploy 'white box' AI models in 3 real-world pilots for common and rare oncological (**Myelodysplastic syndromes** and **Multiple Myeloma**) and non-oncological (**Sickle Cell Disease**) haematological diseases

Diagnosis

AI algorithms for early identification of high-risk individuals

Prognosis

Prediction algorithms for insights on disease development

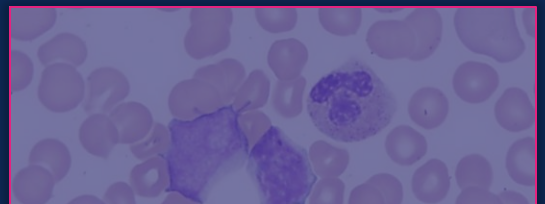
Treatment

Clinical algorithms to aid decision-making in risk stratification

Myelodisplastic syndromes

The disease

Myelodysplastic syndromes (MDS) are a group of bone marrow failure disorders that typically affect the elderly. Patients suffer from blood cytopenia (low blood cell counts), since their bone marrow is no longer able to produce enough healthy blood cells. The disease is also known as a form of blood cancer, and in some patients can evolve into acute myeloid leukemia (AML), which is usually fatal if not treated.



Validation

Prevention based on Genomic Screening

Investigate factors that influence the development of MDS, enabling early-stage identification of individuals at risk.

Omics-based Classification and Prognosis

Personalized predictive models through integration of comprehensive genomic and clinical information.

Omics-based Clinical Decision Making

AI-based algorithms to stratify the individual probability of response to specific treatments.

Drug Repurposing

Build a rationale for drug repurposing in specific subsets of MDS.

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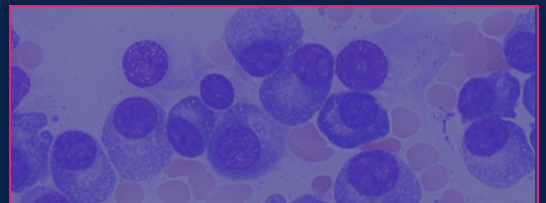
Multiple Myeloma

The disease

Multiple Myeloma (MM) is a type of bone marrow cancer originating in plasma cells, a type of white blood cell responsible for producing antibodies to fight off infections. In patients with MM, cancerous plasma cells accumulate in the bone marrow and produce abnormal proteins instead, which can lead to decreased blood cell numbers, bone and kidney damage.



Validation

Understand Disease Complexity

Describe the different layers of MM heterogeneity integrating baseline genomic and imaging data.

Identify Evolution Dynamics

Define the quantitative and qualitative dynamics of the disease in time.

Study Risk Progression

Develop a prognostic risk score for the baseline and the disease remaining after therapy.

Integrate Radiomics and Radiogenomics

Develop and validate a model to predict treatment response and determine progression-free survival.

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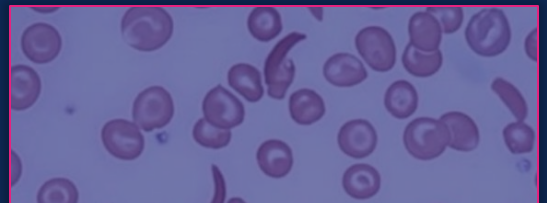
Sickle Cell Disease

The disease

Sickle Cell Disease (SCD) is a group of hereditary red blood cell disorders. It is a rare, chronic and life-threatening disease, in which red blood cells become C-shaped in resemblance to a sickle, the farming tool the disease is named after. Sickle cells die early and tend to clog the blood flow when going through small blood vessels, so patients usually suffer from low red blood cell counts, infections, acute chest syndrome and strokes.



Validation

Identify gene mutations associated to inflammation markers

Correlations between genetic inflammatory risk profiles CRP level to develop high inflammation prediction models.

AI allocation of SCD patients to a sickling risk profile

Understand which genetic loci (GWAS) are associated with SCD patient-specific blood rheology and the point of sickling (PoS).

Develop a combined model to predict clinical outcome

Using the extent of renal damage expressed as microalbuminuria as gold standard, together with other known genetic modifiers.

AI-based Radiomics

Build a probability score using AI-based brain MRI image analysis to predict incidents of silent infarction in young SCD patients.

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USE CASES – DATA COLLECTION

- Available data from GENOMED4ALL partners
- EU Reference Networks on RD (ERN)
- EU REGISTRIES
- EXITING REPOSITORIES / RESEARCH NETWORKS

CONNECTIONS WITH EU FUNDED INITIATIVES OR INFRASTRUCTURES

DATA COLLECTION 1 - data from EXITING REPOSITORIES / RESEARCH NETWORKS

- 20,012 patients with Myeloid Neoplasms
- 103 centers across Europe, America, Asia and Australia

- Diagnosis:
  - 6,311 Acute Myeloid Leukemias
  - 7,378 Myelodysplastic Syndromes
  - 2,597 Myeloproliferative Neoplasms
  - 3,726 Myelodysplastic/Myeloproliferative Neoplasms

DATA COLLECTION 2 - Available data from EU REGISTRIES

SICKLE CELL DISEASE - USE CASE

- ENROL, the European Rare Blood Disorders Platform, has been conceived in the core of ERN-EuroBloodNet as an umbrella for both new and already existing registries on RHD. ENROL aims at avoiding fragmentation of data by promoting the standards for patient registries' interoperability released by the EU Rare Disease platform.

- ENROL will map at the EU level demographics, survival rates, diagnosis methods, genetic information, main clinical manifestations and treatments in order to obtain epidemiological figures and identify trial cohorts for basic and clinical research.

- ENROL will enable the generation of evidence for better healthcare for RHD patients in EU as ultimate goal.

RADeep Rare Anemia Disorders European Epidemiological Platform

Strategy for data sharing within Genomed4all

RADeep is an initiative endorsed by ERN-EuroBloodNet for pooling data from patients affected by a Rare Anemia Disorders (RAD)

- RADeep is built in line with the EU Rare Disease Platform recommendations for patients' registries on rare disorders and ENROL
- RADeep contributes to ENROL by sharing a sub-set of patients' pseudonymised data

➡ possibility to share and pool data

➡ reach critical numbers

➡ perform clinical trials, research projects

➡ knowledge generation (evidence)

➡ **better care for RADs patients**

RADeep

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Other techniques supported by AI

Synthetic and causal modalities

- In order to help the federated learning process and allow further research, there are some research lines in AI as well to be explored
- Synthetic data generation: increasing available data in 2 ways
  - Same feature space to generate similar patients
  - Different feature space to generate novel patients not following the same pattern so novel
- In addition it is providing anonymization to the data!
- In the research of causal models (causal Deep learning) and better explanability of the results of AI
- Better optimal fusion of clinical, genomic and radiomics information

Thanks!

Any questions?

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