ERN | EuroBloodNet

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GenoMed4All & SYNTHEMA AI Federated Platforms in hematology

Providing a new research data infrastructure and AI models for ERN-EuroBloodNet clinical networks addressing oncological and non-oncological diseases

Speaker: Prof. Federico Alvarez,
Universidad Politécnica de Madrid

European Reference Network | Hematological Diseases (ERN EuroBloodNet)

Funded by the European Union

Disclosure of Conflict of Interest

The speaker declares no conflicts of interest

GENOMED4ALL

Genomics and Personalized Medicine for all through Artificial Intelligence in Haematological Diseases (2021-2025)

GENOMED4ALL receives funding from the European Union's Horizon 2020 Research and Innovation programme under Grant Agreement No. **101017549**

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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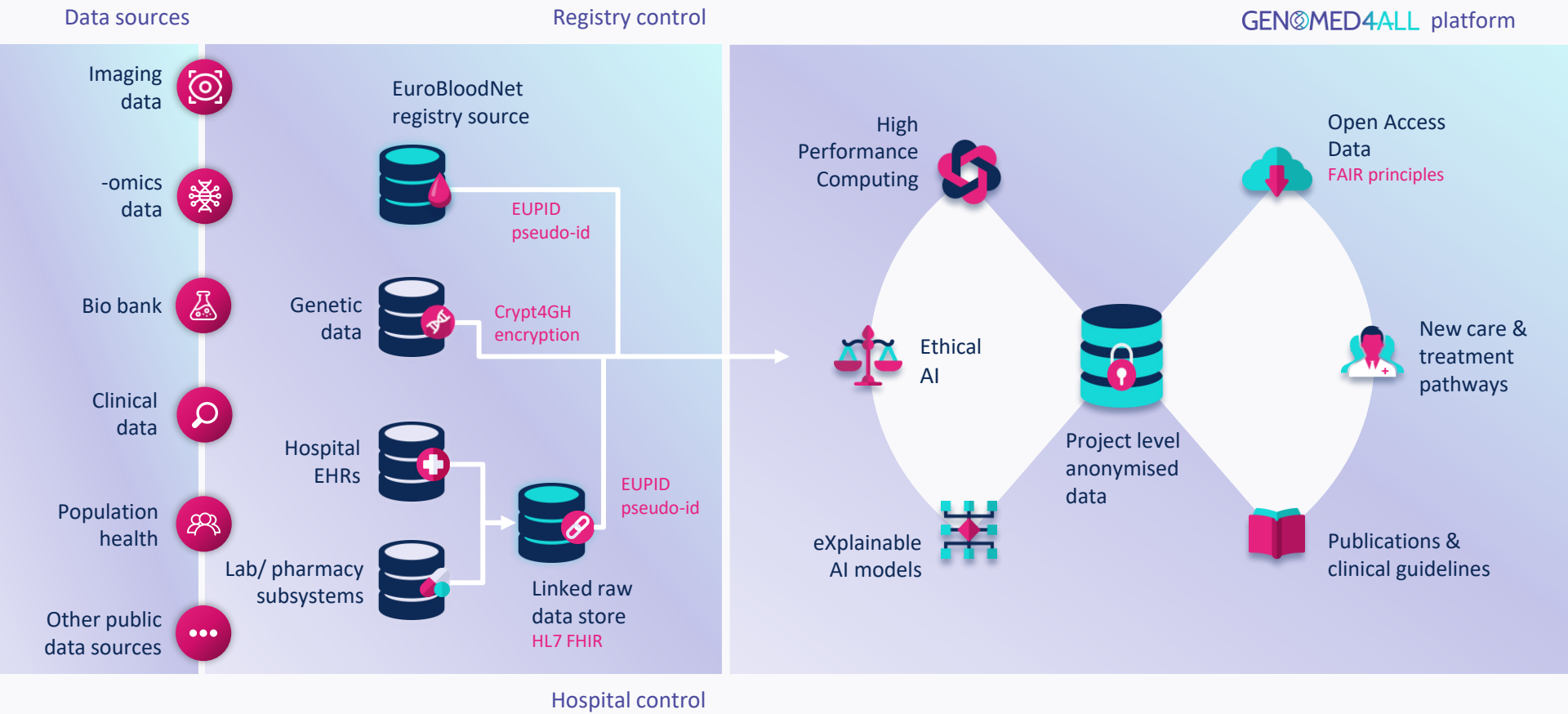
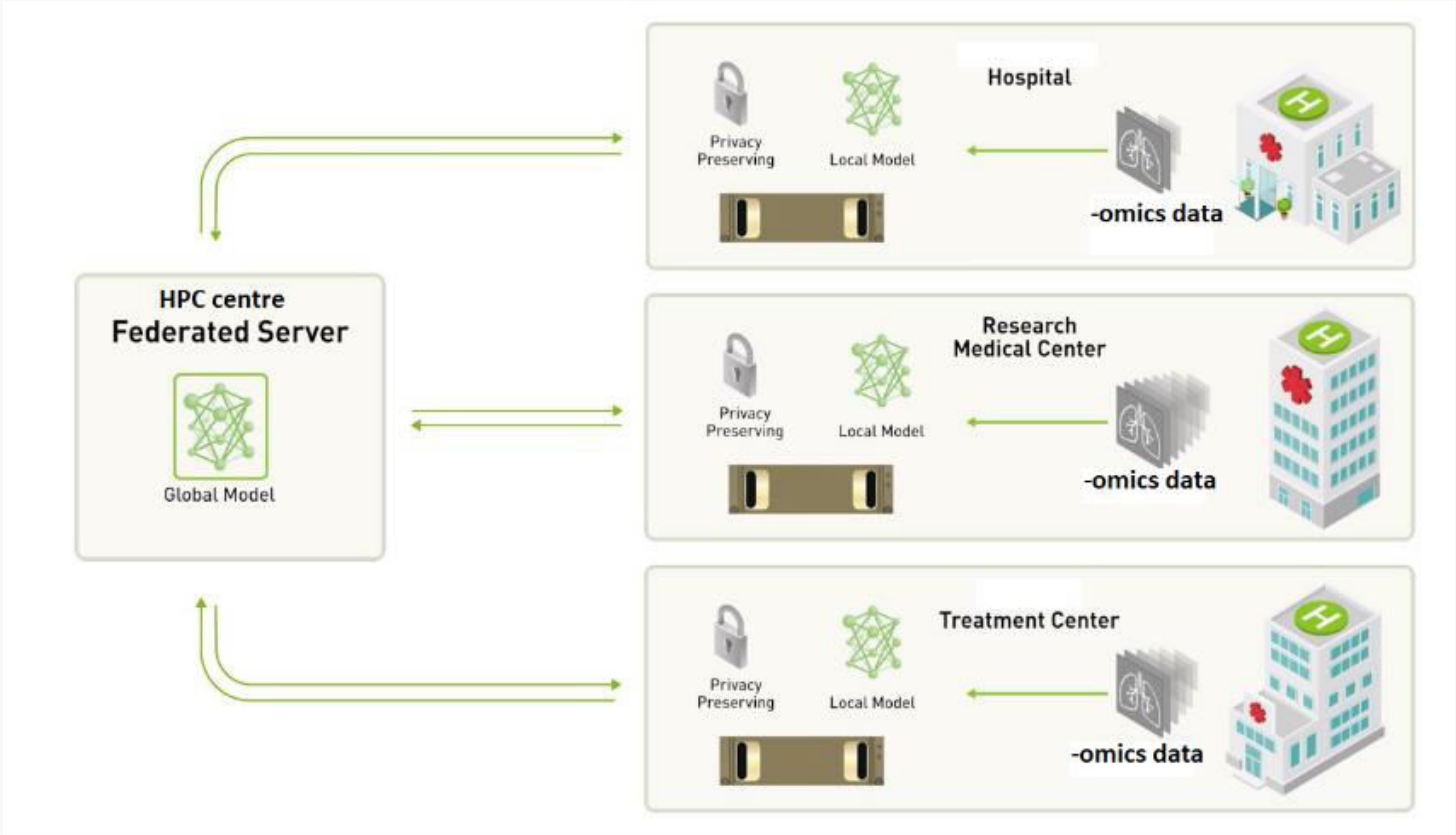

Data sources
Registry control
GENOMED4ALL platform
Imaging data
-omics data
Bio bank
Clinical data
Population health
Other public data sources
EuroBloodNet registry source
EUPID pseudo-id
Genetic data
Crypt4GH encryption
Hospital EHRs
Lab/ pharmacy subsystems
EUPID pseudo-id
Linked raw data store
HL7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

Federated Learnign platform approach:
The approach is going to be used in SYNTHEMA, we should not reinvent the wheel



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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

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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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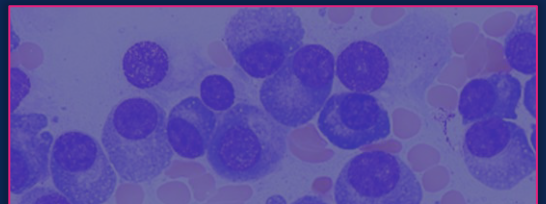
Multiple Myeloma

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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Synthetic haematological data over federated computing frameworks (2022-2026)

Funded by
the European Union

Our Mission

Establish a cross-border hub to develop and validate Artificial Intelligence (AI) techniques for anonymisation and **synthetic data generation to tackle the scarcity and fragmentation of data and widen the basis for GDPR-compliant research in rare hematological diseases (RHD).**

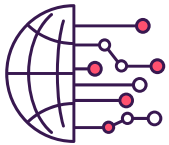
The Key Drivers

Synthetic data generation

To generate virtual patients that keep patterns and features of real-world data

Federated Learning

To facilitate the collaborative training of AI models with no sharing of raw data



Secure multi-party computation

For lower risk, privacy-preserving model aggregation in FL schemes

Differential privacy

To set strict boundaries on the disclosure of private data each clinical site is allowed

Value proposition

SYNTHEMA aims to generate reliable, **high-quality synthetic data** that can shape new **virtual patients** to further **enhance diagnostic capacity, assess treatment options and predict outcomes** in rare haematological diseases.

The platform will be based on a **privacy-preserving federated learning (FL) network**, equipped with **secure multi-party computation (SMPC)** protocols and **differential privacy (DP)**, connecting health data centres, academic research centres, industries and SMEs to advance translational and clinical research and care in RHDs.

Our Use Cases

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 patients with SCD, red blood cells become C-shaped in resemblance to a sickle, the farming tool the disease is named after.

Cohort

The SCD use case will include **2,000+ patients** with genetic diagnosis for SCD disorder, including paediatric (1+ year-olds) and adult patients (exclusion criteria: transplanted patients, 1 year-old or younger).

Acute Myeloid Leukaemia

The disease

Acute Myeloid Leukemia (AML) is a type of blood cancer that starts from young white blood cells in the bone marrow. The bone marrow produces white blood cells called granulocytes or monocytes too quickly because they grow and divide too fast.

Cohort

The AML use case will include **2,500+ patients** with "de novo" AML (2016 WHO classification criteria) and **2,000+ patients** with myelodysplastic syndrome (MDS), a pre-leukemic condition that can evolve into AML and allows us to study the transition between the chronic and acute disease phase.

The Team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Vall d'Hebron
Institut de Recerca
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HÔPITAUX
DE PARIS


CHARITÉ
UNIVERSITÄTSMEDIZIN BERLIN


HOSPITAL DA LUZ
LEARNING
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TRAINING, RESEARCH
& INNOVATION CENTER


UNIVERSITÀ
DEGLI STUDI
DI PADOVA


netcompany
intrasoft

Federated Platform offer to EurobloodNET

Synthema and GenoMED4All enjoys the support, resources and active participation of **ERN-EuroBloodNet**, as the European Reference Network on rare haematological diseases (RHDs).

The platform, along with other developments in GenoMED4All will be offered as open for research supporting the federated processing of clinical data among different clinical sites, with the aim of improving the research in hematological diseases and with a non-commercial approach. There will be different uses for clinical research among sites such as:

- Analysis with powerful AI algorithms
- Synthetic data generation
- Statistical analysis models
- Multimodal omics integration and whole analysis (images, genomics, tabular, …)

- Without the need to transfer data from the original site.

Due to their specificities, 2 data platforms will be offered: one for oncological diseases, one for non-oncological diseases. The deployment and testing is currently happening and interested members can contact for more details. 2 clinical sites offered to act as central providers of the platform, whereas UPM will provide the support to technical matters.

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Time for questions

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THANK YOU!

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