GenoMed4All

Artificial Intelligence-based Deep Learning algorithms for patients with Sickle Cell Disease

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Keywords Sickle Cell Disease | Artificial Intelligence

Objective

Improve **prediction of SCD disease severity** by developing AI-based **Deep Learning** algorithms.

Background

- Sickle Cell Disease (SCD) is a **hereditary red blood cell disorder** characterized by **hemolytic anemia**, periodic painful **ischemic vascular occlusion** and **long-term multiorgan failure**.
- Pathophysiology of SCD is not completely understood and **disease phenotypes vary** largely.
- The only curative treatment is **hematopoietic stem cell transplantation**. This is however **limited** in its availability and not without **risks**.
- **ERN-EuroBloodNet** now **leads a SCD use case** within the GenoMed4All initiative.

Patients & Methods

ENROLLMENT

- **1,000 steady state SCD patients** over 1 year of age from ten European hospitals, with **HbSS**, **HbSβo**, **HbSC** and **HbSβ+** genotypes will be enrolled.

DATA REQUIREMENTS

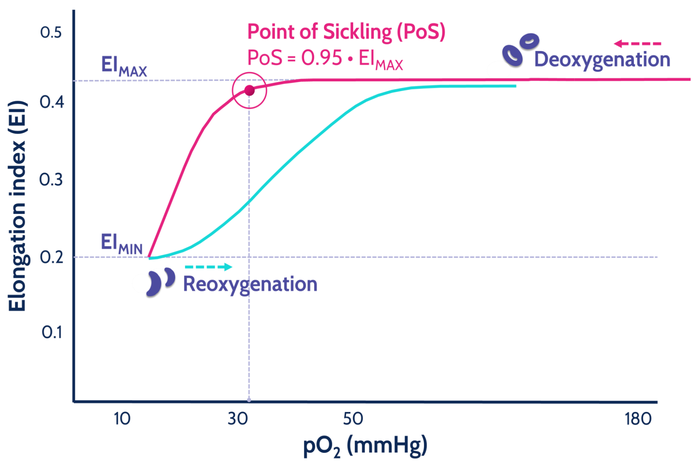
- Standardized collection of data will be performed, including **GWAS**, **metabolomics**, **radiomics**, **Lorrca Oxygenscan**.

CLUSTERING

- Based on predefined and newly identified Single Nucleotide Polymorphisms (SNPs), **patient clustering** will be performed.

AI MODELLING

- Integration of collected covariates will **facilitate Deep Learning AI-based algorithms**.



Representative Oxygenscan curve and Point of Sickling (PoS)

Update on results

- **Study protocol** has been **validated** and **approved**.
- **Enrollment** of patients has just started in two hospitals. All 1,000 patients will be enrolled by **2023**.
- Deep Learning algorithms will be developed and validated to achieve **overall aims** of GenoMed4all (see **Aims of the Sickle Cell Disease use case**).
- Following **a stepwise-approach**, we will first predict **single clinical outcomes** and **radiomics** patterns, then develop predictions of **complete clinical phenotypes**.

Aims of the Sickle Cell Disease use case

AIM 1	Investigate **SNPs associated with C-Reactive Protein (CRP)** levels in SCD
AIM 2	Investigate **SNPs associated with Point of Sickling (PoS)** using the Lorrca Oxygenscan
AIM 3	Predict **microalbuminuria** in SCD
AIM 4	Predict **silent cerebral infarctions** based on radiomics of the brain
AIM 5	**Predict clinical outcomes**: ischemic stroke, hemorrhagic stroke, vaso-occlusive crisis (VOC), acute chest syndrome, microalbuminuria, liver failure and retinopathy

Diagnosis	Prognosis	Treatment
AI for early identification of high-risk individuals	Predictions for insight on disease development	Clinical decision support models for risk stratification

GenoMed4All's AI models for hematological diseases

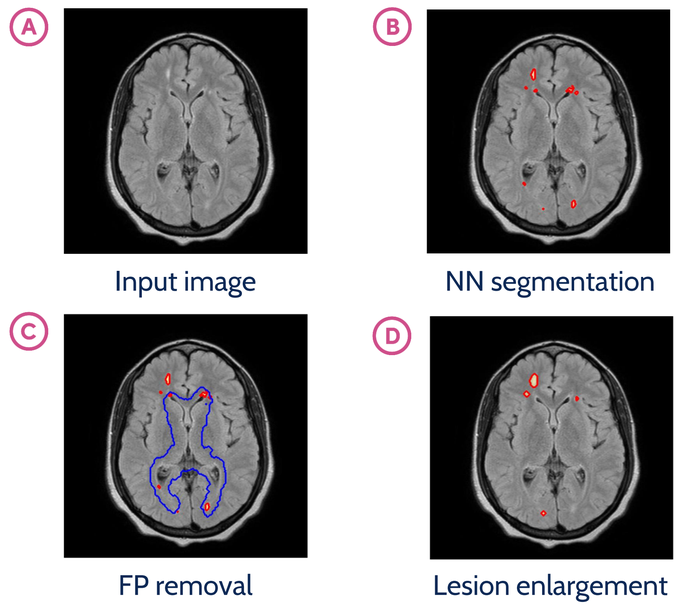
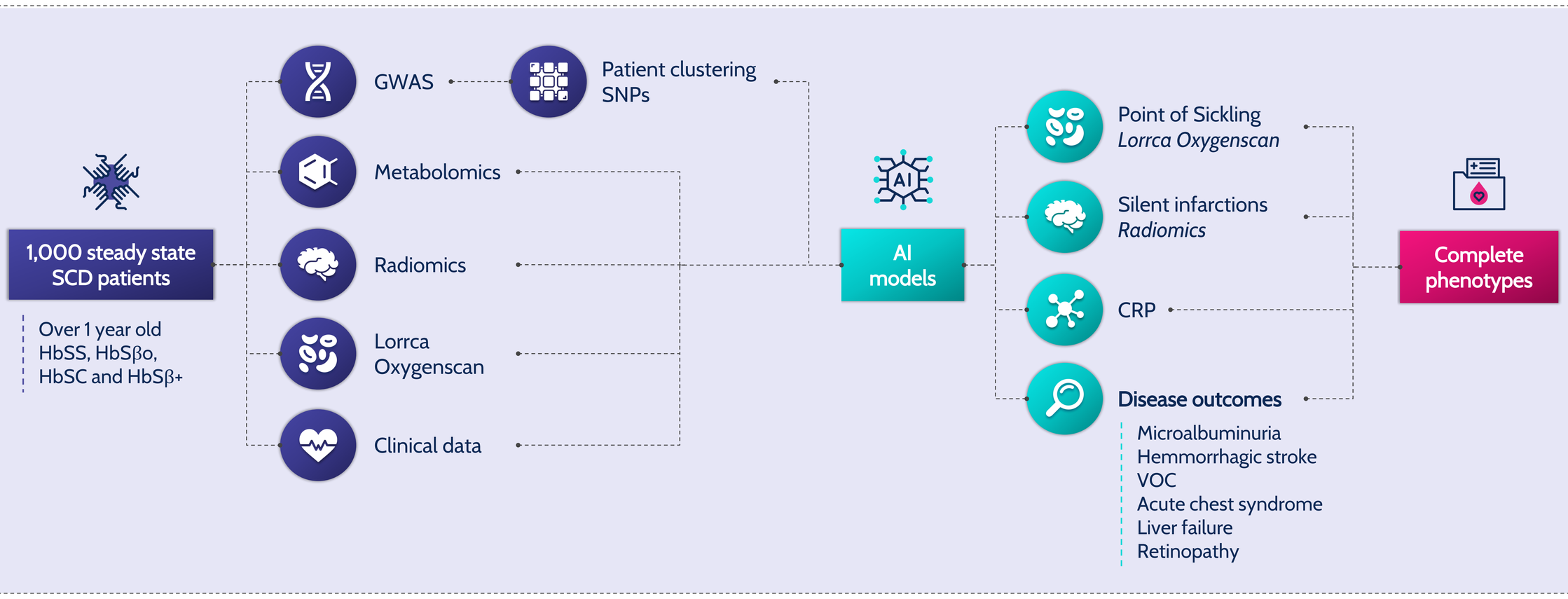


Image Segmentation Pipeline. A. Input image, B. Neural Network results (thr = .45), C. Defined area for FP removal, D. Enlarged region by active contours

Conclusions

- The widespread use of AI has already modified diagnostic research. Combining data of 1,000 SCD patients in GenoMed4All is a necessary step to **improve robustness of study results**.
- Concomitantly, **standardization** and **linkage** of **SCD data repositories** is promoted through this ERN-EuroBloodNet collaboration. Full compliance with **data protection legislation** and **ethical principles** is safeguarded.
- Implementation of AI algorithms and linking of SCD health repositories promises to **enhance diagnostics**, **predict disease outcomes** and **individualize treatment** options, so urgently needed for SCD.



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