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AMIGO Proposal:
A Concrete Use Case for Paediatric Innovation
From Fragmented Data to Integrated Translational Pathways

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AMIGO in one sentence

A federated European paediatric multi-omics infrastructure for precision medicine

Monday
Routine clinical sample
Genomics + omics + phenotypes

Federated AI
Algorithms travel across sites
No raw data leave the clinic

Friday
AI-assisted diagnosis
Therapy and repurposing hypotheses

Take-home message: draw blood on Monday, run genomics and multi-omics locally, and by Friday provide diagnosis support from a model trained federated across all participating paediatric clinics.

Why this matters for seriously ill children

≈30%
maximum diagnostic yield reported for Whole Exome Sequencing in this cohort context

8,000+
rare diseases mean extreme fragmentation of patient examples

100k
scale needed to learn across many rare paediatric disease patterns

The bottleneck

One hospital sees too few comparable cases for robust AI.

Genomics alone often does not explain complex phenotypes.

Clinical, omics and longitudinal data remain fragmented across institutions.

AMIGO response

Link multiple data modalities through a clinical knowledge graph.

Train and evaluate models in a federated way across hospitals.

Move from diagnostic support to translational hypotheses for therapies.

Concrete use case for EPTRI: paediatric innovation becomes data-driven, privacy-preserving and scalable across Europe.

Clinical foundation: SCIVIAS and prospective AMIGO-S

SCIVIAS cohort

Currently around 2,500 children; expansion target around 5,000.

Age 3–17 years, disease-overarching design with controls.

Two visits: early disease phase and follow-up after therapy.

Clinical metadata, routine labs and longitudinal course are captured.

Data modalities

Genomics	Transcriptomics	Proteomics
Metabolomics	bulkRNA / scRNA	OCT imaging
Detailed phenotypes	Doctor letters	Clinical labs
Medical history	HPO / SNOMED CT	LOINC2HPO

AMIGO uses this Munich foundation as a starting point and aligns it with additional European paediatric sites.

From local excellence to European evidence scale

Why federation is necessary

Rare diseases are too diverse for a single-centre AI training strategy.

The signal is distributed across hospitals, countries and data systems.

AMIGO brings the algorithm to the data instead of moving sensitive data to a central repository.

Existing biological knowledge helps reduce the amount of patient data needed for learning.

Use existing knowledge: clinical knowledge graph integration

Graph layer

Connect patients, phenotypes, genes, proteins, pathways, variants and diseases.

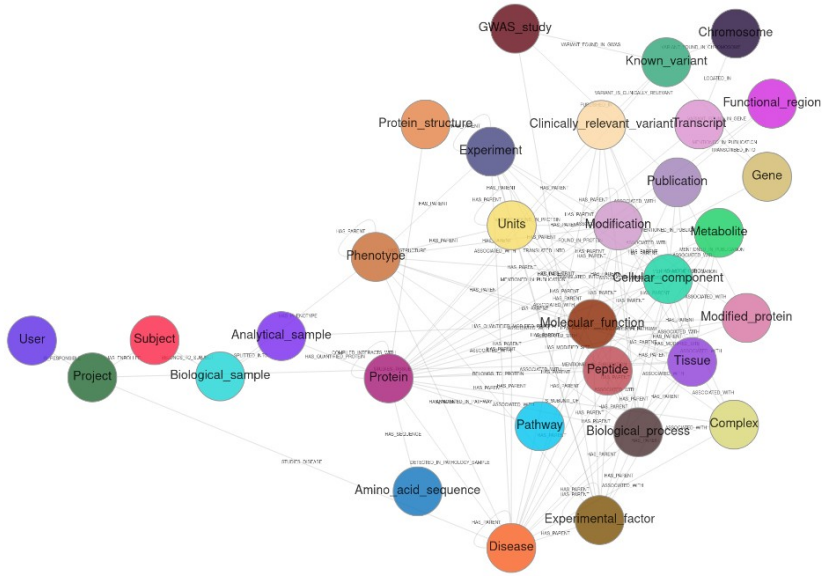
Map EHR text and questionnaires to HPO / SNOMED CT; map routine labs via LOINC2HPO.

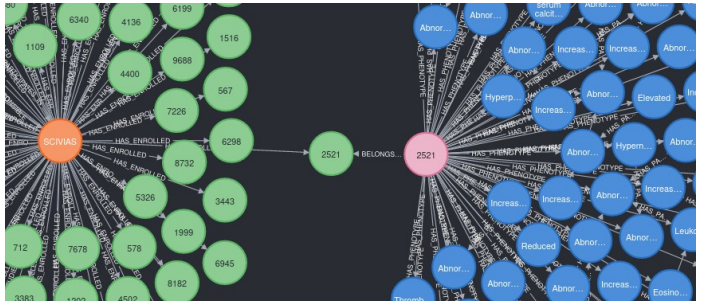
Enable phenotype-driven gene prioritisation, patient clustering and pathway-level hypotheses.

Provide a common representation for AI models and translational interpretation.

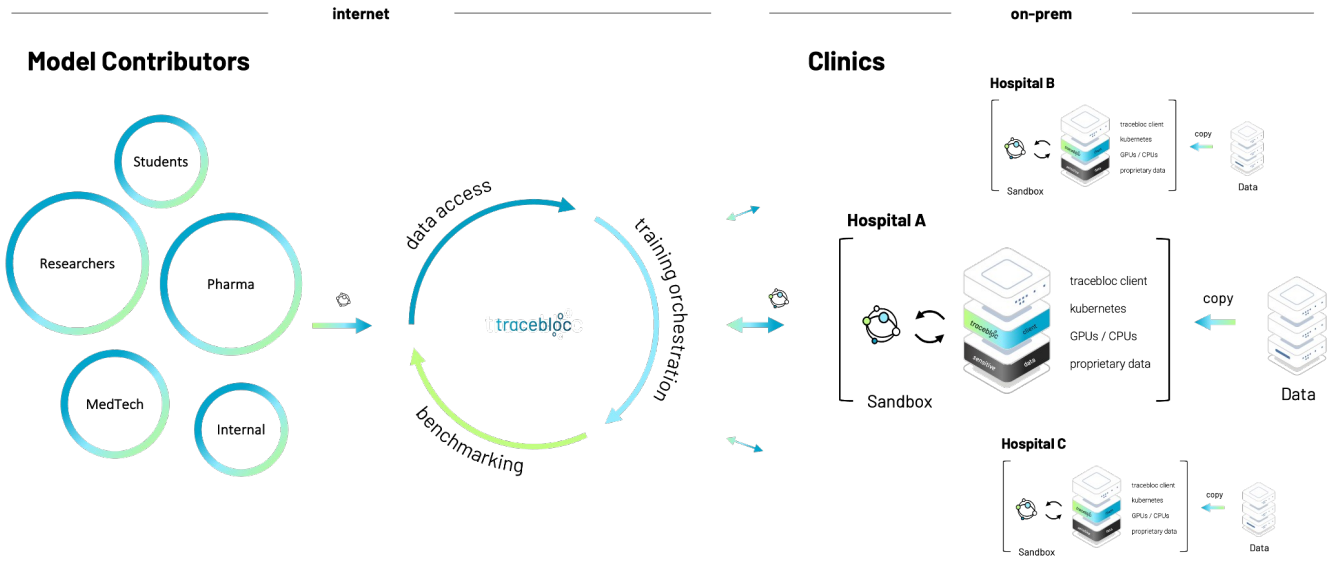
Why it matters

The graph reduces sparsity: models can learn from biology, not only from raw patient counts.





Bringing third-party AI models into the clinic



Model contributor perspective

Developers can contribute algorithms without receiving raw clinical or omics data.

Execution, logging and evaluation become reproducible and auditable.

Only approved outputs, metrics or aggregate results leave the clinic.

Clinical-site perspective

Hospitals keep governance over data and approved use cases.

Sites can compare models and retain control over implementation decisions.

The same architecture can support diagnosis, cohort feasibility and translational research.

Integrated translational pathway: from fragmented data to impact

Primary use

AI diagnostic apps and model benchmarking on comparable multi-omics data.

Patient matching for existing drugs and off-label/repurposing hypotheses.

Trial recruitment and cohort feasibility across paediatric centres.

Secondary use

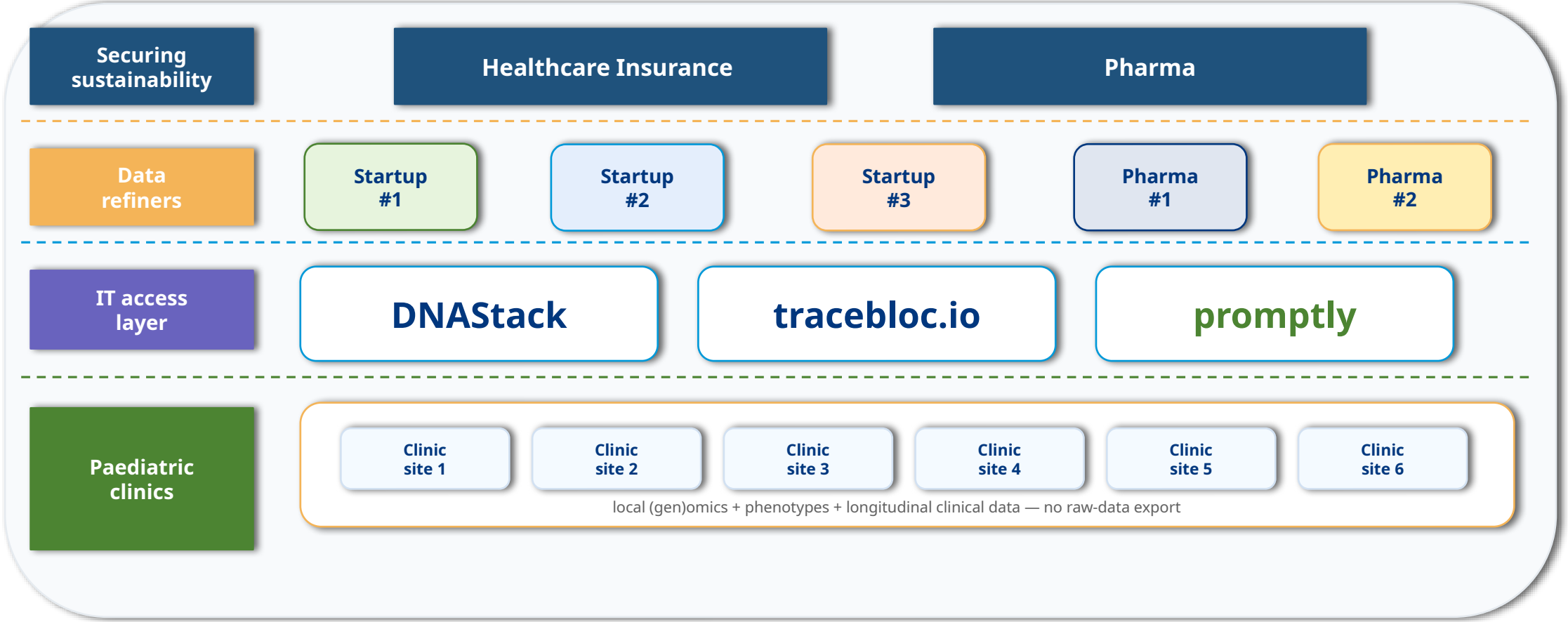
Pharmaceutical market analysis based on real-world biomarker distribution.

Discovery of novel drug targets through pathway and graph-based clustering.

Validation loops with wet-lab or organoid/iPSC models where feasible.

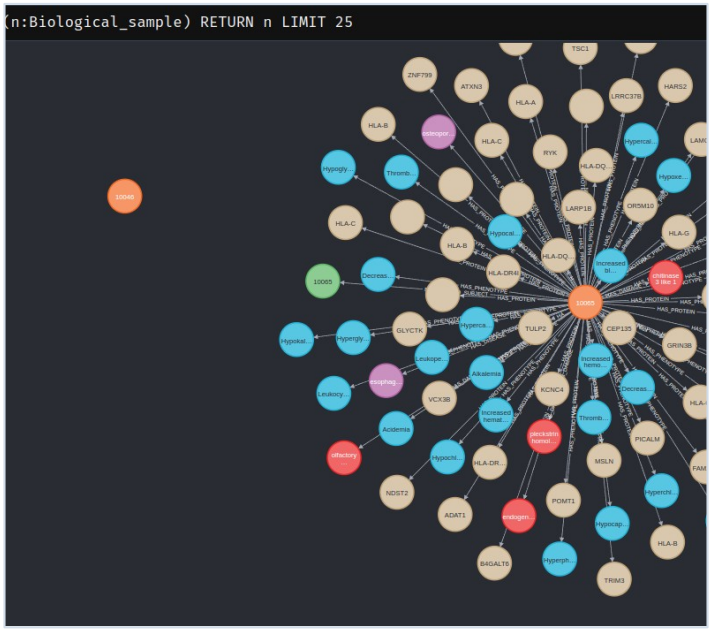
Sustainability: competition at every level

Self-financing paediatric precision medicine: Monday blood draw → Friday diagnosis support, using models trained federated across clinics



Core principle: do not sell children's data — enable governed decentralised access and reinvest the value into personalised medicine for children.

Proof of concept: privacy-safe development already demonstrated



AMIGO = Advanced Medical Intelligence for Guiding Omics-based Medicine

Thank you - AMIGO is built by people

Donated trust, clinical responsibility and volunteer expertise make paediatric personalised medicine possible.

Most importantly:

Children & families

Thank you for your trust and for contributing blood samples for research.

Every graph, model and therapy hypothesis starts with your willingness to help other children.

The foundation of AMIGO

Physicians & clinical teams

For seeing the child behind the data, recruiting responsibly and translating insights back into care.

Volunteers from industry & consulting

For donated expertise in architecture, governance, business models and implementation.

Students and hackathon teams

For creativity, prototypes, energy and the courage to test ideas on realistic data.

Thank you for turning fragmented data into integrated translational pathways - for children.