National Biobanks for Population & Rare Disease Research

Closing the Rare Disease Diagnostic Gap

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BIOBANK

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The Diagnostic Gap in Rare Diseases

- 27–36 million EU citizens live with a rare disease
- Many patients remain undiagnosed or experience 5–7 year delay in diagnostics
- Fragmented symptoms across specialties delay recognition
- Variants of Uncertain Significance complicate interpretation
- National and EU level Health system fragmentation increases cost and inequity

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What Can Help Us to Close the Gap

EHDS → Data access, interoperability & empowerment of Secondary use

National data lakes → Aggregated vast amounts of structured and unstructured health data

Biobanks → Secure genomic-phenotype integration

AI systems → Pattern recognition & decision support

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The Role of National Biobanks

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- Combine genomics + longitudinal phenotypes
- Offer governance frameworks and re-contact capability
- Act as trusted intermediaries within EHDS
- Enable genotype-first screening approaches

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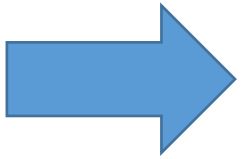
Secondary Use of Phenotypes as a System-Level Opportunity to Close the Gap

- Reuse of EHR, registry, imaging, and laboratory data
- Enables early detection of rare disease patterns
- Improves variant interpretation and patient stratification
- Condition(!)-Requires strong governance and interoperability



- Algorithmic detection of rare disease signatures
- Machine learning clustering of atypical presentations
- Automated triggers for earlier genomic testing
- Shortens diagnostic odyssey

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Example of the pilot study in progress: Early Alport syndrome detection screening in Lithuania

- Integration of HIS and national data
- Integration of genealogy (kinship) into screening algorithms
- Age-stratified AS screening and TBMD (Thin Basement Membrane Disease) monitoring as a predictor of potential disease
- A systematic validation cycle that includes data-driven vocabulary development, retrospective algorithm analysis, and clinical outcome evaluation

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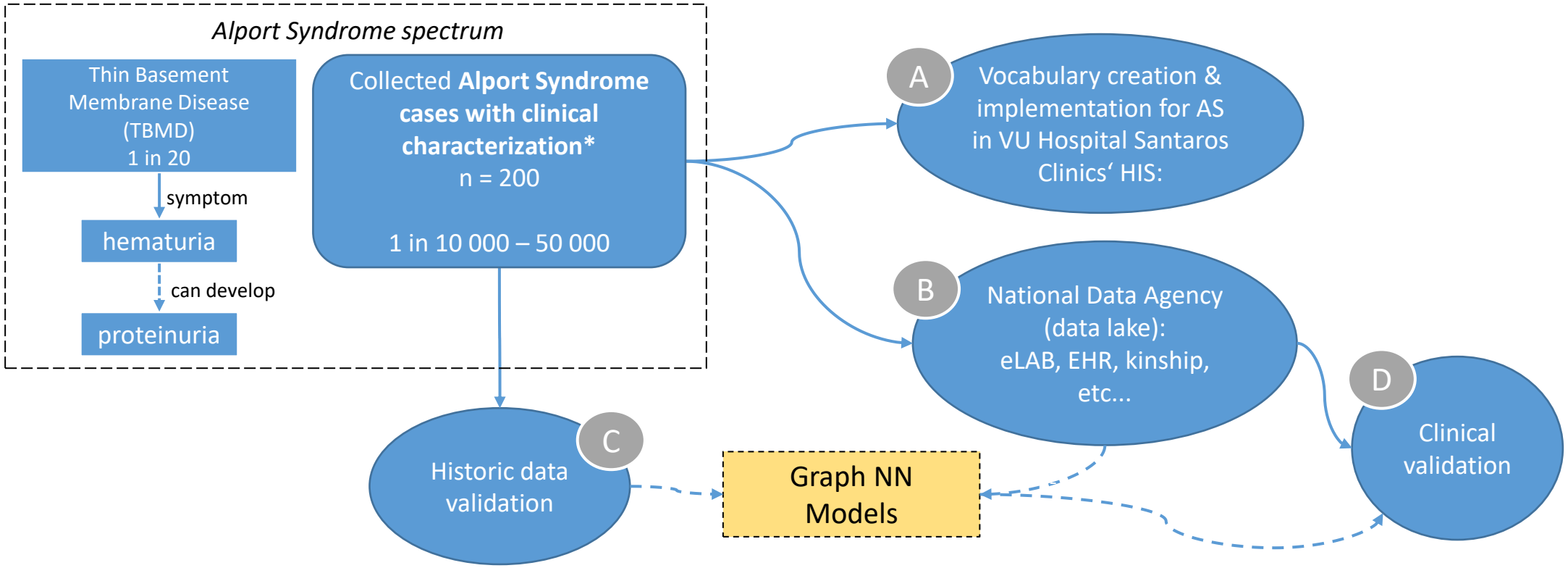
Structure & stages of the AS screening Pilot



*Cerkauskaite-Kerpauskiene, A., Navickaite, M., Savige, J., Mazur, G., Brazdziunaite, D., Azukaitis, K., Slazaite, G., Laurinavicius, A., Miglinas, M., Vainutiene, V., Strupaite-Sileikiene, R., Misevice, A., Mickeviciene, V., & Cerkauskiene, R. (2025). Lithuanian Study on COL4A3 and COL4A4 Genetic Variants in Alport Syndrome: Clinical Characterization of 52 Individuals from 38 Families. International journal of molecular sciences, 26(15), 7639. https://doi.org/10.3390/ijms26157639

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Reverse Phenotyping & Variant Interpretation

Can help to:

- ❖Match variants to deep phenotype data
- ❖Identify individuals with similar genotype + phenotype patterns
- ❖Improve classification of variants of uncertain significance
- ❖Enable family cascade screening

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Societal and Economic Value

- Earlier diagnosis reduces healthcare waste
- Improves equity across Member States
- Accelerates pharmaceutical innovation
- Strengthens EU competitiveness in precision medicine

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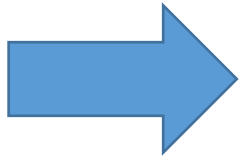
Means & Goals for Identifying Undiagnosed Patients in Health Systems

- Systematic screening for phenotype combinations
- Detection of hereditary patterns
- Proactive referral to genetics services



- Rare disease risk scores integrated into EHR
- Automated phenotype extraction (HPO mapping)
- Federated analysis across EU biobanks
- Embedded rare disease registries

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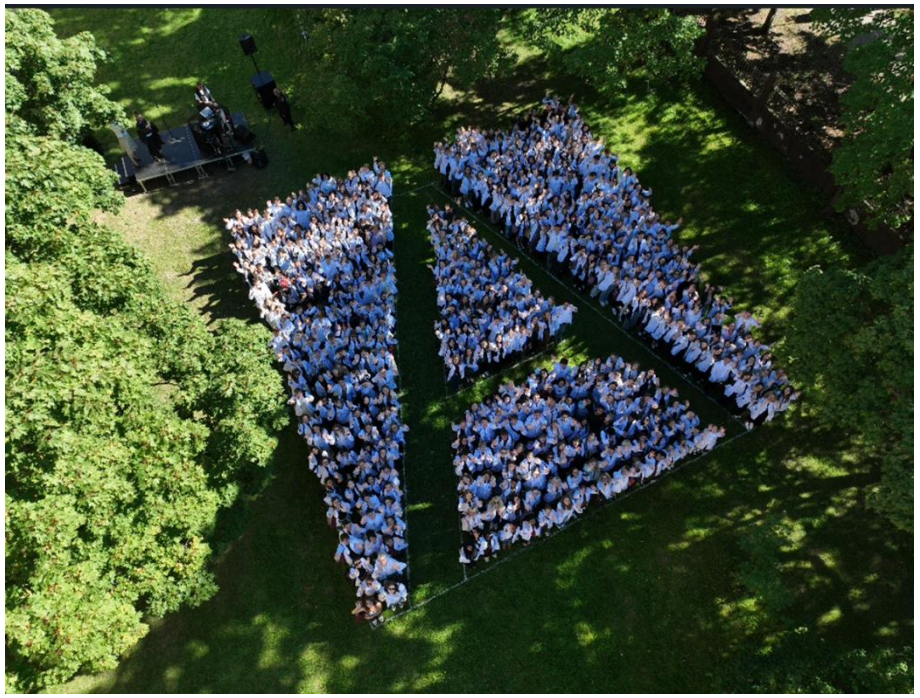
The Messages to Take-away

- Data governance + biobanks + AI = diagnostic acceleration
- Rare diseases can be a flagship EHDS use case
- Opportunity for coordinated EU leadership



Joint solidarity action by Lithuanian population & rare disorders biobank and VU medical students in 2025

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Thank you!

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