

Diagnosis of Rare Diseases via AI

*Supporting **Patients**, Doctors, Clinics with Diagnosis
to facilitate access to treatment or clinical trials*



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— Health —

Introduction to Saventic Health



Who we are

Med-tech combining **AI algorithms**, **implementation platforms** and processes to **support doctors and medical clinics in diagnosing exclusively rare diseases** (“hidden”, “difficult to spot” diseases)

Selected Clients (contracts / negotiations with most of RD pharmas)



Selected partners

Clinics, Patient advocacy groups, Startup accelerators, Universities, Chambers of commerce

Our Team



Szymon Piątkowski, CEO

- +15 years at PwC & EY with focus health consulting
- MA in finance & B.Sc. in computer engineering



Prof. Grzegorz Basak, MD, CMO

- +18 years Dep. of Hematology, Oncology & Internal Medicine
- Ph.D. in Molecular Medicine

Global Management Team



Szymon Piątkowski
CEO



Prof. Grzegorz Basak, MD
Medical Advisor



Maciek Klein
CBO



Karol Lis, MD
COO



Marek Dudziński, MD, PhD
CMO



PhD Michał Dąbrowski
CTO



Maciej Majewski
CIO

REST OF THE TEAM



Canada



Kim King
Hospital
Relationship
Manager /
Saventic Med
Platform Lead

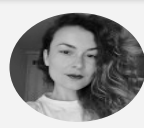


Courtney Bachus
Saventic Care
Platform
Leader

Brazil



Vanessa Koso
Hospital
Relationship
Manager /
Saventic Med
Platform Lead



Claudia Bar
Saventic Care
Platform
Leader

Latam (Spanish speakers)



Laura Restrepo
Hospital
Relationship
Manager /
Saventic Med
Platform Lead

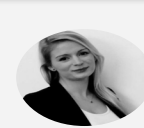


Camila Mejia
Saventic Care
Platform
Leader

Europe



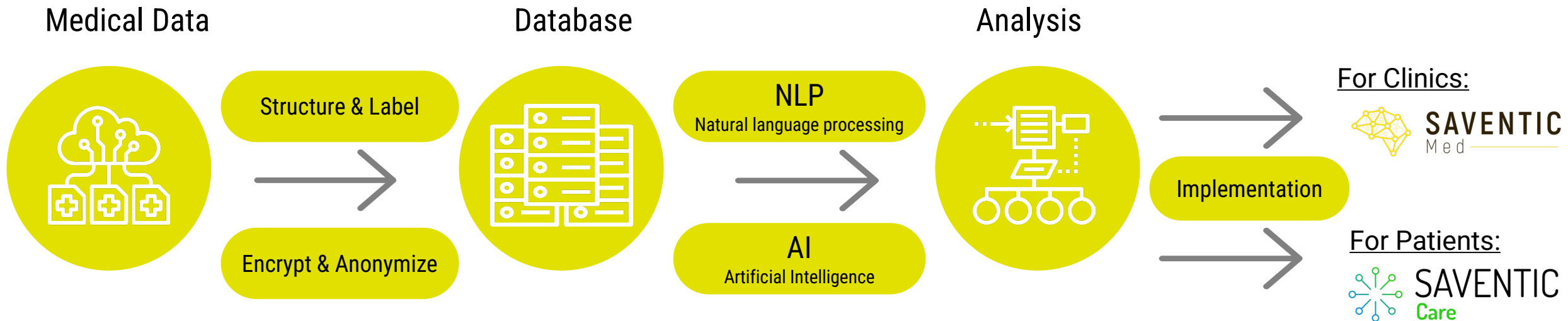
Kai Hamacher
Hospital
Relationship
Manager /
Saventic Med
Platform Lead



Aneta Matysiak
Saventic Care
Platform
Leader

Saventic Health Solution - AI-Tools:

AI algorithms, implementation platforms & innovative processes



30 Co-operating
top clinics

12M Patients in
our database

50 AI-driven
algorithms

2 Platforms

Saventic Health's Portfolio of AI algorithms



Rare Diseases

Blood and bone marrow

- Blood cancers (various including, mastocytosis and myelofibrosis)
- CTCL
- TTP
- PNH
- Castleman
- ITP
- HLH
- MDS (monitoring)
- aHUS
- Waldenström macroglobulinemia

Metabolic

- **Gaucher Disease**
- **Fabry Disease**
- Pompe Diseases
- **HAE**
- MPS 1
- **MPS 2 (Hunter)**
- MPS 3
- ASMD
- **AATD (to be developed)**

Immune system

- Common variable immunodeficiency (CVID)
- Severe combined immunodeficiency (SCID)
- DiGeorge syndrome
- Chronic granulomatous disease (CGD)

Other

- Amyloidosis AL
- Amyloidosis ATTR
- Spasticity
- Hypercholesterolemia
- ILD
- IPF
- C3G

in addition
Approx. 30 algorithms in our pipeline

Implementation

Input data (from patients' EHR)

- a) Age, Gender
- b) Laboratory test results
- c) Medical interviews
- d) Physical examination
- e) Radiology
- f) Many others (ICD-10, drugs, other diseases)

Medical interview example:

newly diagnosed macrocythic anemia ... kidney failure II stage (KIDO 2012) ... Bone pain in the lumbar region increasing for 6 months – requires taking painkillers everyday

Medical imaging example:

X-ray lumbar section: degenerative changes in L1-2, L4-S1, osteolytic changes in the vertebral bodies L2 and S3

Abdominal ultrasound: kidneys of normal size, without hepato- splenomegaly, without lymphadenopathy



Natural
language
processing



AI / ML
classifiers,
algorithms

Output data (feedback for doctors)

Rare disease Risk: Yes

ICD-10: E75.2 - Morbus Fabry

Risk Score: High (76%)

**Rationale: stroke 2× at young age,
unverified cardiomyopathy,
Cornea verticillata, ...**

**Proposed next steps:
DBS test at a reference clinic**

International presence and global expansion



Acute Care Alberta Project

“Rare Disease Diagnosis- Saventic Health Canada”

- Diagnosing rare diseases can take several years, imposing a heavy burden on patients, healthcare providers, and the healthcare system.
- Our project with Acute Care Alberta (AHS) Innovation and Business Intelligence Division and the MAGIC Clinic, a Calgary-based center specializing in rare disease care, is evaluating the use of precision analytics to enhance rare disease detection.
- By algorithmically screening patients based on their risk of having a rare disease, **we hope to empower clinicians to shorten the time to appropriate treatment and care, improve the efficiency of the health care system in Alberta, and improve the quality of life for patients in Alberta with rare diseases.**
- Will help achieve numerous goals of **Canada’s National Strategy for Drugs for Rare Diseases**
- ACA benefits of **modernization, sustainability, care improvement, leadership in adoption of innovation, and a learning health system.**

Acute Care Alberta Project

- 3 major phases

- **Phase 1a (September 2024- complete):** data prep, training, and testing/training Fabry algo with MAGIC Clinic, KOL care pathway/referral workshops with industry partners
- **Phase 1b (~February 2025):** to be completed in two parts, with continual feedback to improve algo and evaluate efficacy. Publications, conferences and economic analysis
- **Phase 2 (~December 2025):** monthly screening of ConnectCare data for 9 months

After each phase: report on algorithms specificity and sensitivity,
and economic analysis

Complete: Signed project synopsis, budget, timelines, 1a ethics approval, technology initial assessment, Phase 1a complete, invitation of industry partners for 1b, phase 1b.1 ethics approval

Underway: Phase 1b data prep, hospital selection, engagement of industry partners, new algo development, KOL engagement

Phase 1a: Fabry retrospective, small population



Summary:

Phase 1a is a focused study on a small population from the MAGIC clinic to check a single rare disease algorithm (Fabry Disease). To evaluate the performance of Saventic Health's Fabry disease screening algorithm on a small retrospective population. September 2024.

Overview:

The algorithm consists of a rule-based classification model, which sorts patients into high-risk or low/no-risk categories. Then, a scoring model ranks the patients from highest to lowest risk.

A retrospective analysis was performed on anonymized clinical data from 30 patients diagnosed with Fabry disease and 15 patients with other lysosomal storage disorders (control group). The data used included descriptive physician notes from patient visits, as well as diagnostic imaging reports. The data was pre-processed to remove Fabry diagnosis.

The classification model accurately identified 29 out of 30 Fabry patients.

Approval to proceed with Phase 1b

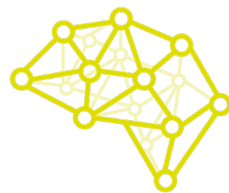
Phase 1b: Next Steps



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1. MAGIC Clinic/Acute Care Alberta/Saventic finalized study design and secured HREBA
2. Amend the project agreement to include Co-Principal Investigators for Phase 1b, including addition of new rare disease targets.
3. Secure data export from Connect Care & analysis environment to run Saventic algorithms.
 - Part 1b.1:
 - Fine-tune algorithms, implement the platform with algorithms on ConnectCare data, retrospectively run the algorithm using fully anonymized Acute Care Alberta (AHS) medical data to validate its specificity and sensitivity
 - economic analysis
 - HREBA approved
 - Part 1b.2:
 - Run the algorithm using ConnectCare data from ~500k patients at ~4 hospitals to flag, review, and appropriately contact patients for further consultation
 - Algo run monthly, for 9 months
 - Will involve an economic analysis
 - This involves a more detailed ethics and Health System Access review due to in-system notifications and patient contact
 - Ethics Chair is discussing this phase with the board and will provide a recommendation for best practice

AHS Rare Disease AI Partnership	Algorithm Development and Confirmation of Diagnosis Pathway
Company	Targets
Pfizer	ATTR CM amyloidosis- wild and hereditary
Novartis	C3G
Boehringer Ingelheim	Interstitial lung disease



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