Identifying Gaucher disease in patients by modelling disease-specific phenotypes using EHR data at Istanbul University: A Single-Center Study in Turkey

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BACKGROUND

Rare diseases are often undiagnosed or misdiagnosed, and patients do not have access to the right treatment. Lysosomal storage diseases (LSD) are characterized by the accumulation of specific metabolites in lysosomes and include more than 50 inherited rare diseases. Gaucher disease is an autosomal recessive LSD that results from mutations in GBA1 gene, which leads to a deficiency in lysosomal acid b-glucosidase function and is associated with damage to multiple organ systems. In this study, which was approved by the ethics committee with a single-center, retrospective research design, it was aimed to identify the patients by modeling the phenotypes specific to Gaucher disease among the patients diagnosed with the ICD10 code "E75.2" in the electronic health records (EHR) of Istanbul Faculty of Medicine Hospital.

METHODS

Using Patient Network Explorer, a patented technology of Clinerion; demographics, diagnosis, medications, procedures, and laboratory tests, relevant health data were compiled and analyzed simultaneously in an anonymous and aggregated way. Clinerion's automated transfer tool for electronic data capture integration from EHR strongly supports each patient's EHR data with retrospective information in e-CRF. In the analysis process, patient personal data is excluded, by using Clinerion ANID server, which provides a fully anonymized record. Data collected are evaluated in terms of validity, accuracy, consistency, integrity and completeness. Keeping duplicate records for the same patient is also prevented.

RESULTS

A total of 3,420,751 unique patient data were screened between 2007 and 2019 in the EHR, which is used as RWD source. 128 female and 117 male patients, their current age and gender, age at diagnosis date, incidence, comorbidities, medications, procedures, and lab tests were analyzed.

CONCLUSIONS

With this approach to the diagnosis of Gaucher patients, significant time and cost savings are achieved through streamlined operations and strategic intelligence to treat more patients.

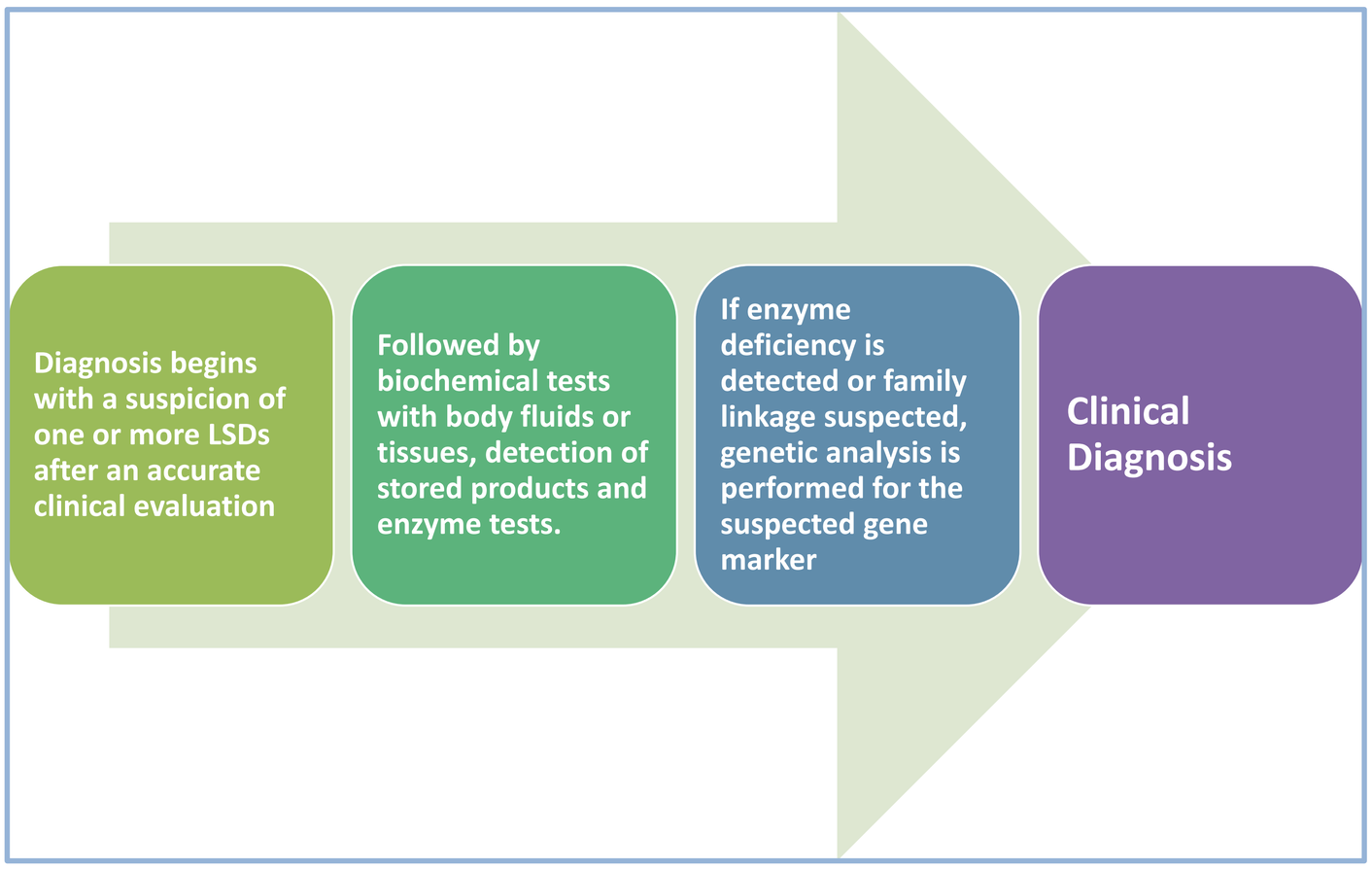

Figure-1 Clinical diagnosis of LSD

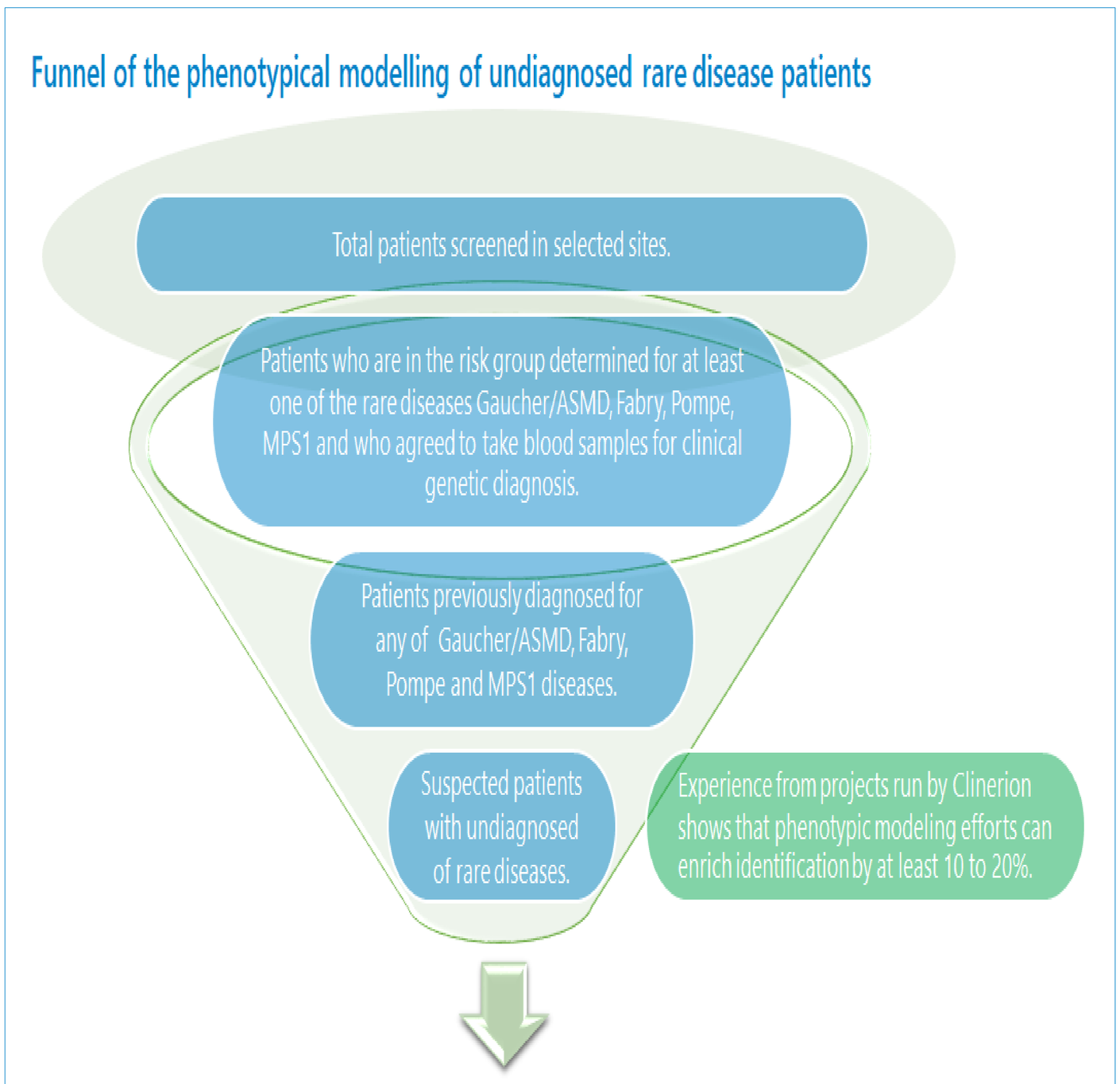

Figure-2 Illustrative screening and phenotypic symptom screening funnel to highlight possible under diagnosed rare disease LSD patients, based on learnable criteria and disease models.

CURRENT STATUS AND NEXT STEPS

- This demonstrates the use of EHR data for rare disease classification as well as potential identification of suspected patients. This white paper outlines a methodology for the use of Patient Network Explorer in identifying patients for earlier diagnostic testing with a wider reach and enhanced clinical trial cohort selection
- Through collaborations with us, sponsors who enable their clinically validated patient models for our queries will be able to perform targeted outreach for the specific genetic diagnostic test needed for corrective diagnosis. In addition, the Clinerion approach also provides significant time and cost savings from streamlined operations and increased strategic intelligence.
- In the next step of our research into AI/ML model training, our models will be clinically validated by sponsor-partners, with follow-up diagnostic testing being done in a validated and approved fashion.

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