

Hidden in Plain Sight: Identification of Genomic Information within EHR Allergy Documentation

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Background

Pharmacogenomics (PGx) plays an important role in precision medicine by using genetic variability to guide medication selection, dosing, and safety. Despite its clinical value, widespread implementation remains limited, largely due to how genomic data are captured within electronic health records (EHRs). In many healthcare systems, genomic and pharmacogenomic information is not stored in structured fields but instead appears within free-text allergy or adverse reaction documentation. This fragmented approach limits the ability of clinicians and clinical decision support (CDS) tools to identify and act on clinically relevant genetic information at the point of care.

Objective: To evaluate the presence of genomic and pharmacogenomic information within EHR allergy documentation and assess how this information is represented, structured, and potentially utilized in clinical practice.

Methods

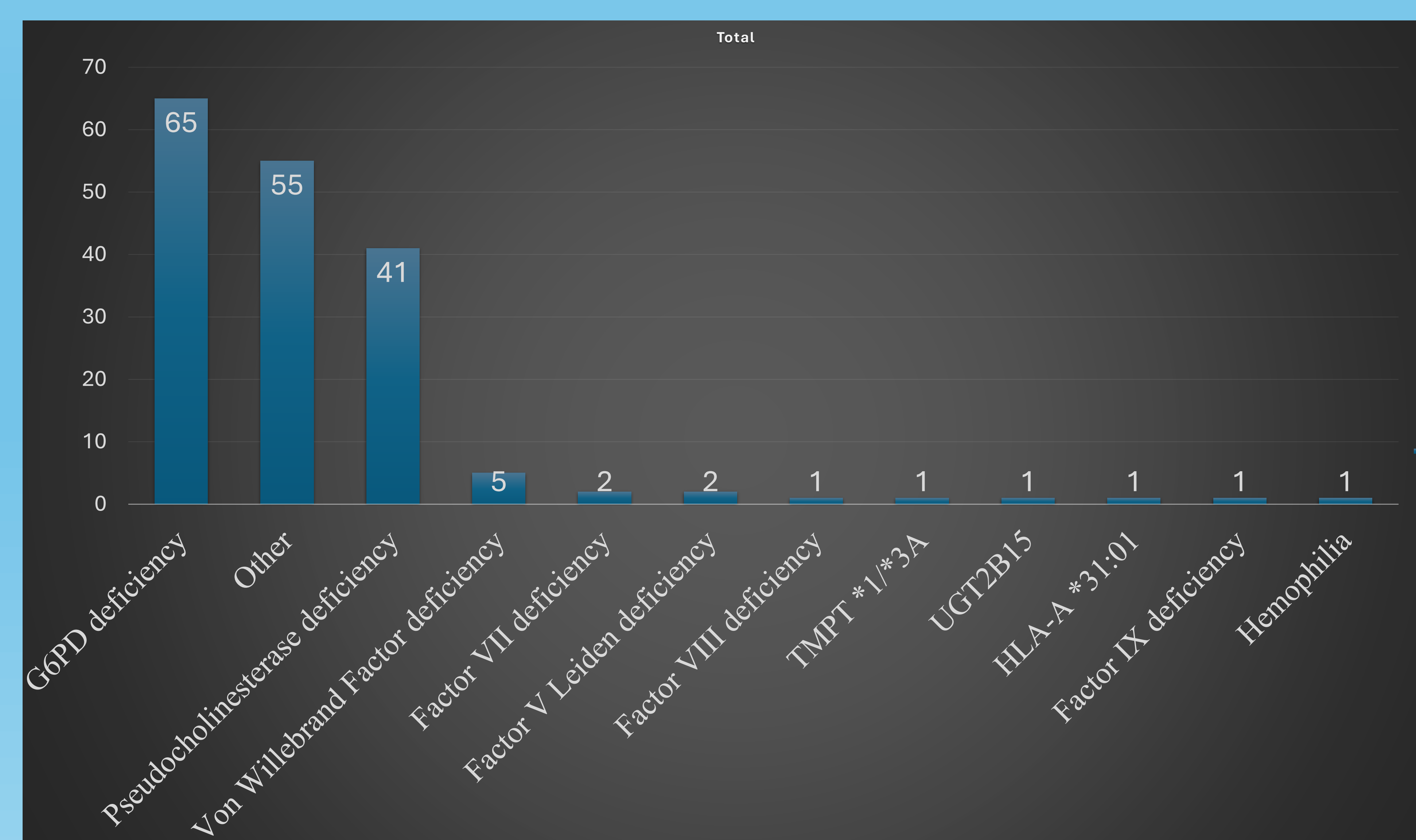
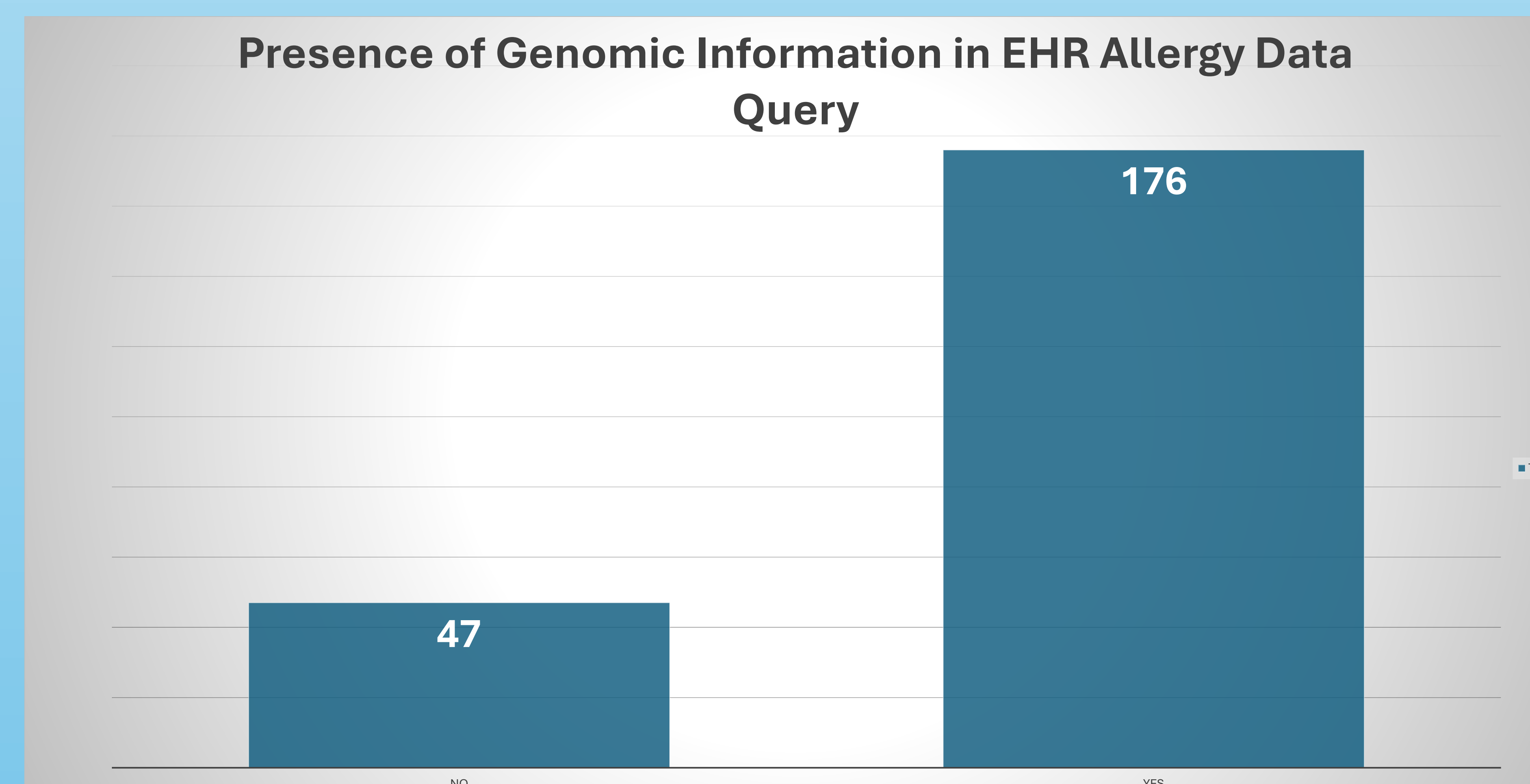
- Retrospective analysis of 223 EHR allergy and adverse reaction records
- Data fields included medication (DESCRIPTION), free-text reaction details, severity, and patient demographics (race, sex, birth date)
- Genomic indicators were identified using a keyword-based search of free-text reaction fields
- Identified records were categorized by genomic condition type (e.g., G6PD deficiency, pseudocholinesterase deficiency, CYP-related metabolism)
- Descriptive statistical analysis was performed in Excel to evaluate frequency, distribution, and associations

Results

- 78.9% (176/223) of records contained genomic or pharmacogenomic indicators
- Most common G6PD deficiency (36.9%)
- Pseudocholinesterase deficiency (23.3%)
- Genomic indicators were strongly associated with clinically relevant medications:
 - Succinylcholine → pseudocholinesterase deficiency
 - Sulfa antibiotics → G6PD deficiency
 - Opioids → altered CYP-mediated metabolism
- Demographic variation was observed:
 - G6PD deficiency was highly prevalent among Black or African American patients
 - Greater variability in genomic documentation was observed among White or Caucasian patients

Limitations

- **Keyword-based identification:** Genomic indicators were identified using a keyword search of free-text fields, which may have led to missed cases or misclassification due to inconsistent wording.
- **Unstructured data:** Reliance on free-text reaction descriptions introduces variability and subjectivity, making it difficult to distinguish confirmed genetic conditions from suspected or patient-reported information.
- **Single dataset:** Data were derived from one EHR dataset, limiting generalizability to other institutions with different documentation practices or patient populations.
- **Lack of validation:** The analysis does not confirm whether genomic indicators were based on formal pharmacogenomic testing or clinical inference.
- **Documentation variability:** Differences in how providers record information may influence findings and may not reflect true biological differences across patients.



Conclusion

Genomic and pharmacogenomic information is frequently documented within the EHR but remains underutilized due to its unstructured and fragmented representation. Establishing a dedicated genomic informatics section within the EHR could improve data standardization, accessibility, and integration into clinical decision support. This would enhance medication safety, support precision medicine, and optimize patient care outcomes.