Standardizing Return of Pharmacogenomic Results into EHR Systems

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BACKGROUND

Clinicians require clear and actionable pharmacogenomics (PGx) results for efficient, effective patient care. Challenges persist:

- Reporting for genes with multiple alleles like HLA-B can be inconsistent or lack resolution.
- Reports vary between labs with content delivered as a PDF or VCF file instead of structured data[1].
- PGx test results differ from other types of genetic tests and require special consideration to decrease ADR risk[2].
- Gaps remain despite efforts by CPIC, LOINC, ACMG, AMP, and HL7/FHIR Working Groups. While CPIC provides guidelines for individual HLA-B alleles[3], the terminology used in the ACMG guidelines for reporting variants differs from PGx community norms[4].
- The HL7 Genomics Reporting Implementation Guide offers PGx reporting guidance but lacks detail on specific genes, making it difficult to standardize data structure.

A more detailed exploration of data transmission methods is essential for enhancing clarity and standardization.

METHODS

We performed a retrospective analysis on 12,680 patients tested for HLA-B alleles using the OneOme RightMed® Test. This study evaluated carrier frequencies to assess the potential impact of enhanced data transmission.

We also reviewed lessons learned and input from integrated health systems throughout 2023-24.

REFERENCES

1. Carter AB, Abruzzo LV, Hirschhorn JW, *et al., J Mol Diagn.* 2022 Jan;24(1):1-17.
2. Gammal RS, Berenbrok LA, Empey PE, Massart MB., *J Pers Med.* 2021 Dec;11(12):1296.
3. Clinical Pharmacogenetics Implementation Consortium (CPIC). HLA-A and HLA-B CPIC guidelines available at: https://cpicpgx.org/gene/hla/
4. Richards S, Aziz N, Bale S, *et al., Genet Med.*2015 May;17(5):405-24.
5. Health Level Seven International (HL7). Genomics Reporting Implementation Guide available at: https://hl7.org/fhir/uv/genomics-reporting/pharmacogenomics.html
6. Dexter P, Ong H, Elsey A, *et al. AMIA Ann Symp Proc.* 2019;2019:363-70.

Lack of standardized, high-resolution data transfer from laboratories into EHR systems may impact patient safety, efficiency and PGx utility.

RESULTS

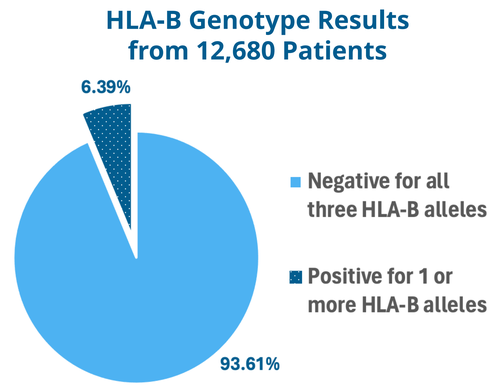
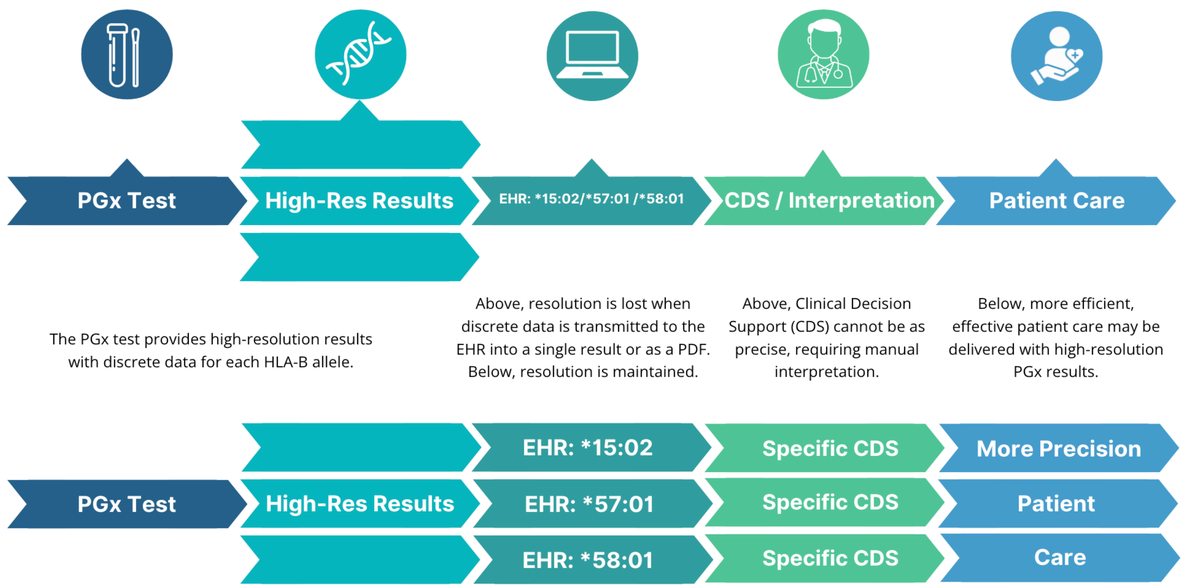


FIGURE 1. Final Genotyping Outcomes for HLA-B Alleles **15:02, *57:01, and *58:01*. This figure highlights that 6.39% of the patient cohort tested positive for at least one of these alleles.

HLA-B Genotype	# of Patients	% of Total Sample
*57:01 Positive	613	4.84%
*58:01 Positive	179	1.41%
*15:02 Positive	13	0.1%
*57:01 + *58:01 Positive	5	0.04%
Total Positive	**810**	**6.39%**

TABLE 1. Detailed Breakdown of Positive HLA-B Allele Results Presented in Figure 1. Patients who tested positive for specific alleles may be at an increased risk for severe ADRs or drug hypersensitivity[3].

WORKFLOW EXAMPLE: OLD (TOP) VS. NEW (BOTTOM)



The PGx test provides high-resolution results with discrete data for each HLA-B allele.

Above, resolution is lost when discrete data is transmitted to the EHR into a single result or as a PDF. Below, resolution is maintained.

Above, Clinical Decision Support (CDS) cannot be as precise, requiring manual interpretation.

Below, more efficient, effective patient care may be delivered with high-resolution PGx results.

LESSONS LEARNED

Non-standardized guidance for transmitting structured, high-resolution for HLA-B contributes to several issues:

(1) Access and Usability:

- Data from PDFs cannot be used outside of the context of their interpretation – they also require data manipulation or manual entry.

(2) Clinical Implication and Risk :

- Unclear reporting or combined genetic data can lead to suboptimal patient care.
- Data cannot easily be used to create precise clinical decision support (CDS) alerts, including CPIC recommendations for HLA-B.

(3) Data Sharing:

- HLA-B data combined into a single result cannot be seamlessly shared with other institutions.
- Lack of high-resolution data hinders large-scale research, such as the eMERGE Genomic Risk Assessment and Management study.[6]

CONCLUSION

Consistent, high-resolution PGx data into the EHR is essential, as highlighted by the prevalence of patients with at least one HLA-B allele and the potential issues that arise.

By updating data formatting to return each HLA-B allele as a separate data point, we aim to standardize the return of PGx data into EHR systems and enhance the use of PGx data to improve patient outcomes.

AFFILIATION

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