Pharmacogenetic Testing to Identify Presence of the HLA-B*57:01 Biomarker for Abacavir Treatment

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INTRODUCTION
Testing for the HLA-B*57:01 biomarker is clinically significant to predict the risk of potentially life-threatening hypersensitivity reactions in patients to be treated with the antiretroviral drug abacavir.\(^1\)
Screening for HLA-B*57:01 is required prior to initiating abacavir therapy and alternative therapy should be prescribed in patients who test positive for the biomarker.\(^1,2\)

METHOD
1. HLA-B*57:01 genotyping methods were identified from the Clinical Pharmacogenetics Implementation Consortium guidelines supplementary information.\(^3\)
2. A literature search was performed to identify studies describing genotyping for HLA-B*57:01 with respect to abacavir.
3. The genotyping methods identified were reviewed and compared according to twelve genotyping method characteristics.

RESULTS
- Fourteen articles (2005-17) describing HLA-B*57:01 genotyping were reviewed.
- Eight genotyping methods were identified: 7 molecular (Sequence-based typing (SBT), Sequence Specific Primer PCR (SSP-PCR), Sequence Specific Probes (SSOP), Allele-Specific PCR, Real-time PCR, Direct PCR, Reverse hybridisation strip assays) and 1 serological (Flow Cytometry).
- The 8 methods have high sensitivity (>99%) and specificity (>97%) and 100% concordance with reference methods. For all 8 methods single-sample genotyping is not possible.
- Four methods allow use of non-invasive genomic DNA sources, 3 methods have integrated controls, turnaround time ranges from 2 to 24 hours and estimated cost/test ranges from £1-35.

AIM
To review and compare genotyping methods for the identification of the HLA-B*57:01 pharmacogenetic biomarker in relation to abacavir treatment.

CONCLUSION
The genotyping methods reviewed show high sensitivity, specificity and concordance. Possibility of non-invasive testing, availability of integrated controls and on-demand testing are limited. The review highlights the need for development of rapid point-of-care HLA-B*57:01 genotyping methods to promote use of pharmacogenetic testing in practice to personalise abacavir therapy in the interest of patient safety.

REFERENCES