

HEMO ID – A ROBUST AND FLEXIBLE PANEL FOR BLOOD GROUP GENOTYPING, AND PREDICTED GENOTYPING

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Background:

Immunohematology labs have access to a range of systems for blood group genotyping. Each system offers distinct benefits such as the range of blood groups interrogated, precision, robustness, throughput and cost. Here we describe the performance characteristics of a newly available research-use-only blood group genotyping panel called Hemo ID.

The Hemo ID panel consists of six independent modules each containing a distinct set of assays which are used to interrogate 108 single nucleotide variations and indels in the human genome. The six modules consist of: (i) Kell, Kidd, Duffy, (ii) MNS, (iii) Rare Blood Groups, (iv) RhD/CE Broad, (v) RhD Variant and (vi) HPA & HNA. The modular setup provides a flexible high-throughput solution that can easily adapt to newly discovered blood groups. In this work we assess the performance of the Hemo ID Panel regarding its reproducibility and its flexibility with respect to different sources of DNA, customizability of panel content and reporting software.

Methods:

For Hemo ID analysis, human DNAs derived from buccal swabs, blood and cell lines were amplified by multiplex PCR followed by single base extension and analysis by MALDI-TOF mass spectrometry. Hemo ID software was used to assign genotypes, predict phenotypes and generate reports. For the reproducibility study this process was repeated at three separate sites using the same set of 47 HapMap DNA samples.

Results:

Feasibility study: We observed >99% call rate and 100% concordance of genotypes and predicted phenotypes across all six modules and independent of the different DNA sources. The entire study from buccal swab sampling through DNA isolation, Hemo ID biochemistry, data interpretation, and report generation was completed within two days.

Reproducibility study: Across the three sites the observed call rates were 99.8%, 98.0%, and 99.8%. The genotype concordance was above 99%. All sites correctly identified the major and minor alleles of the interrogated blood group, including the correct identification of Rhesus negative samples, one DAU sample, two Psi variant samples, three SS samples, and one sample each that was Fy(a-b-), Js(a+b+), Co(a+b+), Yt(a+b+), and heterozygous VEL negative.

Conclusion:

The Hemo ID panel is a robust solution for molecular blood group genotyping. It offers high flexibility regarding DNA sampling, sample through-put, choice of blood groups interrogated, and demonstrates both excellent reproducibility between analyses and a standardized analysis and reporting software that can easily be adapted to extended, customized modules.