# Cross platform comparison of HLA typing from whole-genome sequencing

C. Cremin[1],J. Quilez[1],A. El-Khani[1], A. Saad[1], A. Attia[1], C. Minas[1], F. Aldhuhoori[1], G. Katagi[1], H. Wu[1], H. Sidahmed[1], J. Mafofo[1], O. Soliman[1], S. Elavalli[1], S. Behl[1], T. Cardoso[1], F. Sedlazeck[2], L. Paulin[2],T R. Magalhaes[1];
[1]M42, Abu Dhabi, United Arab Emirates,
[2]Baylor Coll. Med., Houston, TX

## EXPERIMENTAL DESIGN

1. Two sample datasets were processed as per figure 1.
    a. The Coriell dataset is a 24 publically available cell line sample set from the Coriell Institute for Medical Research.
    b. An internal dataset derived from the blood, buccal and saliva swabs.
2. HLA typing was done on the BAMs from each processed sample.
3. Output was then compared to determine HLA typing robustness.

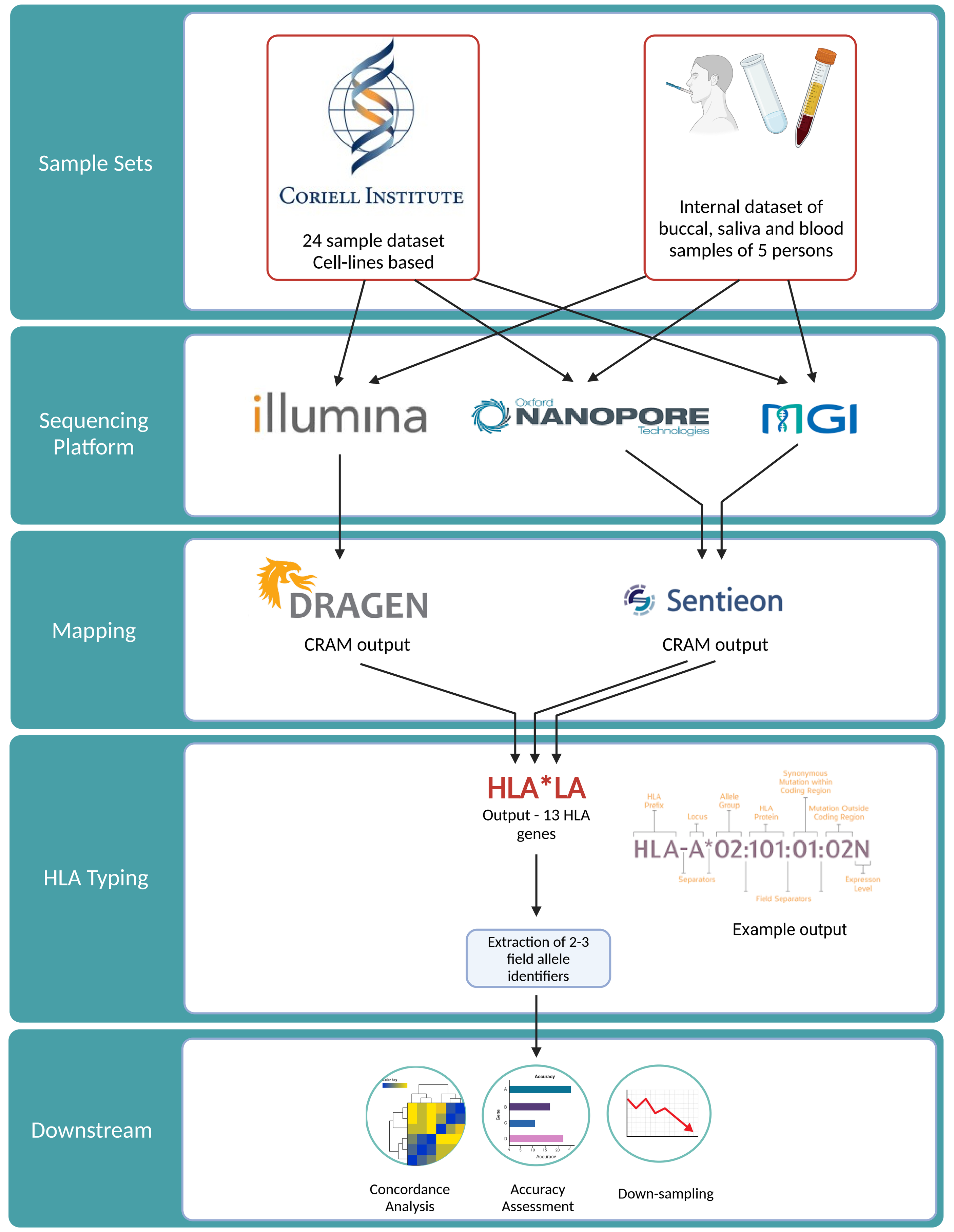


Figure 1: Schematic of Cross-Platform HLA typing methodology

**Outstanding HLA recall accuracy** across all sample types and sequencing platforms.

**Seamless WGS integration** proves HLA typing reliability.

**Huge potential for large-scale HLA studies** across populations.

**Paving the way for national HLA registries** and precision medicine.

## Concordance across sample types

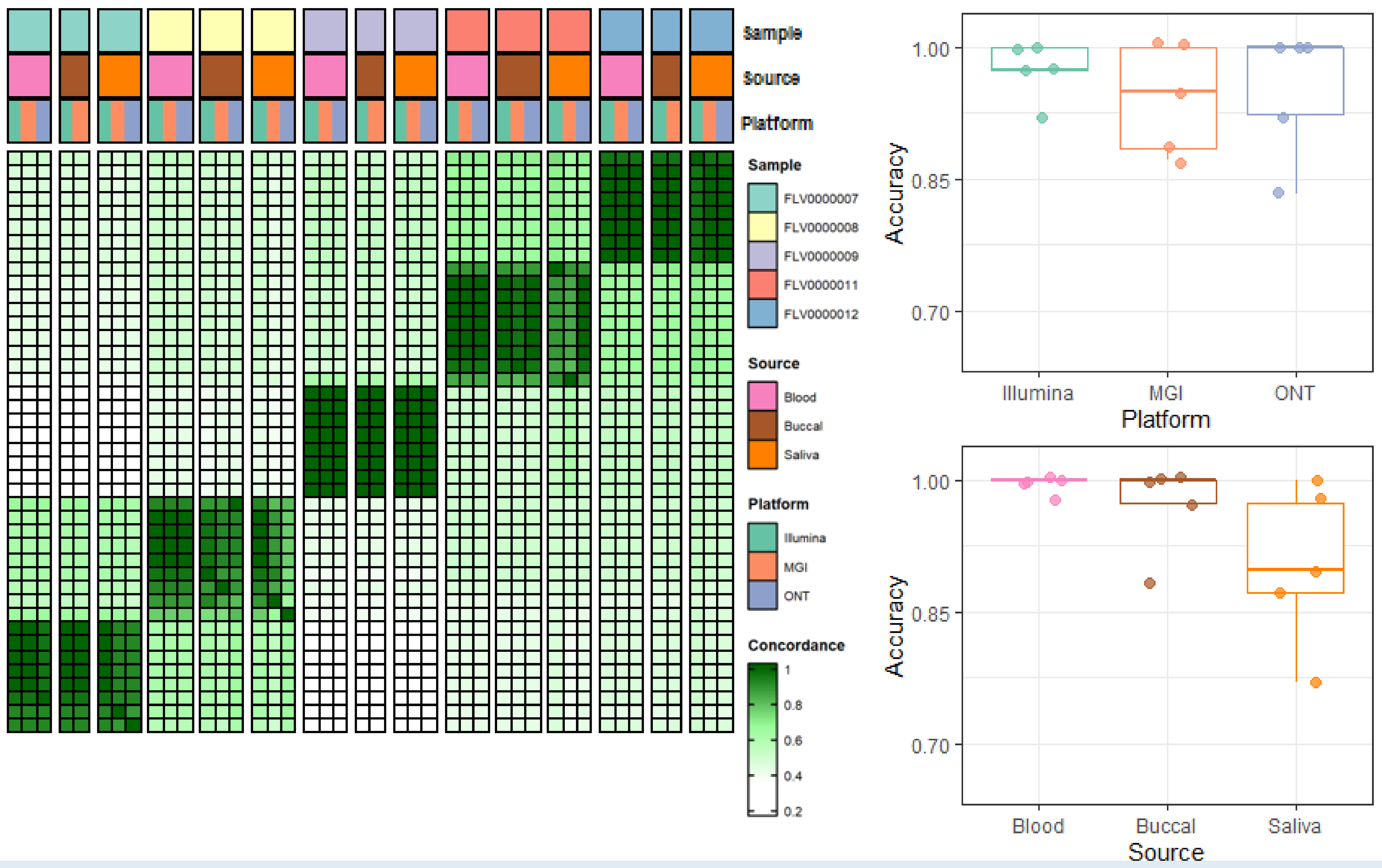


Figure 3: Cross sample dataset: (A) HLA concordance across WGS platforms and sample types. Accuracy distributions; (B) per platform and (C) per sample type.

## RESULTS

### Coriell Dataset

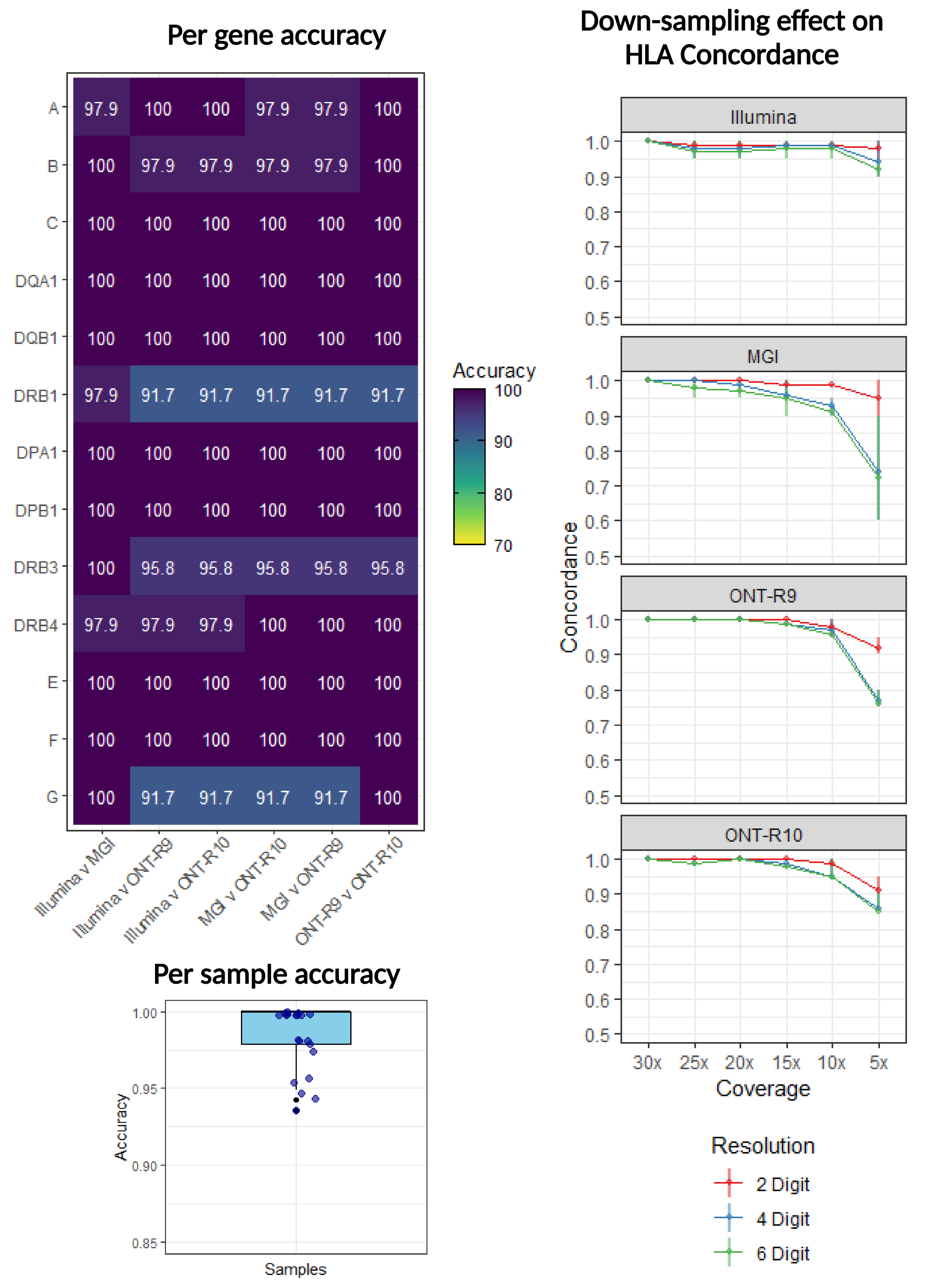


Figure 2: HLA concordance results showing the HLA recall accuracy across each sample and across each HLA gene. Down-sampling effect on HLA allele recall was shown to be preserved to the 15x threshold

### ACKNOWLEDGEMENTS

We followed all ethical guidelines and declare no conflicts of interest. Special thanks to our colleagues at M42 Health, Abu Dhabi, UAE, and Baylor College of Medicine, Texas, USA for their invaluable support.