



# **IMGT/HLA 3.29.0 Reference File Release Notes**

**For use with  
TruSight<sup>®</sup> HLA Assign<sup>™</sup> Analysis Software**

**January 2018**

For Research Use Only. Not for use in diagnostic procedures.

## IMGT/HLA 3.29.0 REFERENCE UPDATE

All alleles were validated for consistency with the IMGT/HLA release database version included in the IMGT/HLA release notes, which can be found with this link [www.ebi.ac.uk/ipd/imgt/hla/docs/release.html](http://www.ebi.ac.uk/ipd/imgt/hla/docs/release.html) except for the following alleles:

- DRB1\*14:152N—Modified to include intron sequence.
- DRB4\*01:03:01:02N—Modified to be consistent with the scientific literature<sup>1</sup>
- DRB4\*03:01N—Removed due to lack of exon 2 reference sequence in the IMGT/HLA database

## REGIONS INCLUDED IN THE CORE EXON ANALYSIS

- Core regions are determined to have the highest impact on biological function.

Locus	Region
A	Exon 2, 3, 4 5'UTR, position 200 Intron 1, position 478 Intron 2, positions 774-778, 1008 Intron 4, position 2167
B	Exon 1, 2, 3, 4, 5 5'UTR, position 133 Intron 1, position 461 Intron 4, position 2232
C	Exon 1, 2, 3, 4, 7
DPA1	Exon 2, 3
DPB1	Exon 2, 3
DQA1	Exon 2, 3
DQB1	Exon 2, 3
DRB1, DRB3, DRB5	Exon 2, 3
DRB4	Exon 2, 3 Intron 1, position 9977

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<sup>1</sup> Immunogenetics 31: 112-117, 1990

## BASE POSITIONS MASKED FOR VARIANT CALLING

Locus	Region
A	None
B	None
C	Intron 6, position 2957
DPA1	None
DPB1	Intron 2, positions 9094-9150, 9154-9156, 9175-9240
DQA1	5'UTR position 378-422 Intron 1 position 2010-2213, 3943-3946, 4199-4207 Intron 3 position 5722-5729 3'UTR position 6504-6510
DQB1	5'UTR position 254-258, 350-352 Intron 1 position 852-888 Intron 2 position 3190-3227, 3801-3852, 5020-5022
DQB1G06	Intron 1 position 867-871 Intron 2 position 4589-4591, 4489-4491, 4629-4631 Exon 3 position 5020-5022
DRB1G01	Intron 2 position 6333-6409
DRB1G03	Intron 1 position 8231-8252 Intron 2 position 9063-9128
DRB1G04	Intron 2 position 9290-9349
DRB1G07	Intron 2 position 11341-11388
DRB3	Intron 2 position 8451-8531
DRB4	None
DRB5	Intron 2 position 8710-8758

The masked regions that were added at position 3 of the 3' UTR in A and positions 210-218 of intron 2 of B in release 3.27.0 have been removed in release 3.29.0, in order to restore compatibility with projects that were saved using earlier references.

### 3.29.0 CWD LIST UPDATE

All alleles in the CWD list supplied by the Immunogenetics Data Analysis Working Group, which have identical entries in the relevant IMGT database release, are included in the TruSight HLA Assign CWD list. In addition, when CWD alleles have been split into subgroups after the most recent CWD list, all alleles in the subgroups are included in the TruSight HLA Assign CWD list.

### UNRESOLVED AMBIGUITIES

Please note that the alleles DQA1\*01:02:02 and DQA1\*01:16N cannot be resolved in the presence of a second allele with a deletion at position 153 in Exon 2.



## REFERENCE COMPATIBILITY

The IMGT HLA Reference 3.29.0 is compatible and validated with TruSight HLA Assign v2.1.

## REFERENCE UPDATE INSTRUCTIONS



### NOTE

In a default installation, the reference library files and the CWD, P, and G group files are installed.

1. In the System group, select **Update**.
2. In the Update window, under Import, select **References**.
3. Browse to the IMGT HLA Reference folder from Illumina. In the bottom right of the dialogue window, 'Reference Files' is indicated. Select all the files in the folder and select **Open**.  
The dialogue window indicates files successfully copied.
4. In the System group, select **Update**.
5. In the Update window, under Import, select **References**.
6. Browse to the IMGT HLA Reference folder from Illumina. In the bottom right of the dialogue window, select **P&G Group File**. Select all the files in the folder and select **Open**.  
The dialogue window indicates files successfully copied.
7. In the System group, select **Update**.
8. In the Update window, under Import, select **CWD Update**.
9. Browse to the IMGT HLA Reference folder from Illumina. In the bottom right of the dialogue window, 'CWD File' is indicated. Select all the files in the folder and select **Open**. The dialogue window indicates files successfully copied.
10. Select 'CWD 2012 v3.29 cat 1' or 'CWD 2012 v3.29 cat 1-2' as needed from the 'CWD Set' drop down list on the 'Annotation' panel.
11. Click 'Update' on the 'Settings' panel and choose 'Yes' to confirm the changes.

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