Benchmarking the HLA typing performance of Polysolver and Optitype in 50 Danish parental trios - DTU Orbit (23/01/2019)

Benchmarking the HLA typing performance of Polysolver and Optitype in 50 Danish parental trios

Background: The adaptive immune response intrinsically depends on hypervariable human leukocyte antigen (HLA) genes. Concomitantly, correct HLA phenotyping is crucial for successful donor-patient matching in organ transplantation. The cost and technical limitations of current laboratory techniques, together with advances in next-generation sequencing (NGS) methodologies, have increased the need for precise computational typing methods. Results: We tested two widespread HLA typing methods using high quality full genome sequencing data from 150 individuals in 50 family trios from the Genome Denmark project. First, we computed descendant accuracies assessing the agreement in the inheritance of alleles from parents to offspring. Second, we compared the locus-specific homozygosity rates as well as the allele frequencies; and we compared those to the observed values in related populations. We provide guidelines for testing the accuracy of HLA typing methods by comparing family information, which is independent of the availability of curated alleles. Conclusions: Although current computational methods for HLA typing generally provide satisfactory results, our benchmark – using data with ultra-high sequencing depth – demonstrates the incompleteness of current reference databases, and highlights the importance of providing genomic databases addressing current sequencing standards, a problem yet to be resolved before benefiting fully from personalised medicine approaches HLA phenotyping is essential

General information
State: Published
Organisations: Department of Bio and Health Informatics, Metagenomics, Integrative Systems Biology, Genomic Epidemiology, Disease Intelligence and Molecular Evolution, Technical University of Denmark, Aarhus University, BGI-Shenzhen, University of Copenhagen, South China University of Technology, BGI Europe A/S
Number of pages: 12
Publication date: 2018
Peer-reviewed: Yes

Publication information
Journal: B M C Bioinformatics
Volume: 19
Article number: 239
ISSN (Print): 1471-2105
Ratings:
BFI (2019): BFI-level 1
Web of Science (2019): Indexed yes
BFI (2018): BFI-level 1
Web of Science (2018): Indexed yes
BFI (2017): BFI-level 1
Scopus rating (2017): CiteScore 2.49 SJR 1.479 SNIP 0.878
Web of Science (2017): Impact factor 2.213
Web of Science (2017): Indexed yes
BFI (2016): BFI-level 1
Scopus rating (2016): CiteScore 2.54 SJR 1.581 SNIP 0.974
Web of Science (2016): Impact factor 2.448
Web of Science (2016): Indexed yes
BFI (2015): BFI-level 1
Scopus rating (2015): CiteScore 2.77 SJR 1.737 SNIP 1.079
Web of Science (2015): Impact factor 2.435
Web of Science (2015): Indexed yes
BFI (2014): BFI-level 1
Scopus rating (2014): CiteScore 2.91 SJR 1.916 SNIP 1.185
Web of Science (2014): Impact factor 2.576
Web of Science (2014): Indexed yes
BFI (2013): BFI-level 1
Scopus rating (2013): CiteScore 3.38 SJR 1.999 SNIP 1.323
Web of Science (2013): Impact factor 2.672
ISI indexed (2013): ISI indexed yes
Web of Science (2013): Indexed yes
BFI (2012): BFI-level 1
Scopus rating (2012): CiteScore 3.24 SJR 1.9 SNIP 1.145
Web of Science (2012): Impact factor 3.024
ISI indexed (2012): ISI indexed yes
Web of Science (2012): Indexed yes
BFI (2011): BFI-level 1
Scopus rating (2011): CiteScore 3.34 SJR 1.662 SNIP 1.19
Web of Science (2011): Impact factor 2.751
ISI indexed (2011): ISI indexed yes
Web of Science (2011): Indexed yes
BFI (2010): BFI-level 1
Scopus rating (2010): SJR 1.775 SNIP 1.13
Web of Science (2010): Impact factor 3.029
Web of Science (2010): Indexed yes
BFI (2009): BFI-level 1
Scopus rating (2009): SJR 1.893 SNIP 1.295
Web of Science (2009): Indexed yes
BFI (2008): BFI-level 1
Scopus rating (2008): SJR 1.951 SNIP 1.13
Web of Science (2008): Indexed yes
Scopus rating (2007): SJR 1.973 SNIP 1.12
Web of Science (2007): Indexed yes
Scopus rating (2006): SJR 1.913 SNIP 1.21
Scopus rating (2005): SJR 2.635 SNIP 1.61
Web of Science (2005): Indexed yes
Scopus rating (2004): SJR 3.304 SNIP 1.723
Web of Science (2004): Indexed yes
Scopus rating (2003): SJR 3.063 SNIP 1.229
Web of Science (2003): Indexed yes
Scopus rating (2002): SJR 24.693 SNIP 1.02
Scopus rating (2001): SJR 0.527 SNIP 0.457
Original language: English
Keywords: HLA genotyping, NGS, Clinical genomics, Population genetics, Prediction
Electronic versions:
s12859_018_2239_6.pdf
DOIs:
10.1186/s12859-018-2239-6

Bibliographical note
This article is distributed under the terms of the Creative Commons Attribution 4.0 International License (http://creativecommons.org/licenses/by/4.0/), which permits unrestricted use, distribution, and reproduction in any medium, provided you give appropriate credit to the original author(s) and the source, provide a link to the Creative Commons license, and indicate if changes were made. The Creative Commons Public Domain Dedication waiver (http://creativecommons.org/publicdomain/zero/1.0/) applies to the data made available in this article, unless otherwise stated.
Research output: Research - peer-review ▶ Journal article – Annual report year: 2018