Benchmarking the HLA typing performance of Polysolver and Optitype in 50 Danish parental trios - DTU Orbit (01/04/2019)

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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

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