Harnessing the power of functional immunogenomics parameters to discover new associations with diseases

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The HLA system is the cornerstone of immune responses and is the **most polymorphic region** of the genome. In the past 10 years, GWAS identified more than 10,000 associations with many diseases and traits in the form of simple variations in the genome called SNP, and 1/3 of the GWAS catalog was associated with *HLA*.

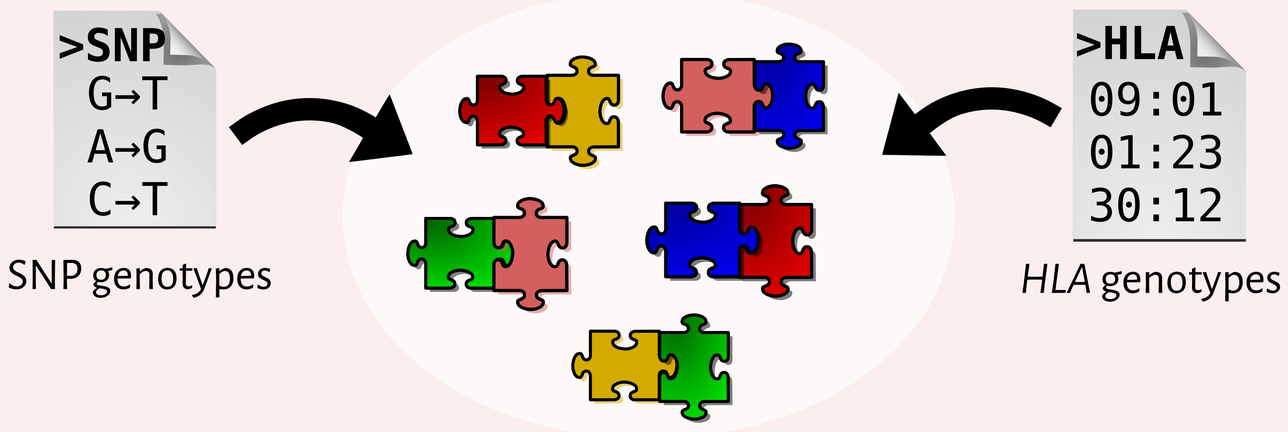
To go beyond the simple SNP associations, we now have the possibility to statistically infer missing information such as *HLA* alleles. We propose two toolboxes to fill the gap between available genomic data and novel functional immunogenomics parameters:

→ SNP-HLA reference consortium

→ Easy-HLA web suite.

SNP-HLA Consortium

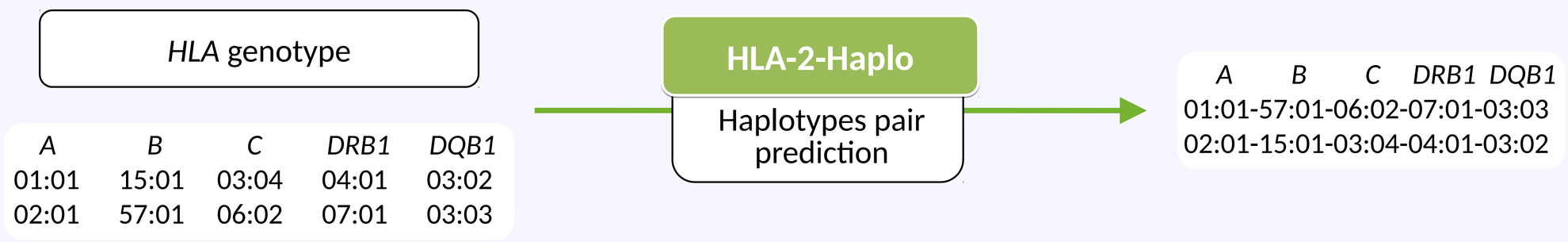
The aim of this consortium is to help *HLA* alleles imputation from SNP data by building and sharing large **reference panels**. The quality of *HLA* information obtained by imputation depends greatly on the similarity between the population and the reference panel.



The SNP-HLA Consortium will provide reference panels from **diverse** ethnicities, genotyping chips and resolutions.

Easy-HLA

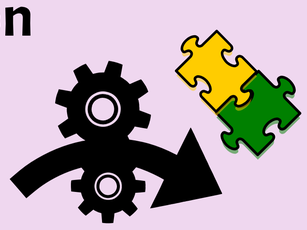
hla.univ-nantes.fr



Easy-HLA provides additional immunogenomics parameters: haplotypes pairs, imputed HLA-C expression, amino-acid equivalence of *HLA* alleles (HLA-AA) and KIR ligand classification of *HLA* alleles.

CAAPA example: *HLA* association study of asthma in an African-American population

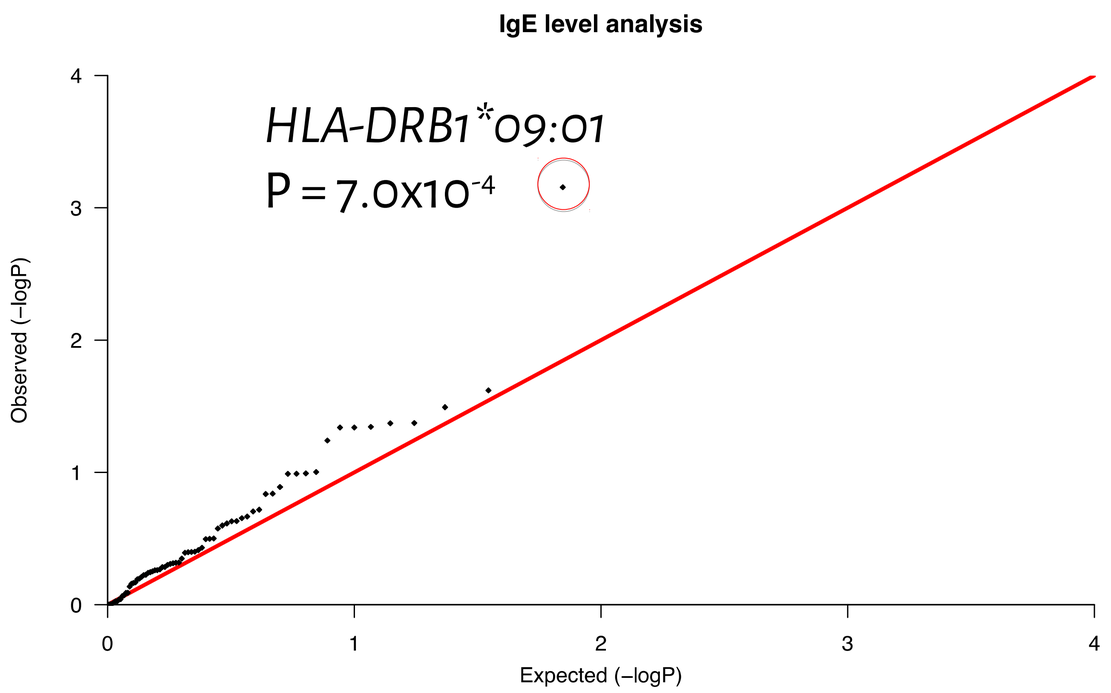
SNP	*HLA* information
G → A	??:??
T → C	??:??
A → G	??:??



HLA imputation using a large & appropriate reference panel

HLA information
01:01
30:01
06:02

Altogether, the immunogenetics tools developed for imputation and enrichment of *HLA* information allow us to investigate the complexity of *HLA* genotypes and to reveal **new associations** with diseases .



After *HLA* imputation, we identified an allele significantely associated with the IgE levels in CAAPA.

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