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► To cite this version:

Nicolas Vince, Estelle Geffard, Venceslas Douillard, Sophie Limou, Pierre-Antoine Gourraud. Harnessing the power of functional immunogenomics parameters to discover new associations with diseases. Labex IGO meeting 2018, Apr 2018, Nantes, France. inserm-02161825

HAL Id: inserm-02161825

<https://inserm.hal.science/inserm-02161825>

Submitted on 21 Jun 2019

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

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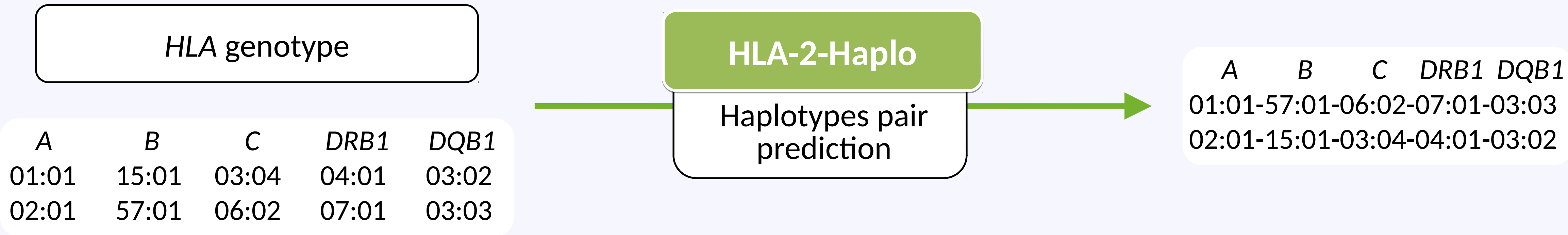
The HLA system is the cornerstone of immune responses and is the **most polymorphic region** of the genome. In the past 10 years, GWAS identified more than 10,000 associations with many diseases and traits in the form of simple variations in the genome called SNP, and 1/3 of the GWAS catalog was associated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

Easy-HLA

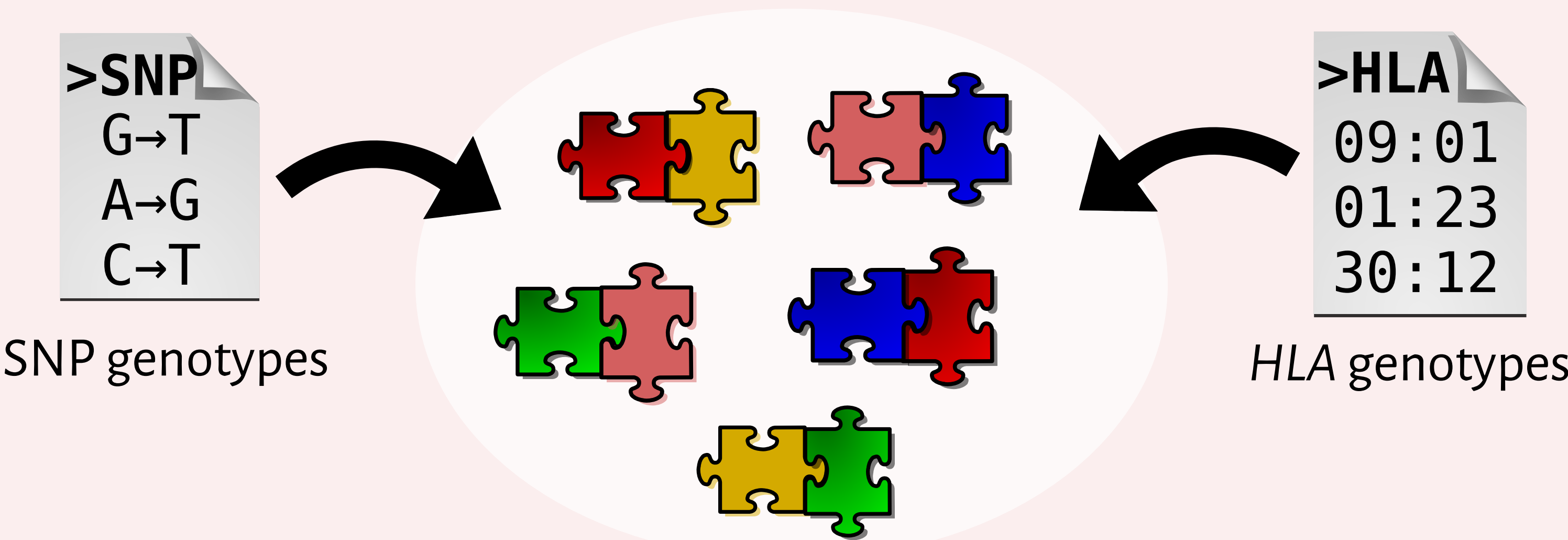
hla.univ-nantes.fr



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.

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.

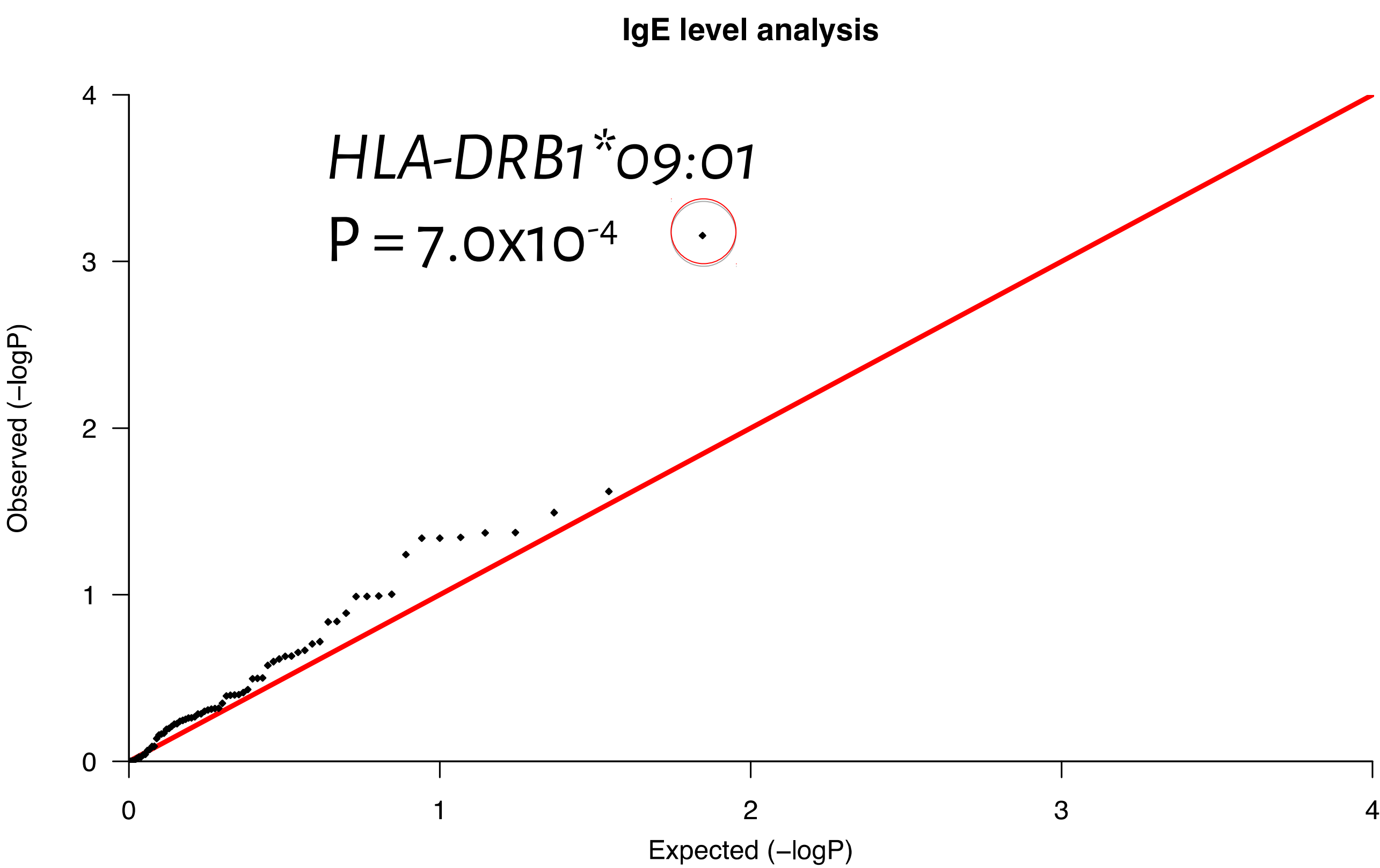
CAAPA example: HLA association study of asthma in an African-American population

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 significantly associated with the IgE levels in CAAPA.