

A Tool for Visual Exploration of Linkage Disequilibrium in Highly Polymorphic Systems of Loci

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Linkage disequilibrium (LD) refers to the non-random association between genetic variants at distinct loci within a population. Understanding the patterns of LD can provide insights into the demographic history and evolution of populations, besides helping in the identification of genes or markers associated with some diseases or traits of interest. However, to analyse LD patterns between highly polymorphic pairs of loci can be challenging, in particular when many haplotypes are present.

To manage such a wealth of information, we present a Shiny application that allows users to interactively visualize LD patterns in one or several populations using a circular graph representation. Thanks to this app, the users can dynamically control the number of haplotypes displayed by setting thresholds on relevant parameters such as haplotype frequency or intensity of LD deviation, but also ignore some loci, loci pairs or populations if desired. While the inputs are simple plain text files, graphical and numerical outputs are easily obtained in well-known open formats. This interactive tool is currently used to explore and analyse LD patterns in highly polymorphic multiallelic loci such as HLA (human MHC), where it allows identifying specific patterns resulting from populations' demography or natural selection.

This new Shiny application reveals the advantages of interactive data visualisation and exploration, in particular for the analysis of complex, non intuitive, parameters such as LD between highly polymorphic loci. Being particularly user-friendly, it will be most useful to researchers working in a variety of scientific domains such as systemic biology, genetics and population genetics.