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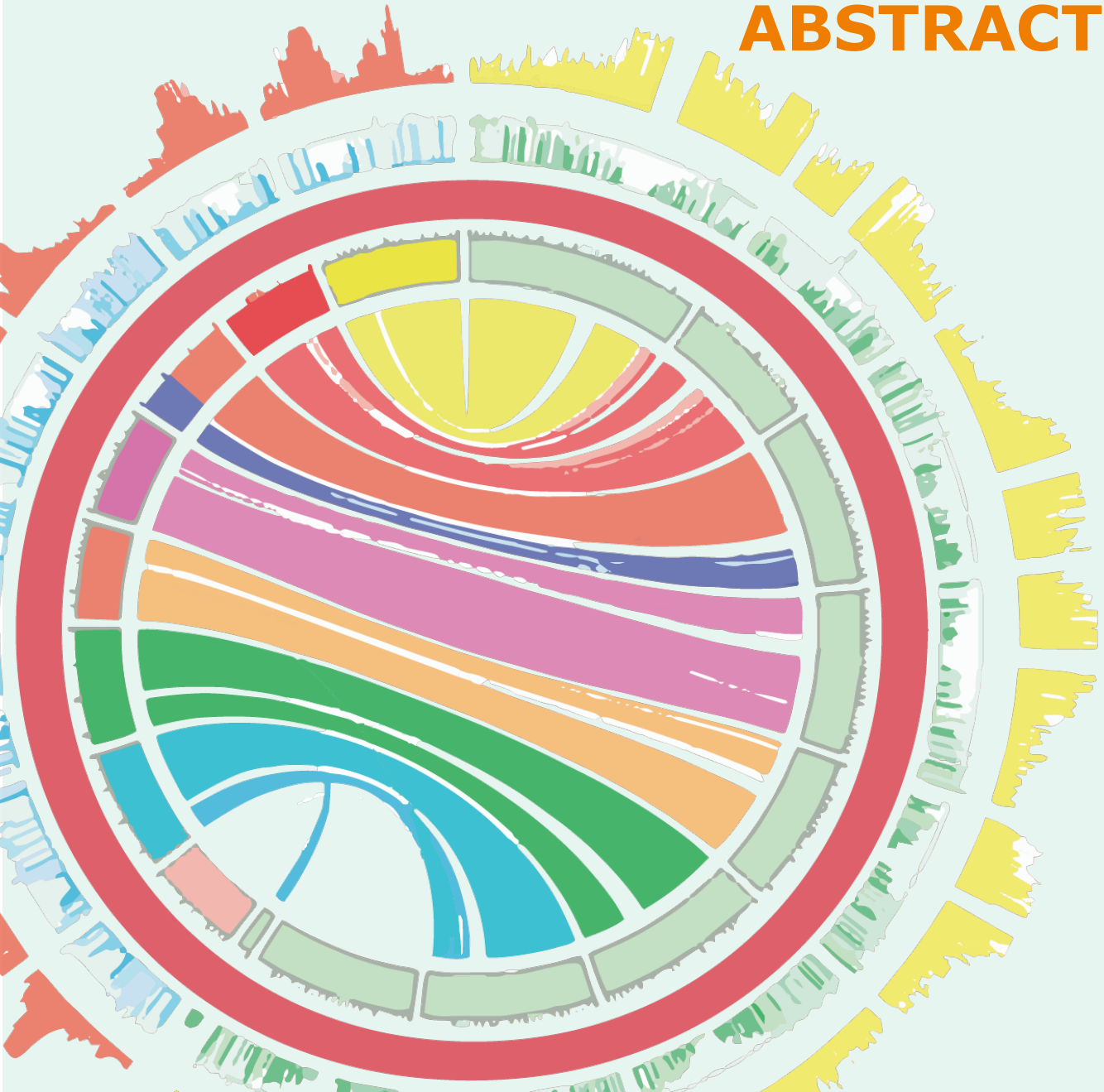
ABSTRACTS



Gigwa - Genotype Investigator for Genome-Wide Analyses

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With the advent of next-generation sequencing (NGS) technology, thousands of new genomes of both plant and animal organisms have become available. In this context, the Variant Call Format (VCF) [1] has become a convenient and standard file format for storing variants identified by NGS / NGG approaches. VCF files may contain information on tens of millions of variants, for thousands of individuals. Having to manage such significant volumes of data involves considerations of efficiency with regard to the following aspects: Filtering features, Storage performance, Sharing capabilities, Graphical visualization. However, existing tools are often limited to command line or programmatic APIs targeted at experienced users, but are not suitable for non-bioinformaticians.
The Gigwa application [2], which stands for "Genotype Investigator for Genome-Wide Analyses", aims at taking into account those aspects. It provides an easy and intuitive way to explore large amounts of genotyping data by filtering it not only on the basis of variant features, including functional annotations, but also on genotype patterns. It is a fairly lightweight, web-based, platform-independent solution that allows to feed a MongoDB [3] NoSQL database with VCF [4], PLINK or HapMap files containing up to billions of genotypes, and provides a user-friendly interface to filter data in real time. Gigwa provides the means to export filtered data into several popular formats and features connectivity with visualization software such as FlapJack [5] and online or standalone genome browsers (GBrowse, [REF]JBrowse [6] and IGV [7]). Additionnally, Gigwa-hosted datasets are interoperable via two standard REST APIs: GA4GH[8] and BrAPI [9]. Thus, we think that Gigwa could serve a large number of scientists by helping them to manage, filter and share their own data.

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Keywords: Genomic variations, VCF, HapMap, NoSQL, MongoDB, SNP, INDEL