Evolution of ohnologous chromosomes following Whole Genome Duplication in apple

Tanguy Lallemand, Sebastien Aubourg, Gilles Hunault, Jean-Marc Celton, Claudine Landès

HAL Id: hal-02928930

https://hal.archives-ouvertes.fr/hal-02928930

Submitted on 3 Sep 2020

Evolution of ohnologous chromosomes following Whole Genome Duplication in apple

Tanguy Lallemand [1], Sébastien Aubourg [1], Gilles Hunault [1], Jean-Marc Celton [1] and Claudine Landès [1]
IRHS-UMR1345, Université d'Angers, INRAE, Institut Agro, SFR 4207 QuaSaV, 49071, Beaucouzé, France

Introduction

Polyploidy has long been known as a driver of genetic innovation in eukaryotic organisms. A high quality genome was recently obtained for domesticated apple (*Malus domestica*) [1]. This new version confirmed a recent Whole Genome Duplication (WGD) event 50 million years ago [2]. This WGD is well preserved and does not exist in other *Rosaceae* which makes the apple tree a model of choice to study genes after WGD.
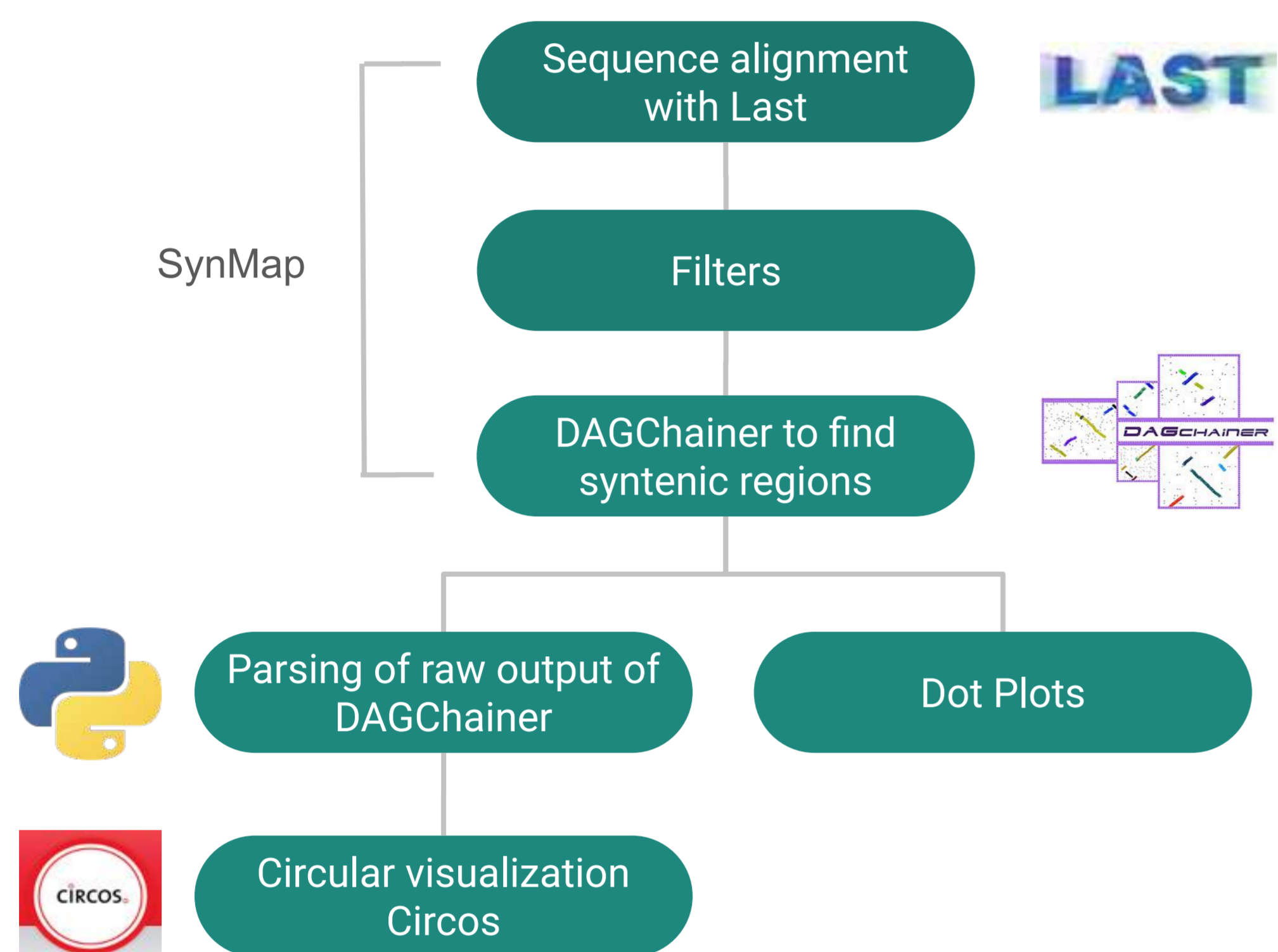
Quantitative Trait Loci (QTL) analysis allows the study of quantitative phenotypic variations, thus making possible to link phenotypic traits to areas of the genome using genetic maps.

Synteny blocks define the preservation of the order of genes between homologous genes along chromosomal segments. Our project aims at comparing ohnologous regions [3] (homologous regions obtained from WGD) to determine whether the proportions of QTLs is equivalent between syntenic pairs.

Methods

Synteny Blocks

In order to compare the different syntenic pairs, it is important to identify syntenic blocks on the apple genome. The SynMap software [4] was used according the following algorithm for this purpose.

- Sequence alignment with Last
- Filters
- DAGChainer to find syntenic regions
- SynMap
- Parsing of raw output of DAGChainer
- Dot Plots
- Circular visualization Circos

QTLs mapping

- Aims: Associate physical positions on the apple genome with QTLs, which are actually associated to genetically localized markers.
- Data: QTLs from GWAS or bi-parental studies, and genetic markers in apple. Files are coming from the GDR database [5].

QTL disequilibrium

- Aims: Check if there is some ohnologous pairs for which the presence of QTL is statistically unbalanced
- Data: QTLs localised on syntenic blocks
- Applied filters:
  - Origin of the QTL (biparental or GWAS)
  - Major or minor QTL by filtered on R²
  - Phenotypic traits categories
- Z test of proportions is used to test a difference of proportions between both syntenic blocks

Results

Successive steps allowed to map QTLs on apple genome. They are summarized in figure 1.
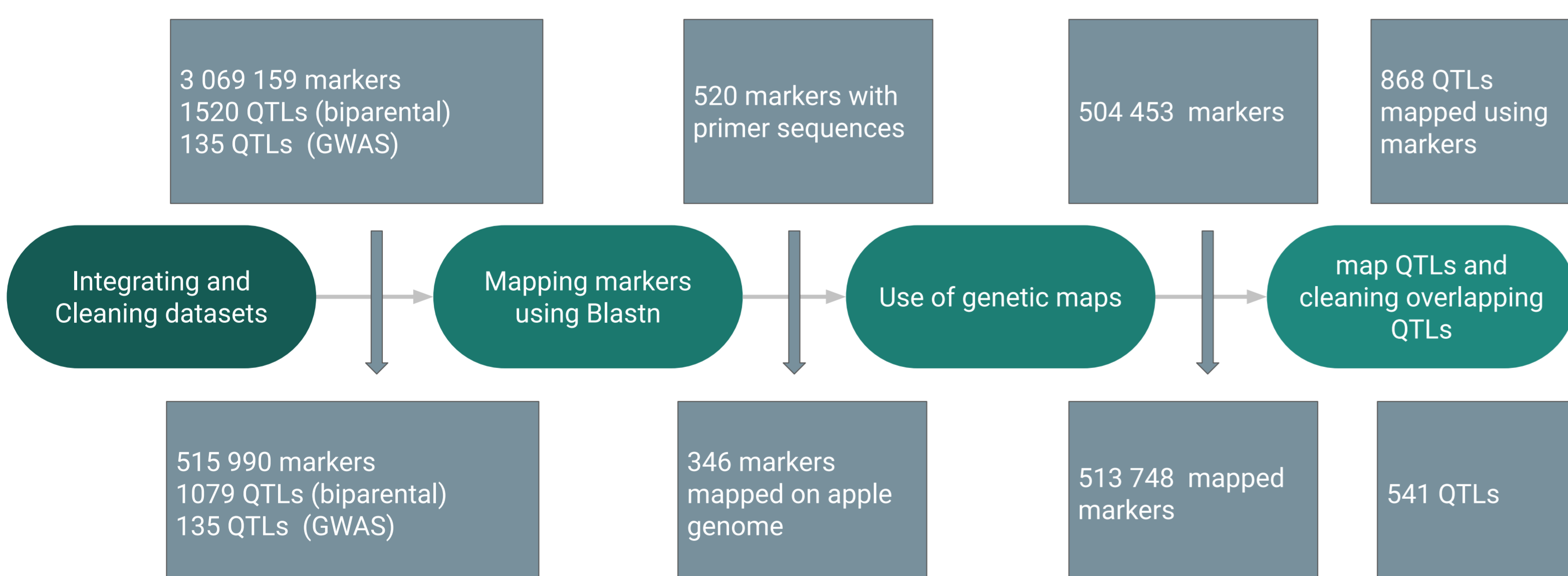
- 3 069 159 markers / 1520 QTLs (biparental) / 135 QTLs (GWAS)
- 520 markers with primer sequences
- 504 453 markers
- 868 QTLs mapped using markers
- Integrating and Cleaning datasets
- Mapping markers using Blastn
- Use of genetic maps
- map QTLs and cleaning overlapping QTLs
- 515 990 markers / 1079 QTLs (biparental) / 135 QTLs (GWAS)
- 346 markers mapped on apple genome
- 513 748 mapped markers
- 541 QTLs

Figure 1: *Summary of the results of the different steps of the script allowing the localization of QTLs on the apple genome.*

Conclusions

This project made possible to locate the different syntenic blocks existing within the apple genome using SynMap.. These blocks were used to check the presence of QTL imbalances between different syntenic pairs. To do so, QTLs were mapped on the apple physical genome using genetic markers. In addition, a cleaning of the possibly redundant data was done to avoid methodological bias in the QTL approach.

Results

A total of 587 blocks were identified. Pairs 3 and 11, 5 and 10, 9 and 17 and finally 13 and 16 are syntenic pairs at the chromosome scale. Other chromosomes such as 1, 2, 4, 7, 12 and 14 are blocks resulting from chromosomes splitting in half.
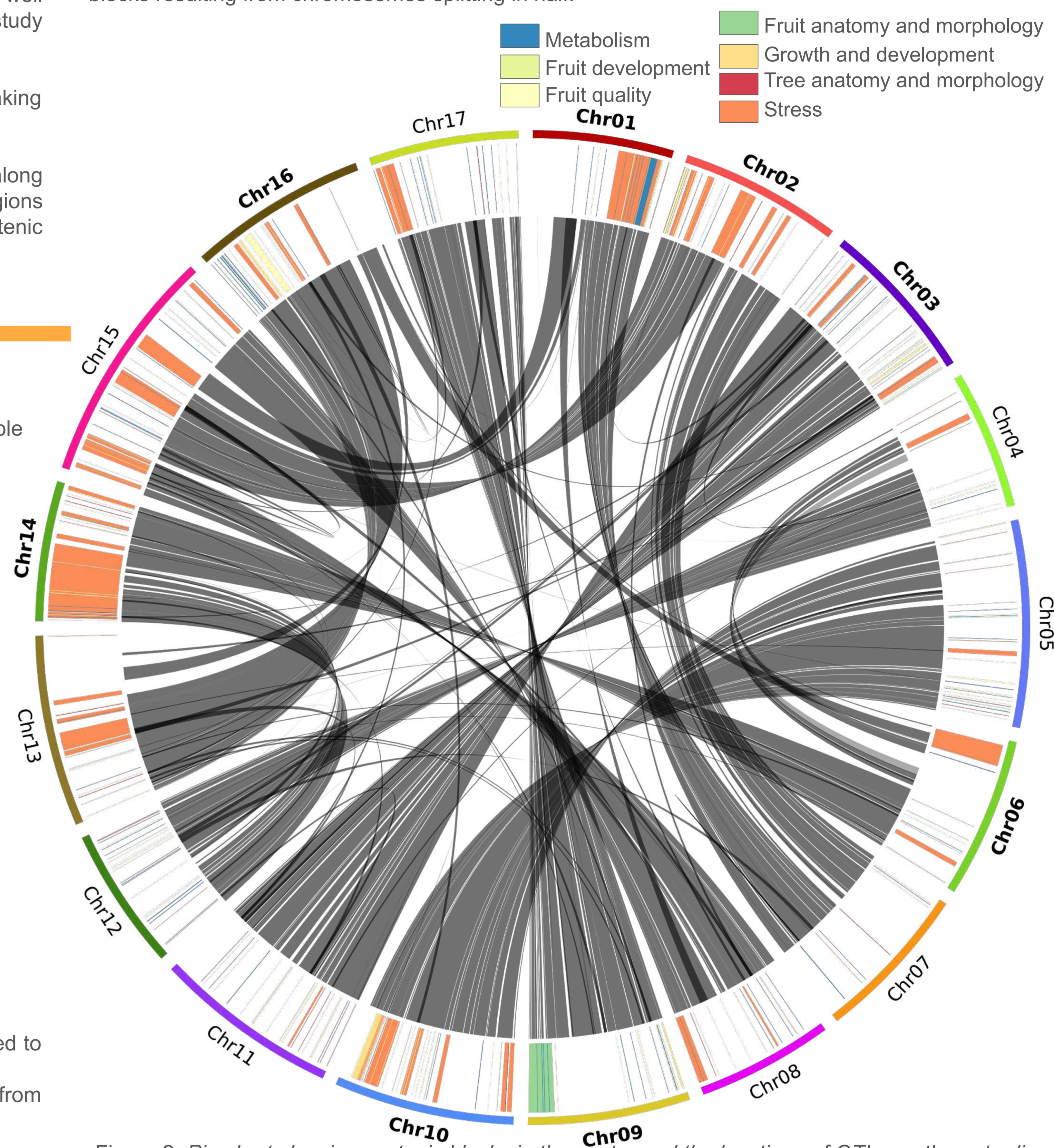

Figure 2: *Pie chart showing syntenic blocks in the center and the locations of QTLs on the outer line. The colors represent the category of phenotypic trait associated with the QTLs. Chromosome names in bold are those with the most QTLs compared to its homologous one. This figure has been created using Circos [6].*

Starting from the 541 physically localized QTLs, 486 were associated with syntenic blocks. A heatmap was constructed to summarize results and is presented as figure 3, which gathers all the results of the proportion tests.
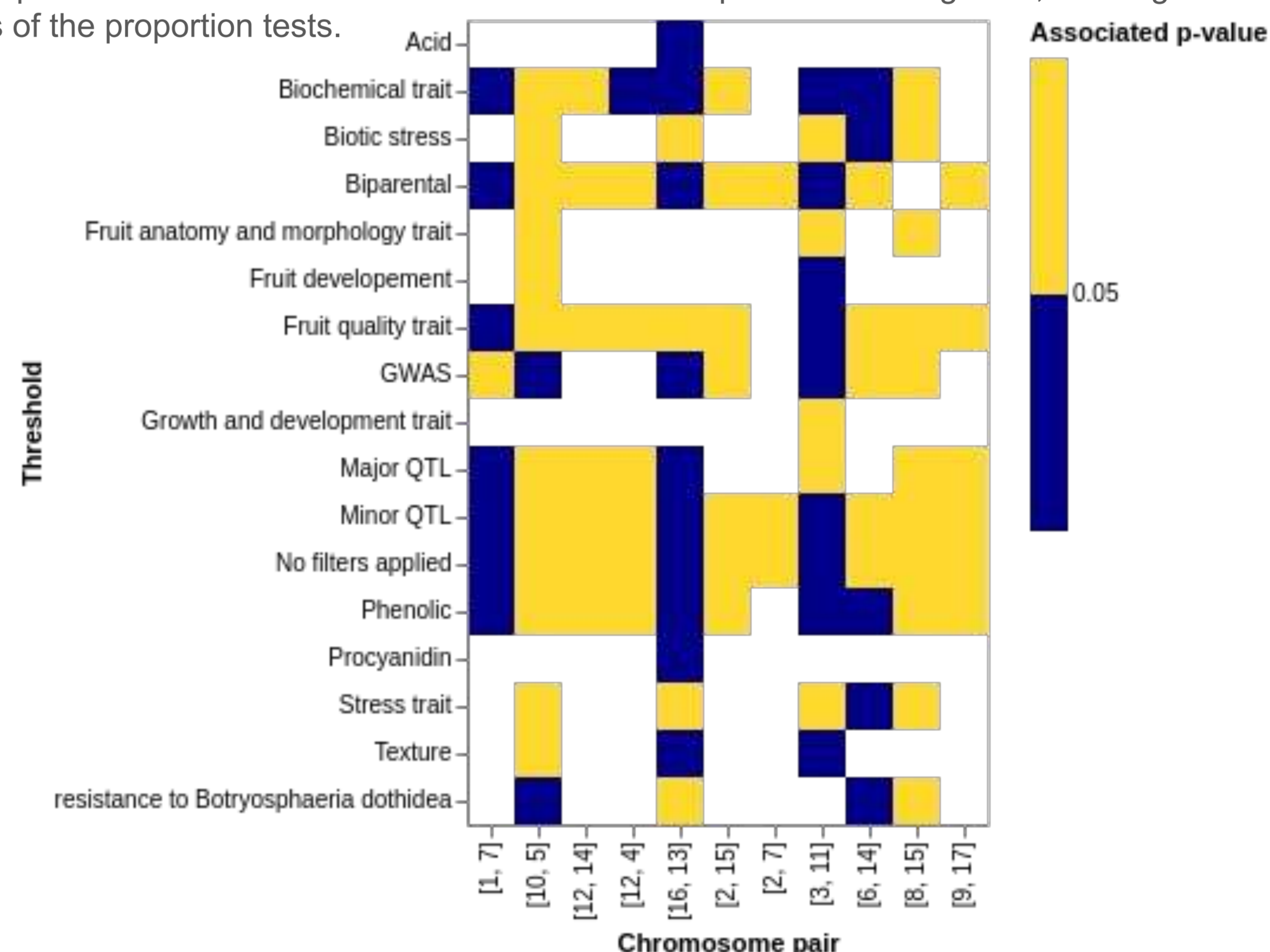

Figure 3: *Visualization of the proportion z test according to the pairs tested and the threshold applied. Significant p-values for a 5% threshold are in blue and non-significant ones in yellow. White cells are due to too small effective for conditions test. The first chromosome of the pair is the one with the most QTLs.*

The positioning of 541 QTLs allowed us to test whether the distribution of QTLs between syntenic pairs was equivalent or not. A disequilibrium seems to exist for some pairs and mainly: (1,7), (13,16), (3,11) and (6,14) and to a lesser extent (2,15), (4,12) and (5,10). Except for the (6,14) pair, the unbalance is always in the same direction whatever the applied filter on the QTLs of the syntenic pair.

References

[1] Nicolas Daccord, Jean-Marc Celton, et al. High-quality de novo assembly of the apple genome and methylome dynamics of early fruit development. 49(7):1099–1106.
[2] Riccardo Velasco, Andrey Zharkikh, et al. The genome of the domesticated apple (malus domestica borkh.). 42(10):833–839.
[3] Ohno S. (1970) Evolution by gene duplication. (Springer-Verlag, NY).
[4] Asher Haug-Baltzell, Sean A. Stephens, et al. SynMap2 and SynMap3d: web-based whole-genome synteny browsers. 33(14):2197–2198. Publisher: Oxford Academic.
[5] Sook Jung, Taein Lee, et al. 15 years of GDR: New data and functionality in the genome database for rosaceae. 47:D1137–D1145. Publisher: Oxford Academic.
[6] Krzywinski M, Schein J, Birol I, Connors J, Gascoyne R, et al. (2009) Circos: An information aesthetic for comparative genomics. Genome Res 19: 1639–1645.