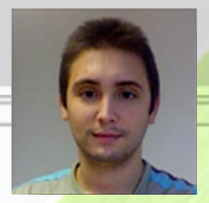
Development of a workflow for SNP detection with Galaxy




Marc Bras, Sandie Arnoux, Nacer Mohellibi, Nathalie Choisne, Olivier Inizan, Delphine Steinbach, Hadi Quesneville

Unité de Recherche en Génomique-Info UR1164, INRA de Versailles-Grignon, Route de Saint Cyr, Versailles, 78026, France

http://urgi.versailles.inra.fr urgi-contact@versailles.inra.fr

MAPHiTS : Mapping Analysis Pipeline for High-Throughput Sequences

Introduction :

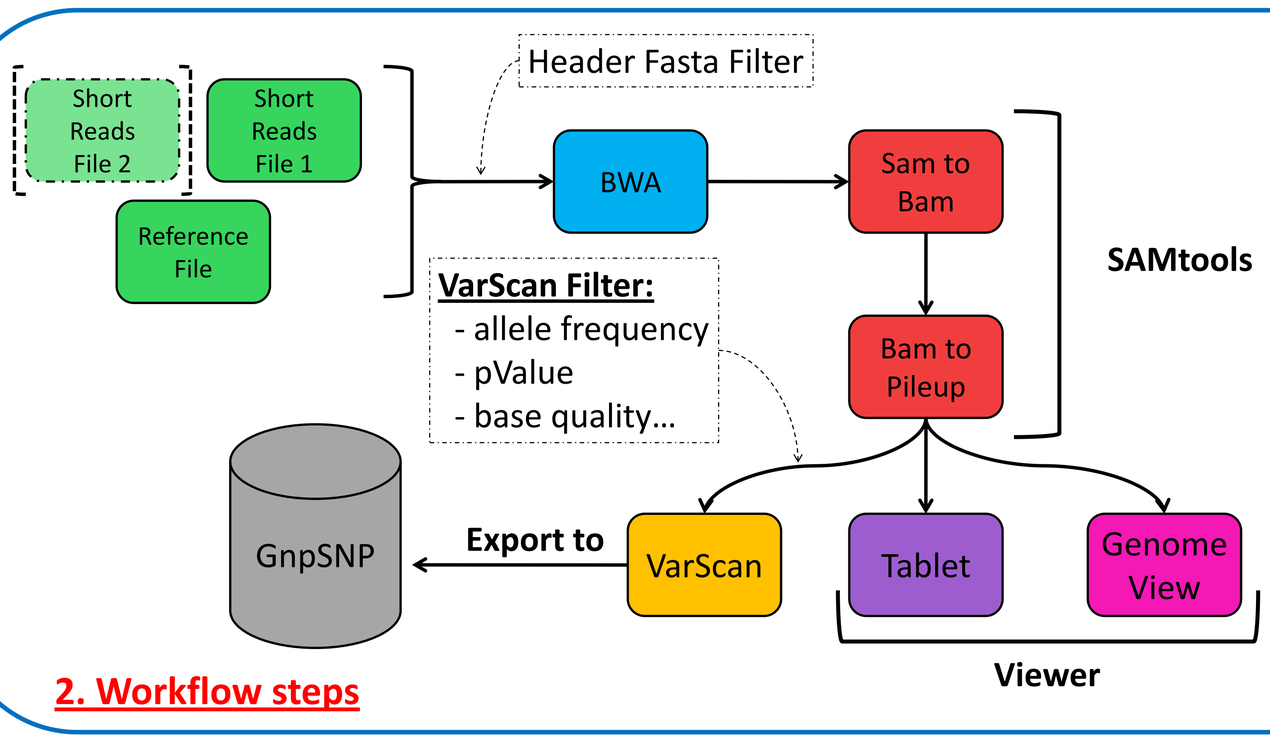
A Single-Nucleotide Polymorphism (SNP) is a DNA sequence variation. It can be used as a marker to characterize genetic variations between lineages. They can be used to detect complex traits such as those involved in diseases or agronomical performance.

The URGI platform developed a pipeline for SNPs detection from short reads, integrated in the Galaxy[1] workflow manager. *Galaxy* allows, through a web page, to chain different tools graphically. In addition, a large number of workflows can be built and shared.

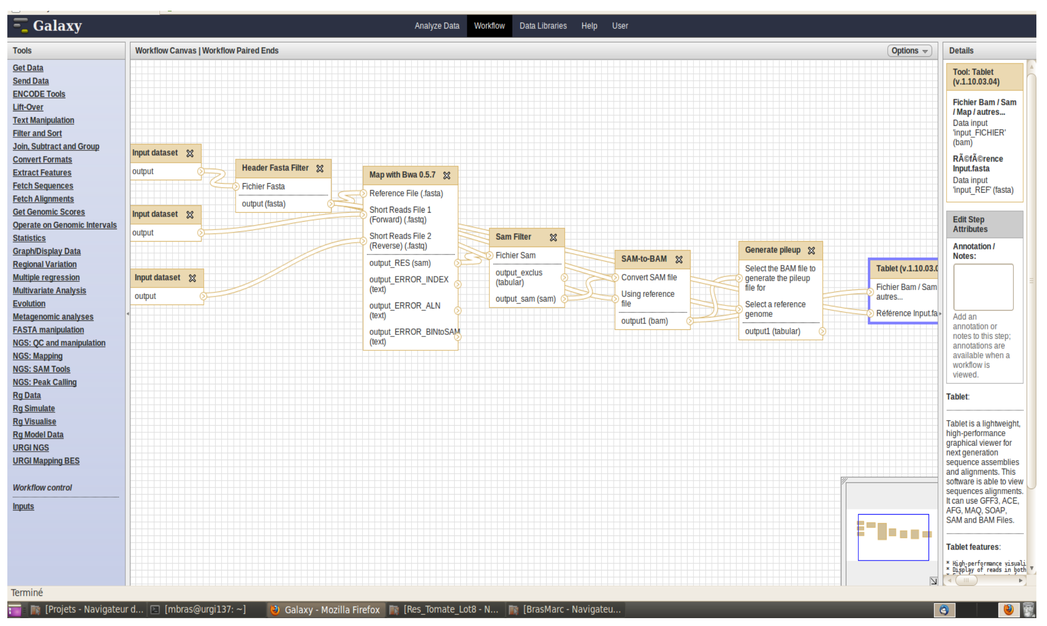
From a reference genome and a set of short reads (single-end or pair-ends), our workflow use *BWA*[2], *SAMtools*[3], *VarScan*[4] and *Tablet*[5] to predict SNPs and indels with number of filters, such as genome coverage, allele frequency, pValue.

Galaxy: web-based platform for genomic research, with many tools for NGS. They can be integrated into workflows.

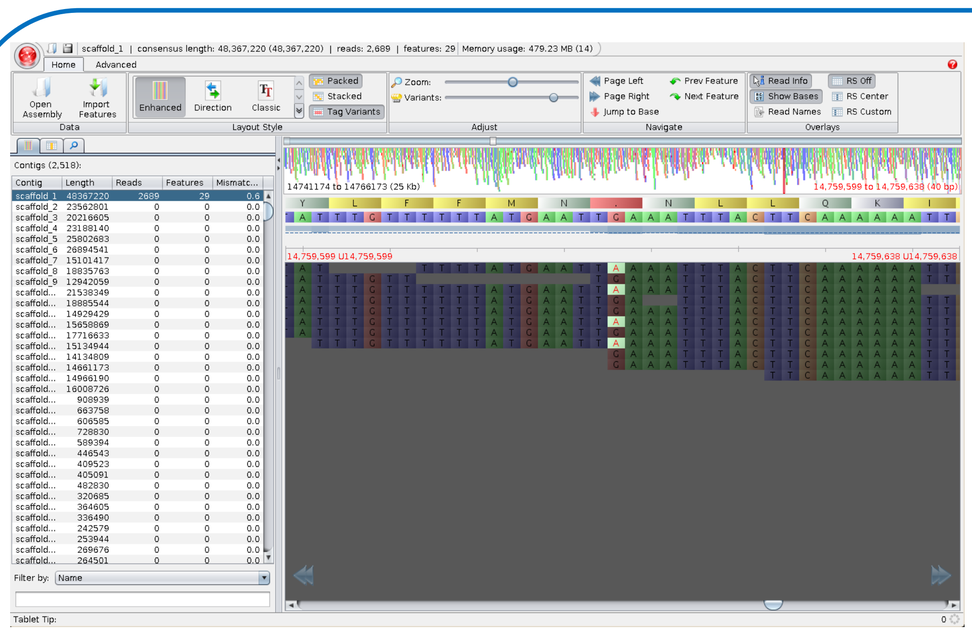
1. *Galaxy* home page



2. Workflow steps



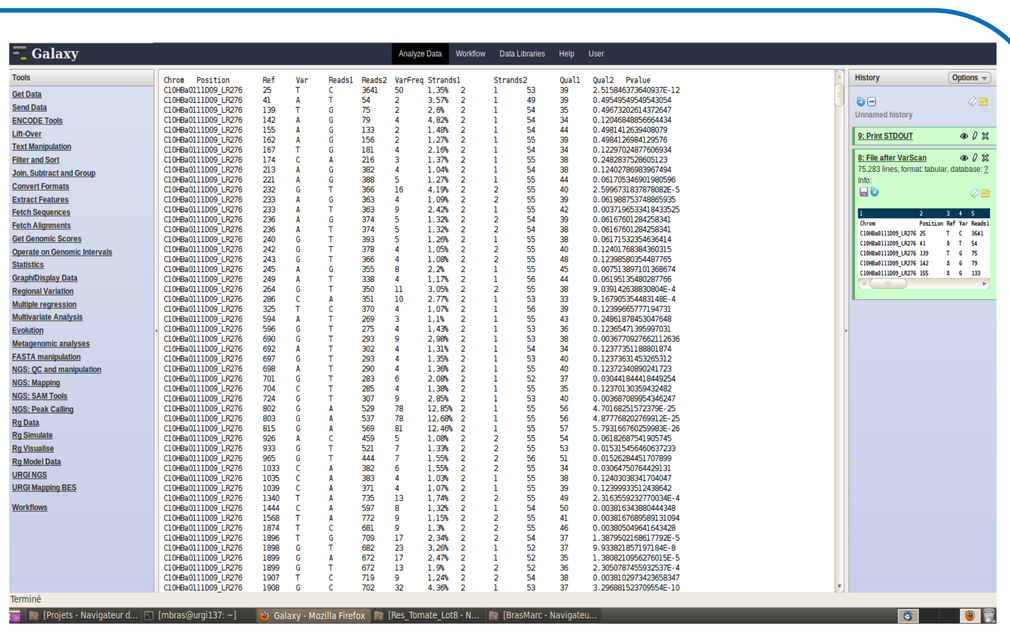
3. Workflow in *Galaxy*



4. Alignment results in *Tablet*

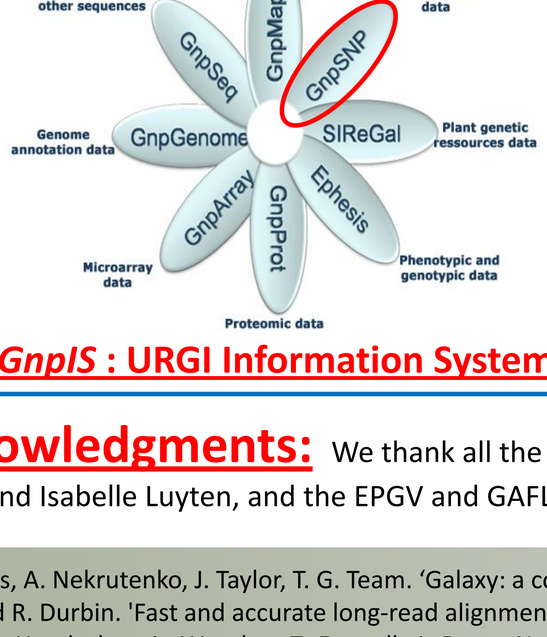
Results:

The alignment can be visualized with *Tablet* (or *GenomeView*[6] to see annotations). By changing the contrast, variants can be easily located (figure 4). *VarScan* can predict SNPs and indels. The output file contains information that can be used to filter them (variant frequency, average base quality, Pvalue…) (figure 5).

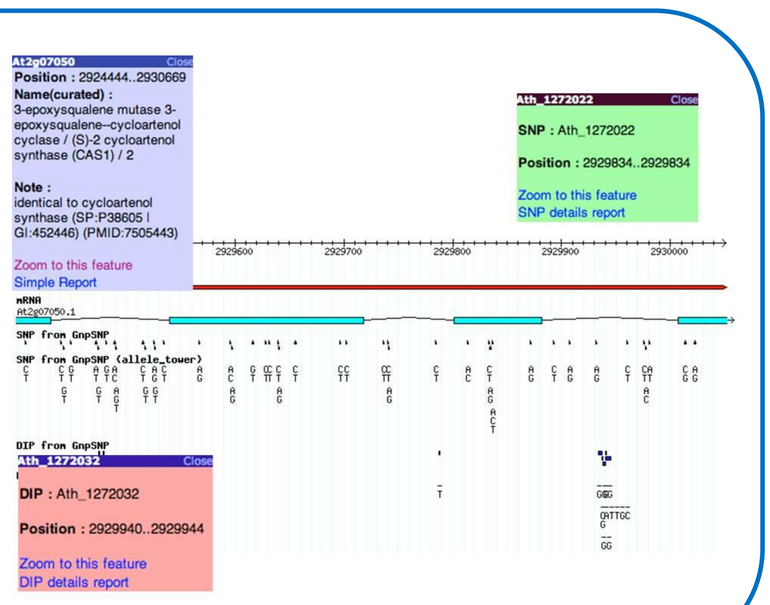


5. *VarScan* results in *Galaxy*

Insertion in database: the tool from *Galaxy* to *GnpSNP* (URGI database) export SNPs and indels to be visualized on Gbrowse.



6. *GnpIS* : URGI Information System



7. Results in *GnpSNP*

Conclusion :

Playing on different filter parameters and tools, this versatil workflow is able to detect SNPs and indels.

It has already been run on different data sets, from *A. thaliana, V. vinifera*, *Tomato* and *Poplar*.

The workflow will soon be available on our *Galaxy* instance.

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[1] J. Goecks, A. Nekrutenko, J. Taylor, T. G. Team. 'Galaxy: a comprehensive approach for supporting accessible, reproductible, and transparent computational research in the life sciences'. Genome Biology 11, R86+ (2010)

[2] H. Li and R. Durbin. 'Fast and accurate long-read alignment with Burrows-Wheeler transform'. Bioinformatics (2010). [PMID: 20080505]

[3] H. Li, B. Handsaker, A. Wysoker, T. Fennell, J. Ruan, N. Homer, G. Marth, G. Abecasis, R. Durbin and 1000 Genome Project Data Processing Subgroup. 'The Sequence alignment/map (SAM) format and SAMtools'. Bioinformatics, 25, 2078-9 (2009). [PMID: 19505943]

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[5] I. Milne, M. Bayer, L. Cardle, P. Shaw, G. Stephen, F. Wright and D. Marshall (2010). 'Tablet—next generation sequence assembly visualization'.Bioinformatics 2010 26(3):401-402.

[6] http://genomeview.sourceforge.net/