

Biological context in C-Sibelia

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C-Sibelia

This tool is designed for comparison between a reference and either a finished genome, or a genome represented as a set of contigs. It is able to detect SNPs/SNVs and indels of different scales. It also finds synteny blocks between these two genomes.

Goals

- 1 Add VCF output
Output variants and rearrangements found by C-Sibelia in VCF format
- 2 Annotation of SNPs and indels
Develop an application that is capable to annotate found variants using external databases.
- 3 Discovery and annotation of mobile elements

Goal 2

Annotation of SNP and indels

Variant annotation tools

- SIFT
- **snpEFF**
- ANNOVAR
- VARIANT
- ...

Problem:

Nobody work with structural variations, in the best case(ANNOVAR) only with most basic as duplications and deletions.

SNP and indels annotation

SnEff is a variant annotation and effect prediction tool. It annotates and predicts the effects of genetic variants (such as amino acid changes).

Work with: indels, SNPs, MNPs

Effects: START_GAINED, SPLICE_SITE_ACCEPTOR, SPLICE_SITE_DONOR, START_LOST, SYNONYMOUS_START, NON_SYNONYMOUS_START, EXON_DELETED, NON_SYNONYMOUS_CODING, SYNONYMOUS_CODING, FRAME_SHIFT, CODON_CHANGE, CODON_INSERTION, CODON_CHANGE_PLUS_CODON_INSERTION, etc

Databases: SnEff supports all bacterial genomes available at NCBI

Goal 3

Mobile elements discovery and annotation.

Transposable elements



A transposable element (TE) is a DNA sequence that can change its position within the genome, sometimes creating or reversing mutations and altering the cell's genome size. Transposition often results in duplication of the TE. Barbara McClintock's discovery of these jumping genes earned her a Nobel prize in 1983.

Why transposable elements are important?

- Transposable elements play an important role in evolution
- Transposable elements can break some gene on insertion
- **Assembled contigs are often starting or ending on mobile element**



Mobile elements discovery and annotation

Done:

- Search for all mobile elements and its fragments in contigs and reference using blast in NCBI database
- Prototype of module for prediction of neighboring contigs splitted on mobile element

To do:

- Finish mobile elements search module
- Filter results output
- Write the manual

Questions

Thank you for your attention!