

#### Speeding up NGS software development

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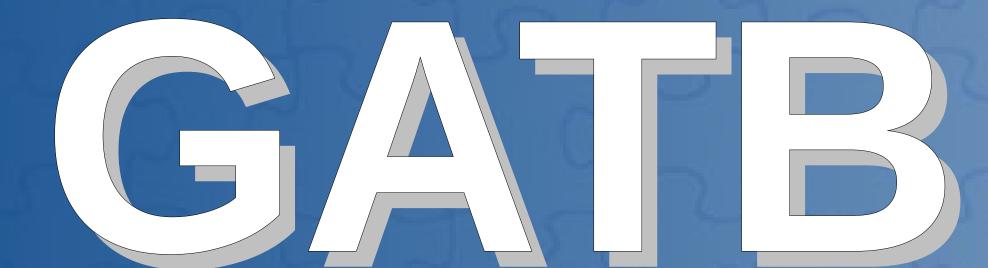
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# Speeding-up NGS software development

Sequencing, Finishing and Analysis in the Future Meeting, Santa Fé, USA, May 2014

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graph node

False Positive

**Critical False Positive** (additional structure)

Howto

Analyze Complex

Genomes on a

Simple Desktop

Computer?

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>read 100.000.000 TCTCCTAGCGCGGCGTATACGC

AAA

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#### 1. What is GATB?

# Motivation

NGS technologies produce terabytes of data. Efficient and fast NGS algorithms are essential to analyze them.

#### Objective

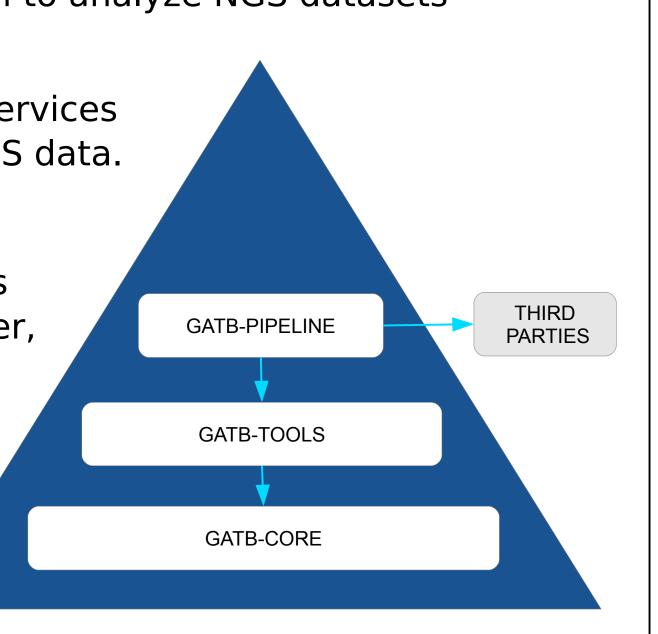
The Genome Assembly Tool Box (GATB)

- ► is an <u>open-source software</u>
- ► provides an easy way to develop efficient and fast NGS tools
- ► is based on data structure with <u>a very low memory footprint</u>
- ► allows complex genomes to be processed on desktop computers

# 2. Software Solution

The GATB philosophy proposes a 3-layer construction to analyze NGS datasets

- 1. **GATB-CORE**: a C++ library holding all the services needed for developing software dedicated to NGS data.
- 2. **GATB-TOOLS**: a set of elementary NGS tools mainly built upon the GATB library (k-mer counter, contiger, scaffolder, variant detection, etc.).
- 3. **GATB-PIPELINE**: a set of NGS pipeline that links together tools from the previous layer.



# 3. Compact de Bruijn graph data structure

The core data structure of GATB is a de Bruijn graph that encodes the main information from the sequencing reads.

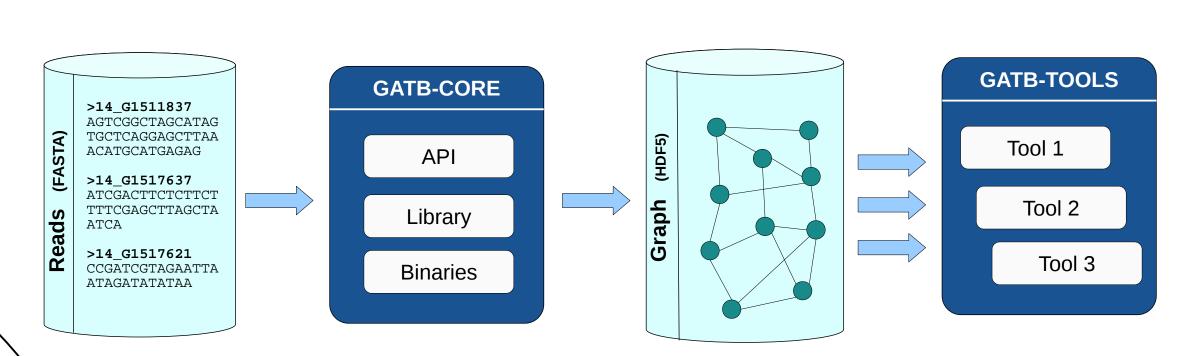
## **Strength of GATB**

GATB makes this graph compact by using a <u>Bloom filter</u> (a space efficient probabilistic

data structure) and by using a CFP additional structure that avoids false positive answers from the Bloom filter due to its probabilistic nature.

# 4. Workflow

Here is a typical workflow when working with GATB



GATB-CORE transforms the reads into a de Bruijn graph, saves it in a HDF5 file that can be opened by other tools developed with the GATB-CORE API.

# 5. GATB helps you as a NGS user

GATB's de Bruijn graph: a basis for families of tools

- **▶** Data error correction
- Assembly
- **►** Biological motif detection \_

a whole human genome sequencing reads can be handled with 5 GBytes of memory

Several tools based on GATB are already available

Bloocoo K-mer spectrum based read error corrector for large datasets

**Minia** Short read assembler based on a de Bruijn graph. Results are of similar contiguity and accuracy to other de Bruijn assemblers (e.g. Velvet)

**DiscoSNP** Discover Single Nucleotide Polymorphism (SNP) from non-assembled reads

**TakeABreak** Detects inversion breakpoints without a reference genome by looking for fixed size topological patterns in the de Bruijn graph

# 6. GATB helps you as a NGS developer

The GATB C++ library gives you the opportunity to quickly develop new NGS tools that fit your needs.

Major facts about the GATB C++ library

- ➤ Object Oriented Design
- Simple and powerful graph API
- ➤ Simple and powerful multithreading model
- ► HDF5 usage for data storage
- ► Fully documented with numerous code samples
- Complete test suite

# **Publications**

G. Rizk, D. Lavenier, R. Chikhi, **DSK: k-mer counting with very low memory** 

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G. Collet, G. Rizk, R. Chikhi, D. Lavenier, Minia on Raspberry Pi, assembling

a 100 Mbp genome on a Credit Card Sized Computer, Poster at the JOBIM

Lecture Notes in Computer Science, Volume 8126, 2013, pp 364-376

conference, 2013 Jul 1-4 (Toulouse) Best poster award.

R. Chikhi, G. Rizk. Space-efficient and exact de Bruijn graph representation

K.I Salikhov, G. Sacomoto, G. Kucherov, **Using Cascading Bloom Filters to Improve the Memory Usage for de Brujin Graphs**, Algorithms in Bioinformatics,

## License & Web Site

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Proprietary licencing for software editors or services providers is currently being studied.

For more details on GATB:

http://gatb.inria.fr

