Speeding up NGS software development

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GATB

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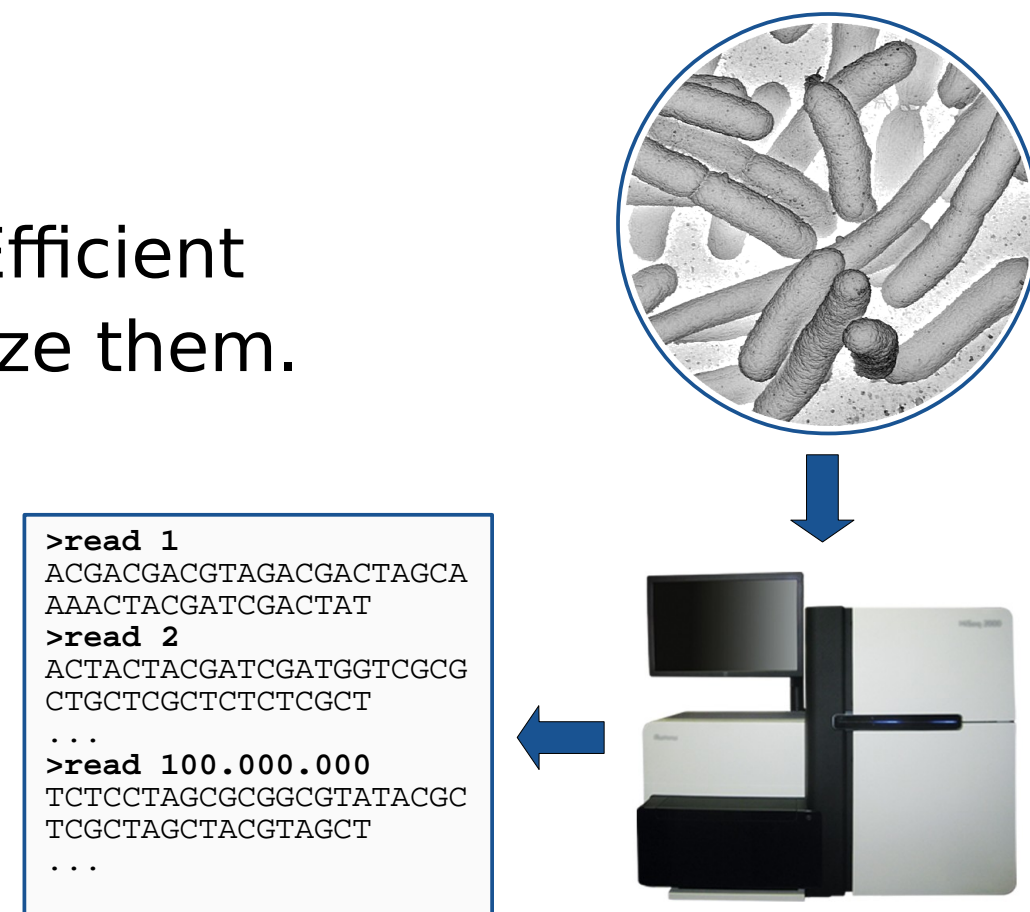
1. What is GATB ?

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

```
>read 1
ACGACGACGTAGACGACTAGCA
AAACTACGATCGACTAT
>read 2
ACTACTACGATCGATGGTCGCG
CTGCTCGCTCTCTCGCT
...
>read 100.000.000
TCTCCTAGCGCGGCGTATACGC
TCGCTAGCTACGTAGCT
...
```

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.

GATB-PIPELINE → THIRD PARTIES
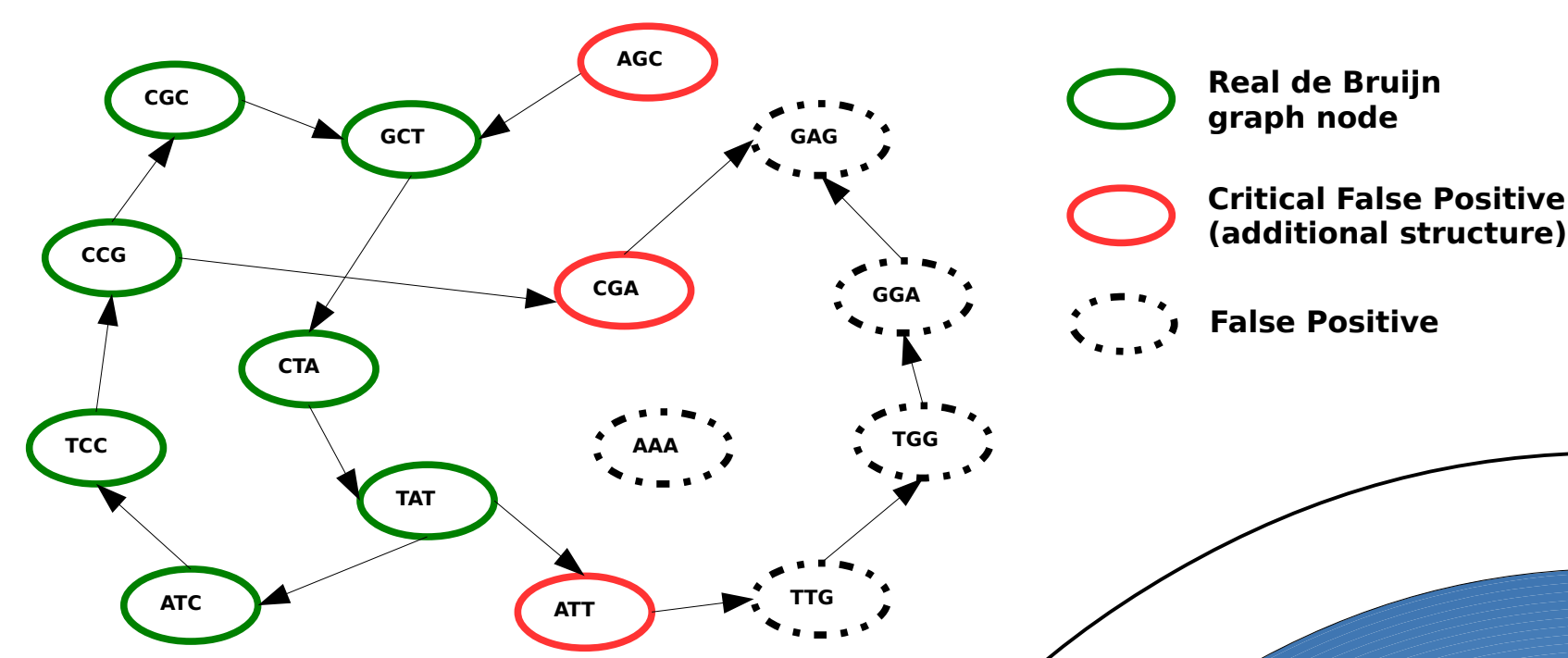GATB-TOOLS
GATB-CORE
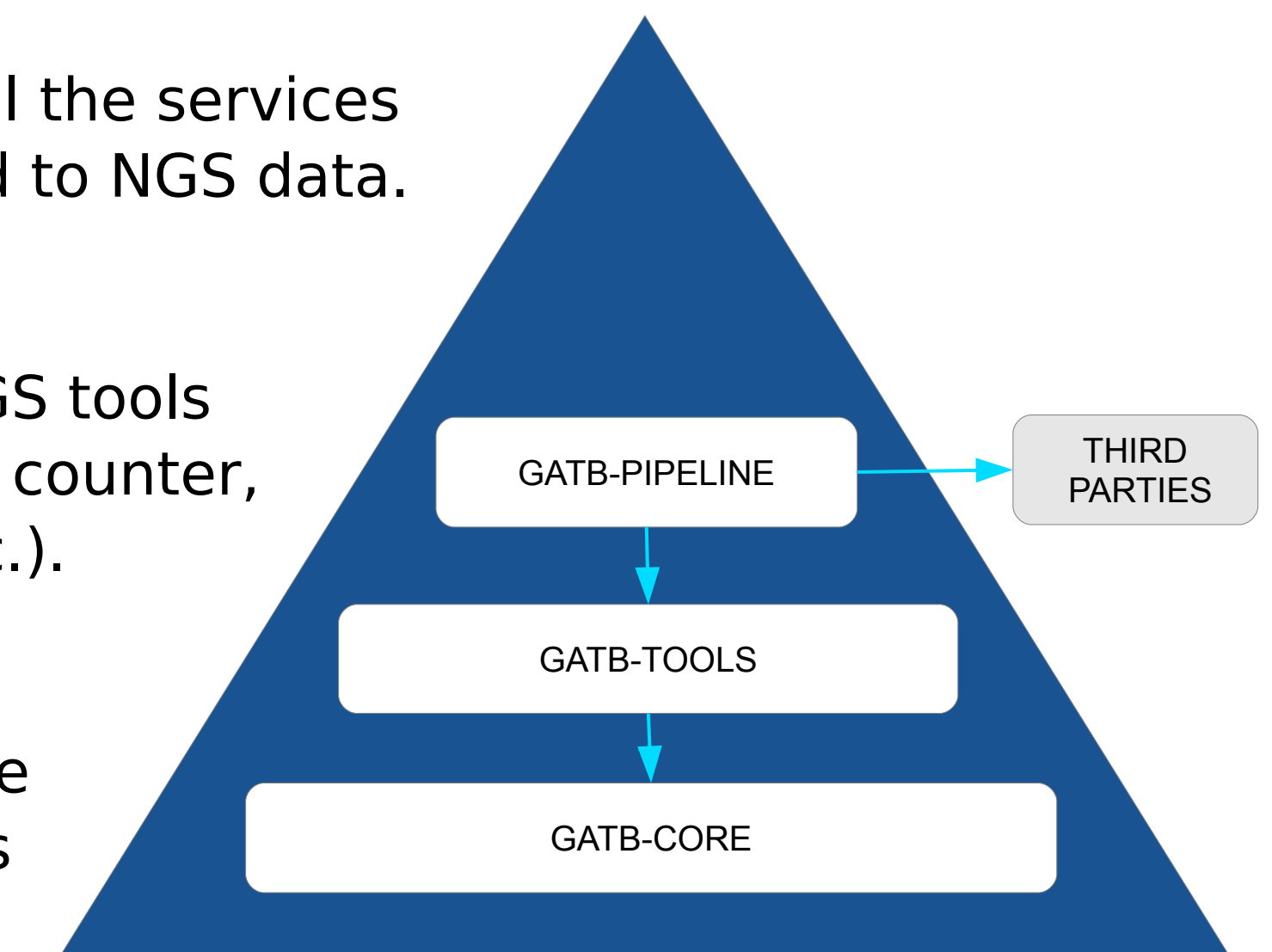
3. Compact de Bruijn graph data structure

The core data structure of GATB is a de Bruijn graph that encodes the main information from the sequencing reads.
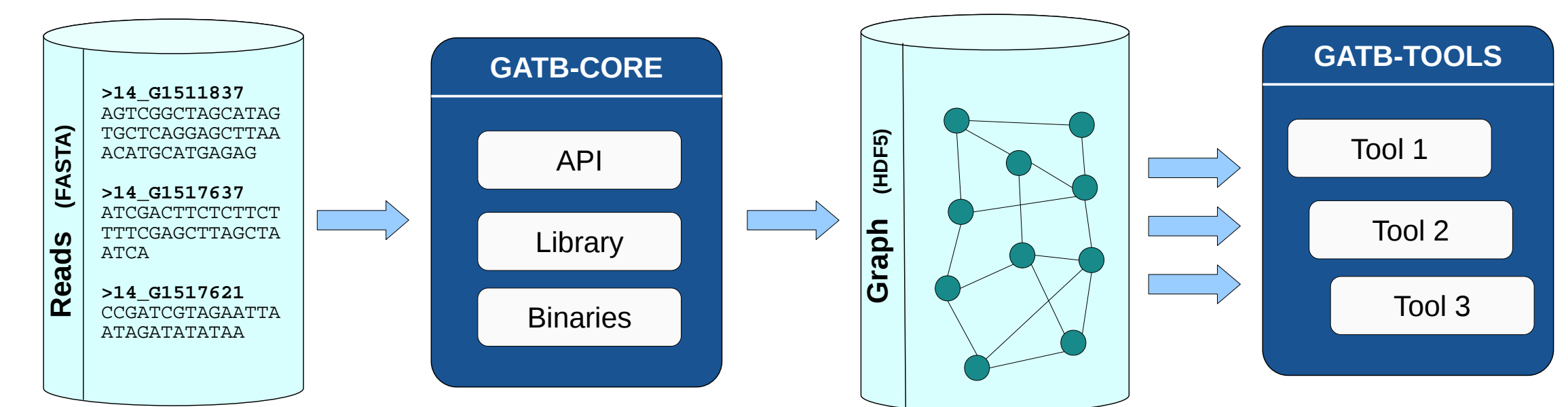
- Real de Bruijn graph node
- Critical False Positive (additional structure)
- False Positive

Strength of GATB

GATB makes this graph compact by using a Bloom filter (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

Reads (FASTA)
```
>14_G1511837
AGTCGGCTAGCATAG
TGCTCAGGAGCTTAA
ACATGCATGAGAG
>14_G1517637
ATCGACTTCTCTTCT
TTTCGAGCTTAGCTA
ATCA
>14_G1517621
CCGATCGTAGAATTA
ATAGATATATA
```

GATB-CORE
- API
- Library
- Binaries

Graph (HDF5)

GATB-TOOLS
- Tool 1
- Tool 2
- Tool 3

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.

How to Analyze Complex Genomes on a Simple Desktop Computer ?

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 usage**, Bioinformatics, 2013 Mar 1;29(5):652-3

R. Chikhi, G. Rizk. **Space-efficient and exact de Bruijn graph representation based on a Bloom filter**, Algorithms for Molecular Biology 2013, 8:22

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

K.I Salikhov, G. Sacomoto, G. Kucherov, **Using Cascading Bloom Filters to Improve the Memory Usage for de Brujin Graphs**, Algorithms in Bioinformatics, Lecture Notes in Computer Science, Volume 8126, 2013, pp 364-376

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For more details on GATB:

http://gatb.inria.fr

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