



HAL
open science

Speeding up NGS software development

Erwan Drezen, Guillaume Rizk, Rayan Chikhi, Charles Deltel, Claire Lemaitre, Pierre Peterlongo, Dominique Lavenier

► **To cite this version:**

Erwan Drezen, Guillaume Rizk, Rayan Chikhi, Charles Deltel, Claire Lemaitre, et al.. Speeding up NGS software development. Sequencing, Finishing and Analysis in the Future Meeting, May 2014, Santa Fé, United States. hal-01088683

HAL Id: hal-01088683

<https://hal.science/hal-01088683>

Submitted on 16 Jan 2015

HAL is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.

Erwan Drézen¹, Guillaume Rizk¹, Rayan Chikhi², Charles Deltel¹, Claire Lemaitre¹, Pierre Peterlongo¹ and Dominique Lavenier¹

¹ INRIA/IRISA/GenScale, Campus de Beaulieu, 35042 Rennes cedex

² Department of Computer Science and Engineering, Pennsylvania State University, USA

1. What is GATB ?

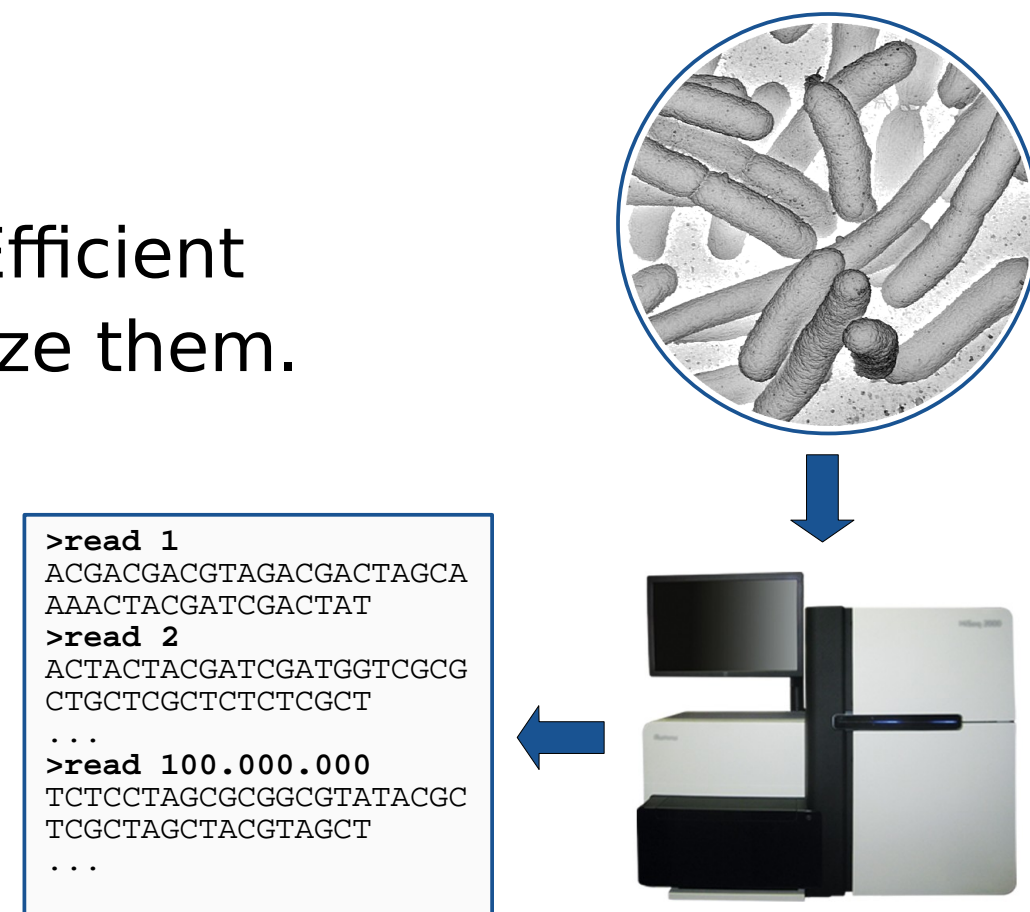
Motivation

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



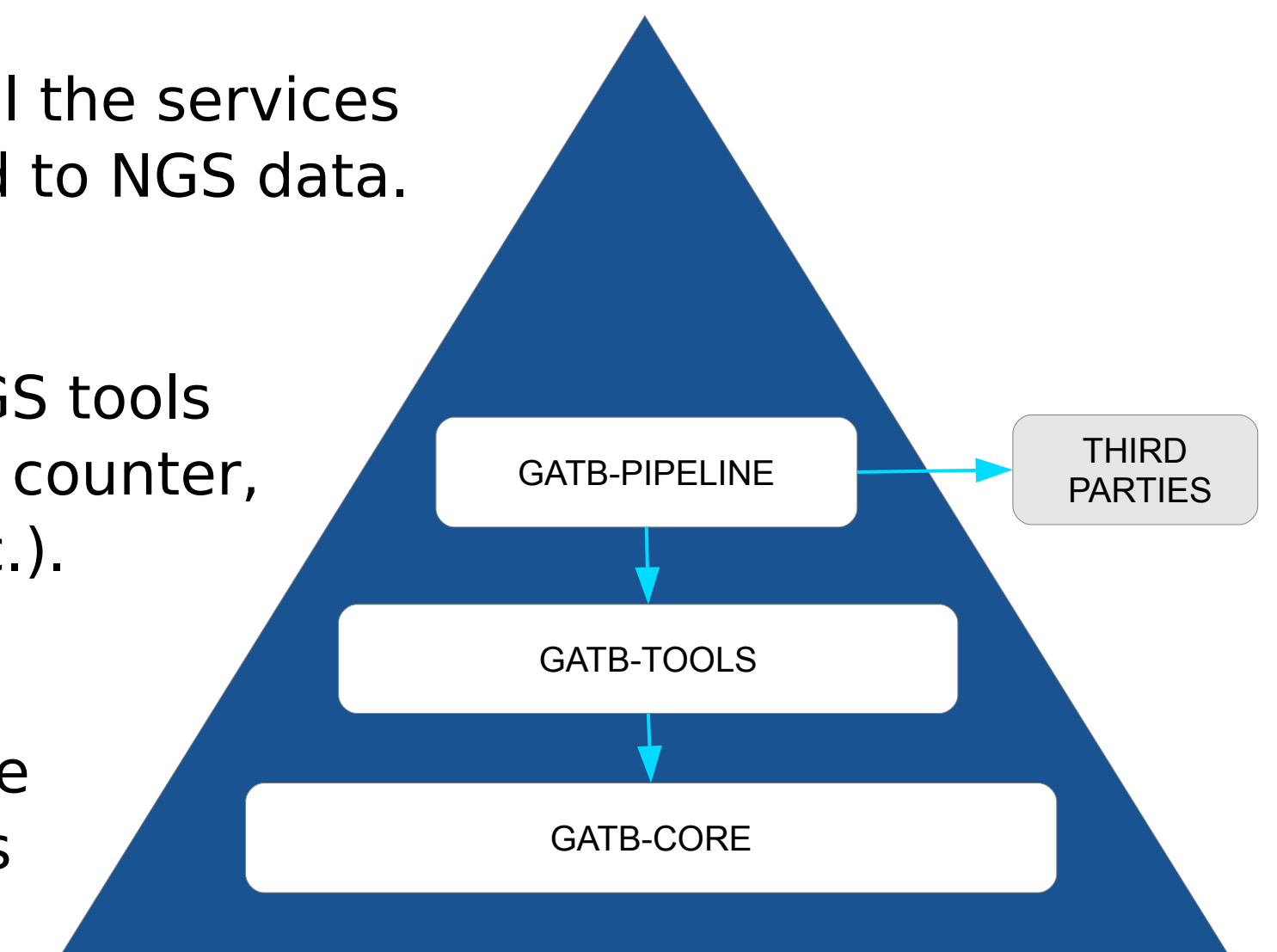
2. Software Solution

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.

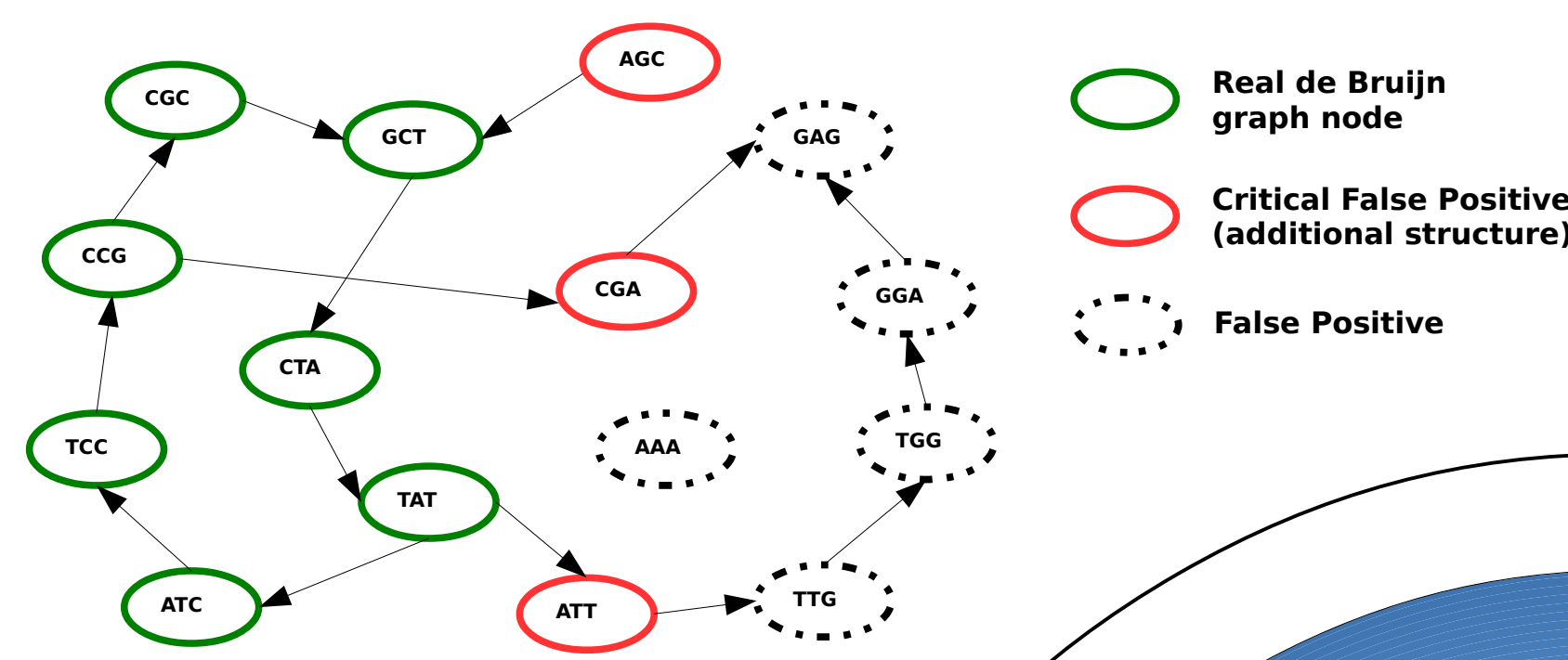


3. Compact de Bruijn graph data structure

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

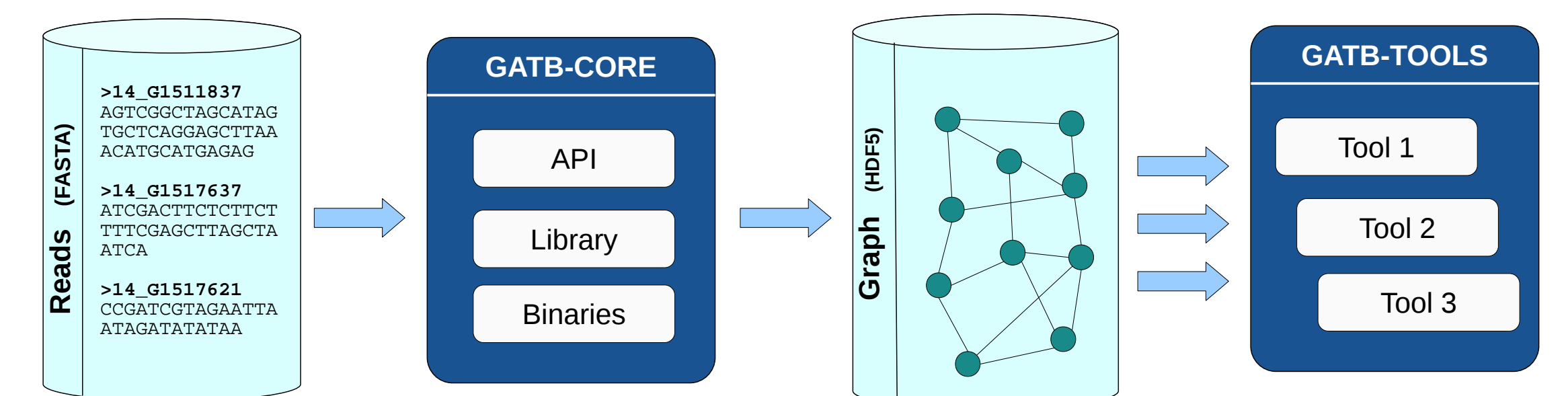
Strength of GATB

GATB makes this graph compact by using a 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



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

- Blooco** 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

How to Analyze Complex Genomes on a Simple Desktop Computer ?

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 Bruijn Graphs**, Algorithms in Bioinformatics, Lecture Notes in Computer Science, Volume 8126, 2013, pp 364-376

License & Web Site

GATB is released under the GNU Affero General Public License.

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

For more details on GATB:

<http://gatb.inria.fr>

Partners

