FOSS
IIT Bombay

Symposium on
FREE & OPEN SOURCE SOFTWARE
Usage in Teaching & Research

Jupyter Notebook: Software Development for Computational Genomics

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Research can begin from the smallest of sparks

Presentation Outline

Introduction

Our Requirements

Our Approach

Our Results

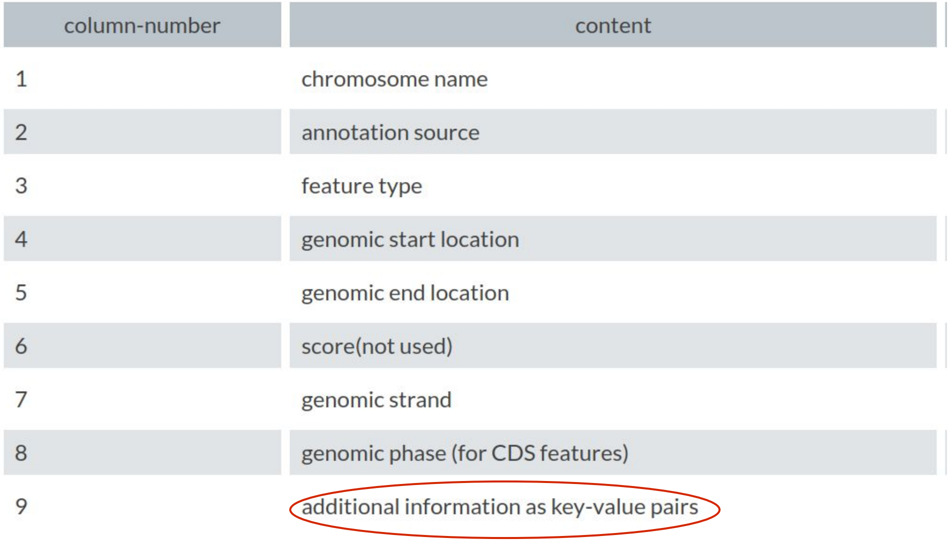
Conclusions and Challenges

Appendix

Introduction

Genomic data is stored in GTF file format:

- Very unintuitive
- Difficult to process
- Space inefficient



column-number	content
1	chromosome name
2	annotation source
3	feature type
4	genomic start location
5	genomic end location
6	score(not used)
7	genomic strand
8	genomic phase (for CDS features)
9	additional information as key-value pairs

Introduction

Existing Software:

- Pyranges (Python)
- Gffutils (Python)
- Plyranges (R)

Problems:

- Long loading times
- Long query times (for certain types of queries)

Stovner, E. B., Sætrom, P. (2019). PyRanges: efficient comparison of genomic intervals in Python. Bioinformatics, 36(3), 918-919.
Dale, R. (2011). gffutils: GFF and GTF file manipulation and interconversion. Retrieved from https://github.com/daler/gffutils.
Lee, S., Cook, D., Lawrence, M. (2019). Plyranges: A grammar of genomic data transformation. Genome Biology, 20, 1-8.

Requirements

Space Efficient

Fast

(easy to query)

Standardized

(well known)

Approach

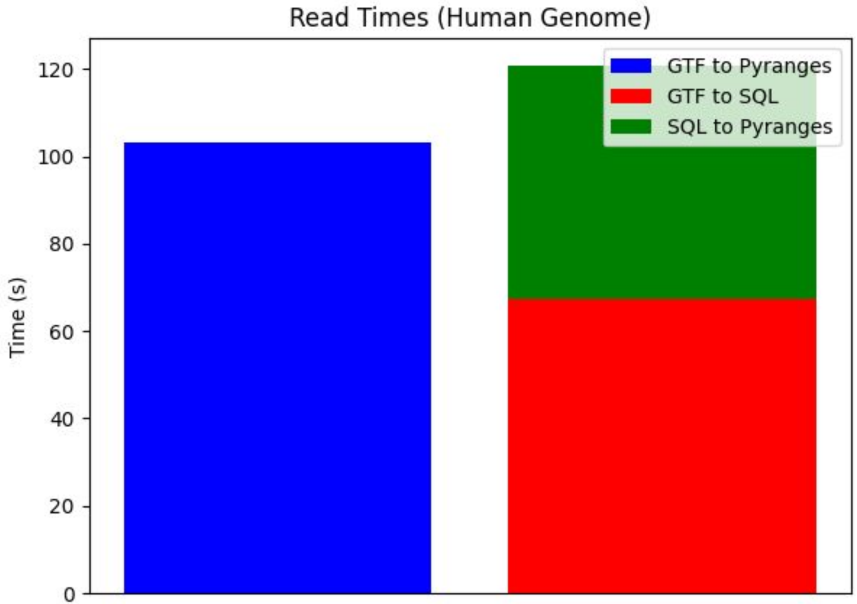
- One of the tools, Pyranges (written in Python), is very good at handling interval queries
- We can add more features to it!
- Specifically, we use a SQL database to store data and handle aggregate queries

Approach

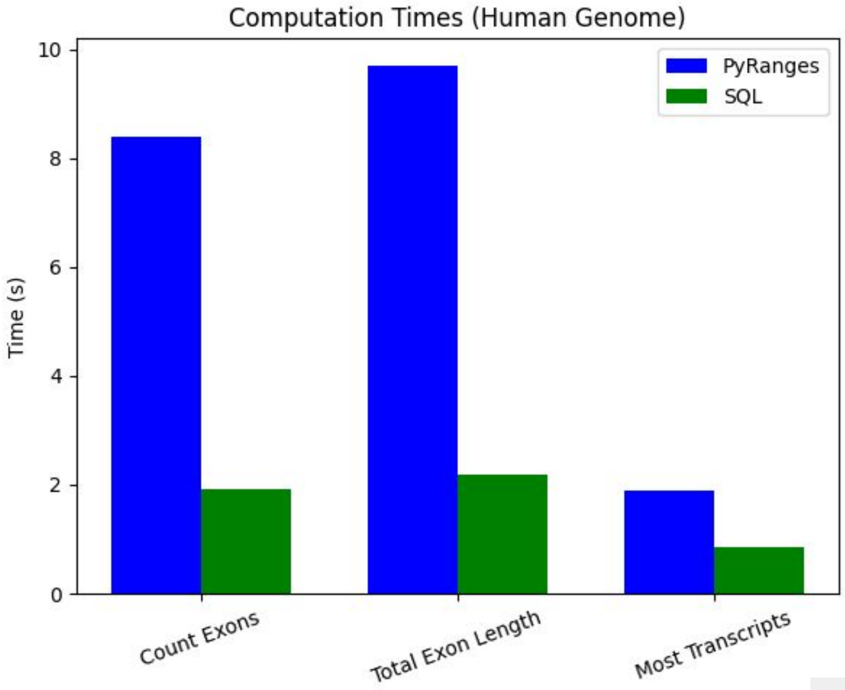
End users are not Software Engineers - abstract out all complicated details:

- Python wrapper around C++ code to convert GTF file to SQL database
- Multithreaded execution for faster processing time
- SQL queries wrapped by self-explanatory Python functions

Results



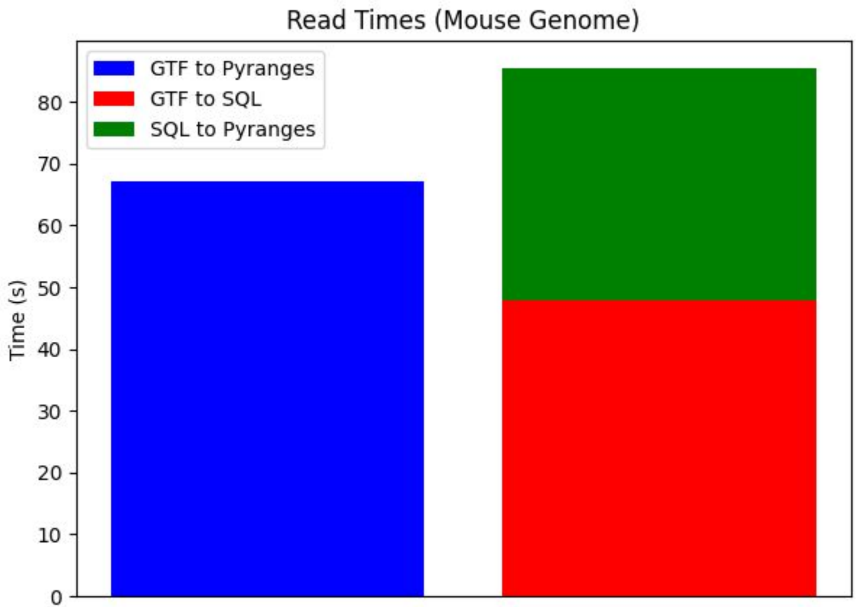
Read Times (Human Genome)

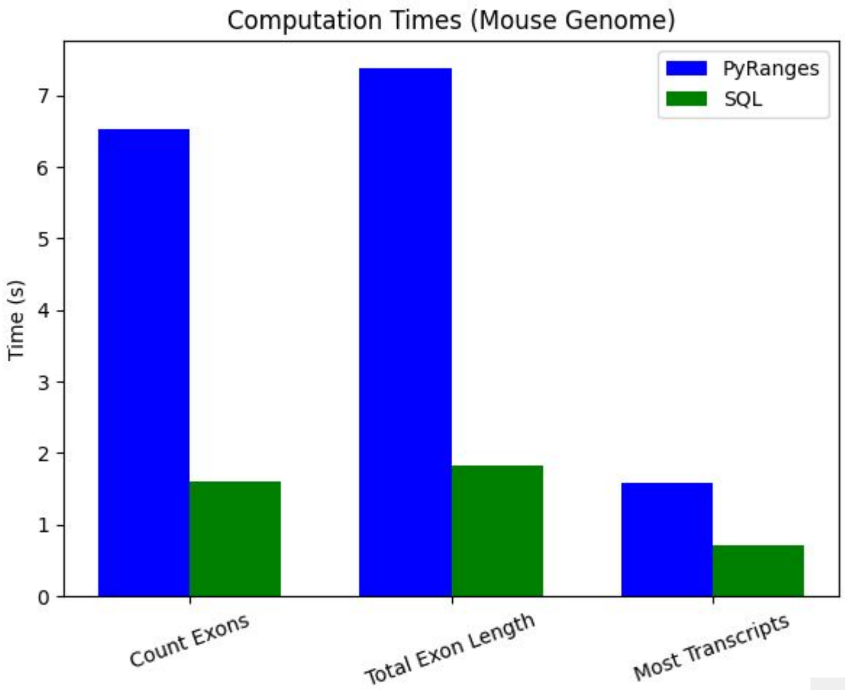


Computation Times (Human Genome)

Storage: GTF - 1.5 GB, SQL: 686 MB

Results





Storage: GTF - 1.1 GB, SQL: 581 MB

Conclusions

- SQL is space efficient
- Good at aggregate queries
- Faster to load than Pyranges object
- Takes slightly longer loading time if Pyranges object also required (say for interval queries)

Challenges

- Optimize multithreading hyperparameters
  - Number of producer and consumer threads
  - Batch Size for reading and writing
  - Choice of synchronization primitive
- Write Python wrappers for a lot of different queries
  - or find a way to automate it
- Merge with Pyranges or create an entirely different tool

Thank you!

Appendix

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Website: cse.iitb.ac.in/~dhano

Code used: https://github.com/DhanoHacks/Genomics-Tools-Analysis

Software Used: Python, C++, Jupyter, SQLite

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