

# ZygosDB: An efficient read-only database for Genome-Wide Association Studies (GWAS)

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## 1 INTRODUCTION & OBJECTIVE

- **Genome-Wide Association Studies (GWAS):** Identify associations between genetic variants and traits.
- **snpXplorer:** Website for visualising SNPs (single-base mutations) and SVs (multi-base mutations) in GWAS summaries.
- **Tabix:** Popular genomic indexing tool, currently in use by snpXplorer.
- **Challenge:** Querying genomic datasets is slow.
- **Solution:** Developed a custom specialized read-only database optimized for query throughput.

How can an efficient read-only database be designed and implemented for querying chromosomal, positional data?

## 2 METHOD

1. **Implement database in Rust**
  - Custom binary format.
  - Store all data sequentially.
  - Encode positions using variable-length integers.
  - Indices using B-trees for fast lookup.
2. **Optimise query throughput**
  - Compress database using Gzip or LZ4.
  - Resolve queries in parallel using multiple threads.
3. **Measure performance**



Figure 1: An example B-tree.

## 3 RESULTS

1

When returning small numbers of results, ZygosDB with Gzip compression is the fastest. As the window size increases, query throughput increases as well, with a not compressed ZygosDB database returning **up to 7 million rows per second**.

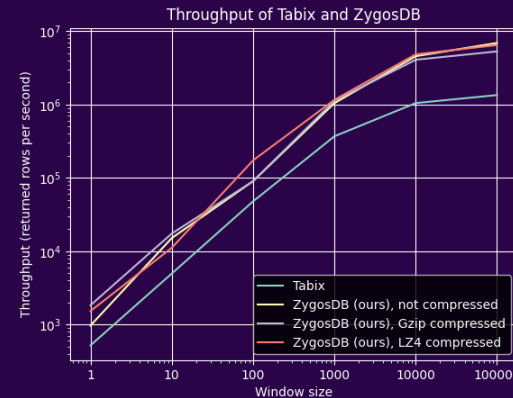


Figure 2: The query throughput, measured in the number of rows returned per window size, for both Tabix and our database.

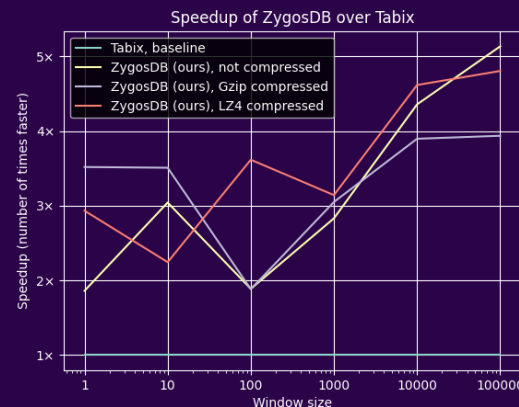


Figure 3: The speedup of ZygosDB over Tabix.

3

The maximum query throughput is reached when using **3-5 threads**. Using **more threads decreases performance**, eventually reaching throughputs lower than when using a single thread.

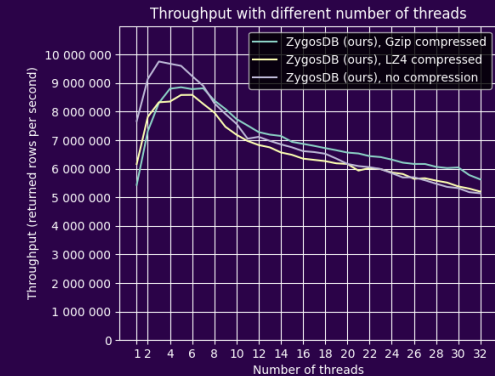


Figure 4: The multi-threaded query throughput with a window size of 100 000, measured with thread counts of 1-32.

2

In a **worst-case scenario**, ZygosDB is approximately **2 times faster** than Tabix. By increasing the window size, ZygosDB can reach throughputs that are **over 5 times faster** than Tabix.

## 4 CONCLUSION

ZygosDB, our read-only database, specialised for storing and querying genomic datasets, has a **2-5 times higher query throughput** than Tabix.