Persona: A High-Performance Bioinformatics Framework

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Agenda

• **Motivation**
• Bioinformatics Data and Tools
• Persona
  • AGD
  • Dataflow Engine
• Performance Results
Sequencing cost

Not a wet lab problem anymore ➔ IT / Systems problem
Implications

Need efficient systems that scale well
Agenda

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- **Bioinformatics Data and Tools**
  - Persona
    - AGD
    - Dataflow Engine
  - Performance Results

~300GB ~hours
What kind of data?

• Common sequencers produce *Reads*
  • Snippets of DNA → AACCGCTAGCGCGCTAGCTCGAGCTAGAA
  • 100-200 bases

@sequence name, metadata
ACGTTTCGATCGCGCCAGGAGGCTAG
+
-++* ' ) ) **55CCF@>>>>>>CCCCCCC

times a few hundred million ...
Alignment

Reference Genome

\[ \ldots TGACCTATAGCGATATAGCTTATTATTG\text{ }GGG-C\text{ }AAAA\text{ }ATGGAATCGAT\text{ }TGATCG\ldots \]

Mismatch

Read: \[ \text{TATTATTGGGATAAAA-TGG} \]

Insertion

Deletion

\text{times a few hundred million ...}

\text{~hours}
Aligned Reads

• Stored in SAM/BAM

```
read_name 16  chr12  85500011  70
18M    *       0       0
TTTTACACACATTTATCTC  CDDFAEEC>EDDFBDEED?FCC@
```

• Followed by
  • Duplicate marking
  • Sorting
  • Recalibrations, analysis (variant calling)

~hours
Data and Tool Issues

- FASTQ
- SAM/BAM
- BED
- VCF

...
Persona – Bioinformatics, Unified
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Aggregate Genomic Data

Storage Subsystem

Bases

Q-Scores

Metadata

Manifest

Header

Index

Data

compressed
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Dataflow

• Dataflow execution framework
  • Base on TensorFlow engine
  • But no machine learning

• Operators perform computation on AGD chunks
Dataflow

- Modularity
- Balance/tuning
- (bounded) Queueing
Fine-grained Threading

• AGD chunks optimized for storage
  • Too coarse for some tasks
• Split into subchunks
• Delegate to *executor* shared resource
  • Task queue + thread pool
Graph Construction

c = persona.read_chunk(path)

d = persona.decompress(c)

o = persona.align(d)

sess = tf.Session()

result = sess.run([o])
Persona Shell

$ persona align local -i hg19 data/my_agd.json
$ persona sort local data/my_agd.json
Distributed Computation

Client
$ persona client bwa-align

Queue Service

Server 0

Server 1

Server 1

Server N

Storage Subsystem
Current Features

• Import data from FASTQ/BAM/SRA, export to BAM
• Sequence alignment with BWA-MEM, SNAP
• Dataset sorting
• Duplicate marking
• Dataset statistics (samtools flagstat)
• Read coverage (depth)
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Evaluation -- Setup

• Focused on sequence alignment using SNAP
• Throughput in bases aligned per second
• Data
  • 223 million 101 base reads (~16GB)
  • AGD chunks of 100K records
• Hardware
  • 32X Ubuntu 16.04, 2x12 Xeon E5-2680v3 @ 2.5GHz
  • Data on 6-disk RAID0 and single spindle drive
  • 7 server Ceph object store for distributed execution
Evaluation -- Questions

• What are the bandwidth-saving effects of AGD?

• What is the overhead of the Persona framework?

• How well do Persona and AGD scale?
Performance – AGD

Significantly less I/O → more efficient use of HW, BW
Persona Overhead

Negligible overhead!
Scaling

Full dataset aligned in ~17 seconds
Scaling Limits

![Graph showing scaling limits with lines for Persona SNAP and Simulation. The x-axis represents the number of nodes, and the y-axis represents gigabases aligned per second. The graph includes a logarithmic scale on the y-axis. The lines show an upward trend as the number of nodes increases.]
Persona – Scalable Bioinformatics

Aggregate Genomic Data

https://github.com/epfl-vlsc/persona
backup
Performance – Sort and Dup. Mark

• **Sort**
  - By metadata or aligned location
  - 1.54x speedup over samtools
  - 5.15x speedup over Picard

• **Duplicate marking**
  - Same algorithm as samblaster
  - 3.73x faster than samblaster

• **Dataset stats**
  - 2x speedup

• **Coverage (depth)**
  - 2x speedup
Profiling
Read/Write Single Disk

![Disk Read Throughput (MB/s)](image1)

![Disk Write Throughput (MB/s)](image2)
Alignment

- Example: SNAP
- Build hash index of reference
- To align a read:
  - Hash a portion (seed)
  - Lookup
    - Evaluate each hit
    - Edit distance computation
- Cores align reads in parallel

TATTATTGGGATAAAATGGTTT

...TATTACTGGGAAAAATGGTTTATG...............

Reference Genome Index (40GB)

\[ \text{edit distance} = 34 \]
Shared Data

• Sometimes need to share data between ops
  • E.g. multi-GB index of reference genome

• Use TF session resource manager
  • [string, string] → refcount object

• Op can create objects, provide handle to other ops
Data Movement

• Tensors not amenable to bioinfo data
• Leverage TF shared resources
• Implement reusable buffers
  • Stable memory use
  • Avoid syscalls
Bioinformatics?

• Biology, computer science, math, statistics
• Started mid 90’s with Human Genome Project
• Broad field
  • Genomics, proteomics, systems biology

• This talk: Whole Genome Sequence (WGS) analysis
  • Reading the letters of your DNA (ATCG ...)

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