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

~300GB

~hours

Need efficient systems that scale well
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*Image of a machine with a DNA strand and a chip icon.*

- ~300GB
- ~hours
What kind of data?

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

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

times a few hundred million ...
Alignment

Reference Genome

\[ \text{...TGACCTATAGCGATATAGCTTATTATTGGG}-\text{AAAAATGGAATCGATGGATCG...} \]

Read: \[ \text{TATTATTGGG}\text{ATAAAA}-\text{TGG} \]

Mismatch

Insertion

Deletion

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

\sim\text{hours}
Aligned Reads

• Stored in SAM/BAM

```
read_name 16  chr12  85500011  70
18M  *  0  0
TTTTACACACATTATCTC  CDDFAEEC>EDDFFBCDEED?FCC@
```

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

~8 hours
Data and Tool Issues

FASTQ  SAM/BAM  BED  VCF

...
Persona – Bioinformatics, Unified
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Aggregate Genomic Data

- Bases
- Q-Scores
- Metadata

Storage Subsystem

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 = \texttt{persona.read\_chunk(path)}

d = \texttt{persona.decompress(c)}

o = \texttt{persona.align(d)}

\texttt{sess} = \texttt{tf.Session()}

\texttt{result} = \texttt{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

* single disk
Persona Overhead

Negligible overhead!

* RAID-0
Scaling

Full dataset aligned in ~17 seconds
Scaling Limits

![Graph showing the scaling limits for Persona SNAP and Simulation with Gigabases Aligned per Second on the y-axis and Number of Nodes on the x-axis. The graph includes a line for Persona SNAP and a line for Simulation. The y-axis ranges from 0 to 1e9, and the x-axis ranges from 0 to 100 nodes.]
Persona – Scalable Bioinformatics

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

• Dataset stats
  • 2x speedup

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

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

![Graph showing disk read and write throughput over time.](image-url)
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
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

BufferPoolOp

[container, name]
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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