TU Delft DELFT UNIVERSITY OF TECHNOLOGY

A Spark Framework for < $100, < 1 Hour, Accurate Personalized DNA Analysis at Scale

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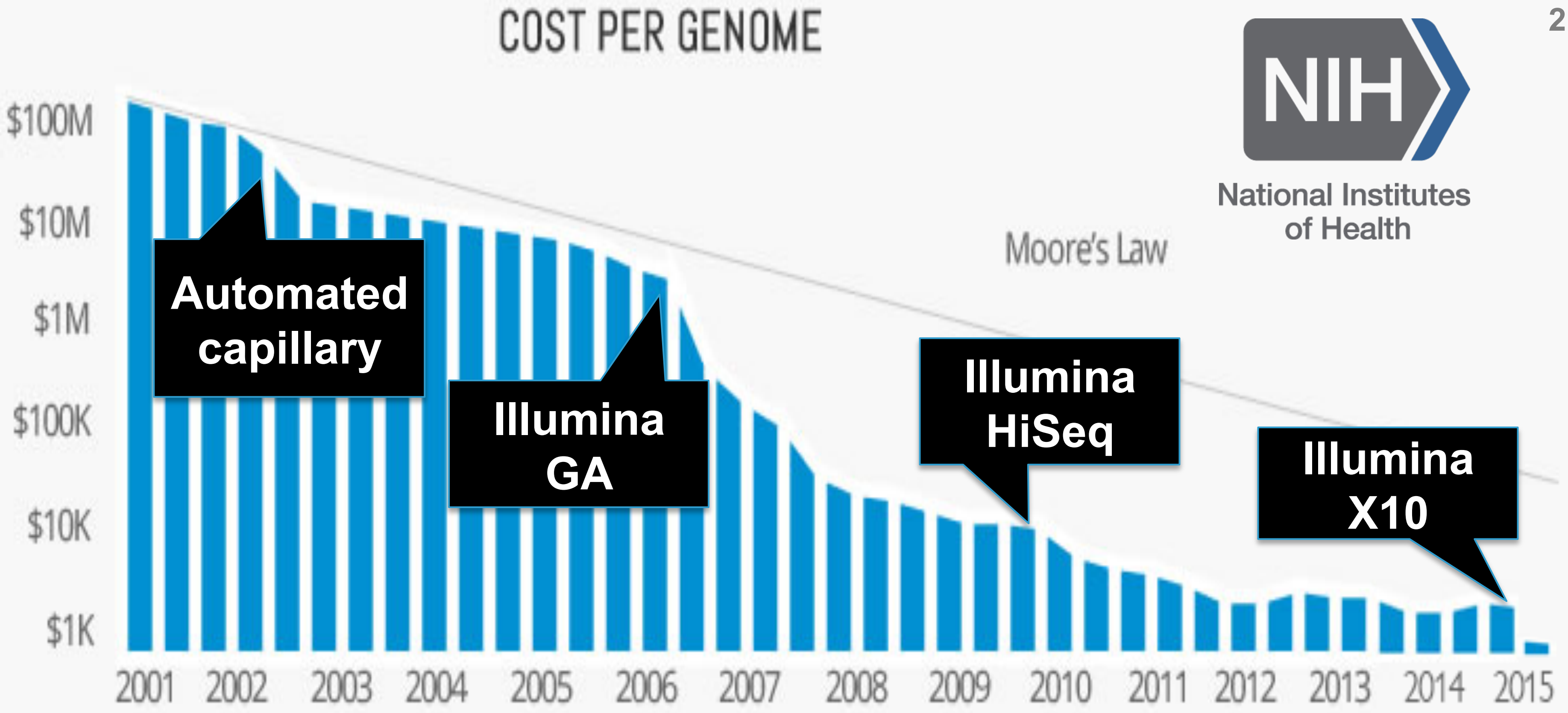
The Netherlands

Spark summit

SPARK SUMMIT 2016
DATA SCIENCE AND ENGINEERING AT SCALE
JUNE 6-8, 2016 SAN FRANCISCO



Automated capillary

Illumina GA

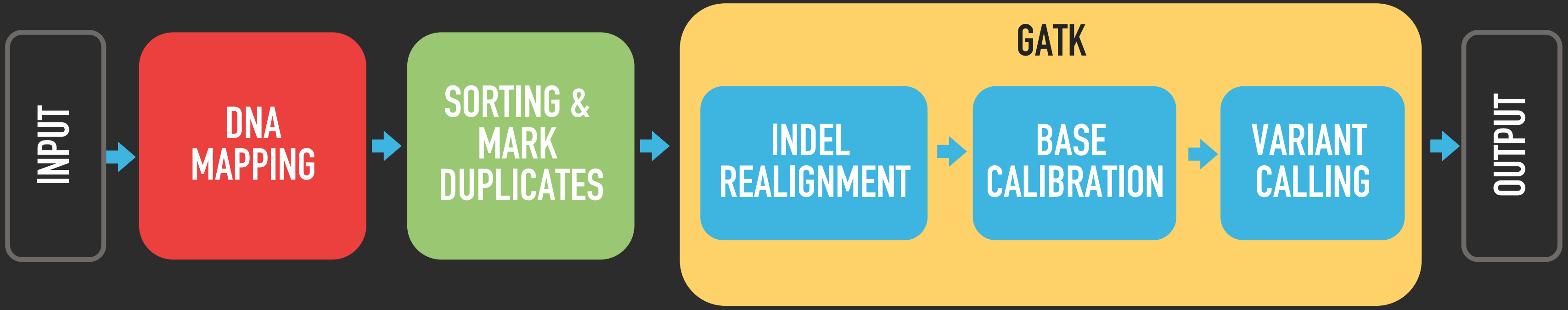
Illumina HiSeq

Illumina X10

National Institutes of Health

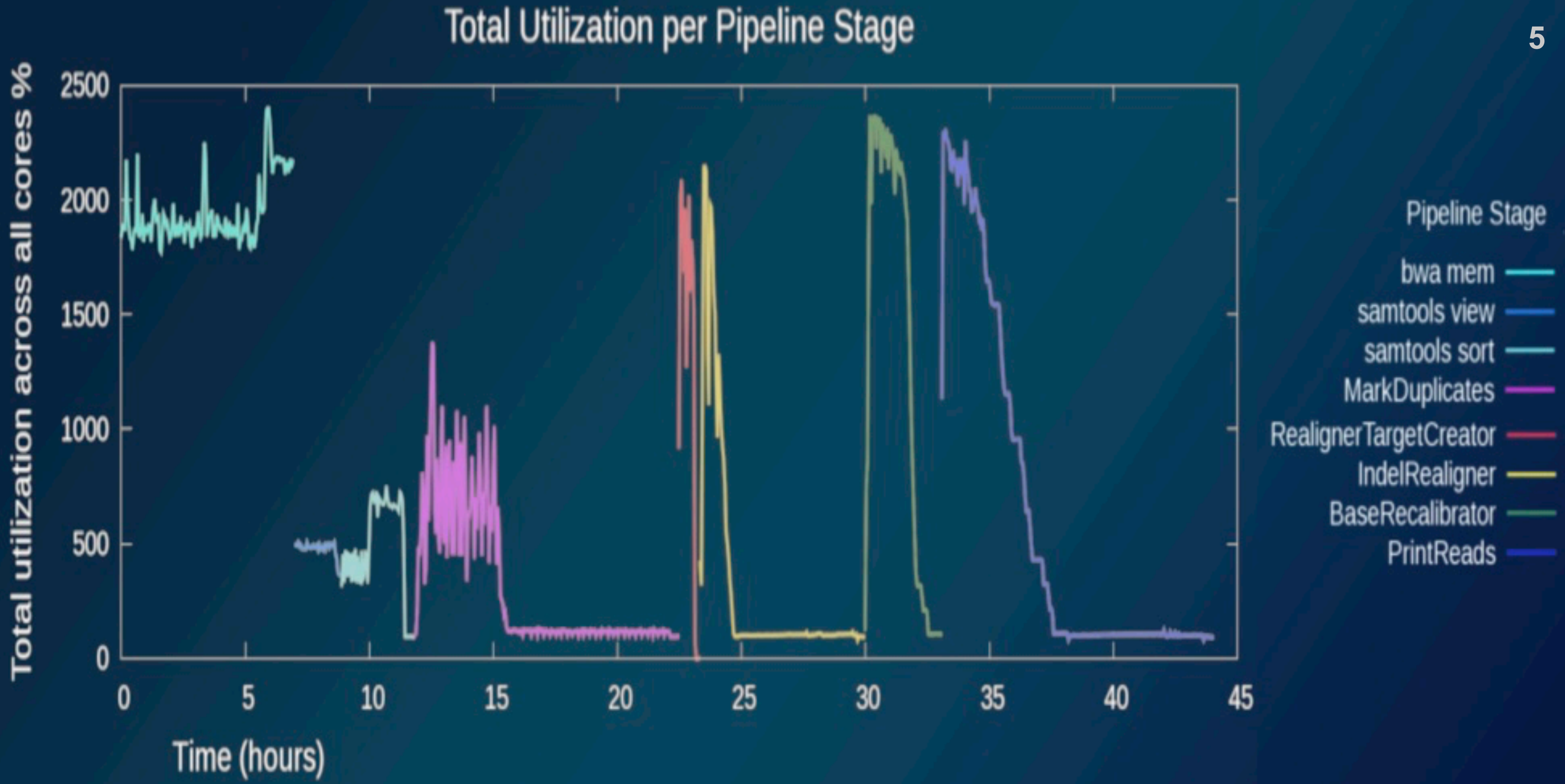
DNA SEQUENCING COSTS 10^5 LESS THAN 15 YEARS AGO!

HUGE DATA SETS REQUIRE INTENSE COMPUTING CAPACITY



STATE-OF-THE-ART DNA VARIANT DISCOVERY IS GATK USES A PIPELINE OF DIFFERENT TOOLS

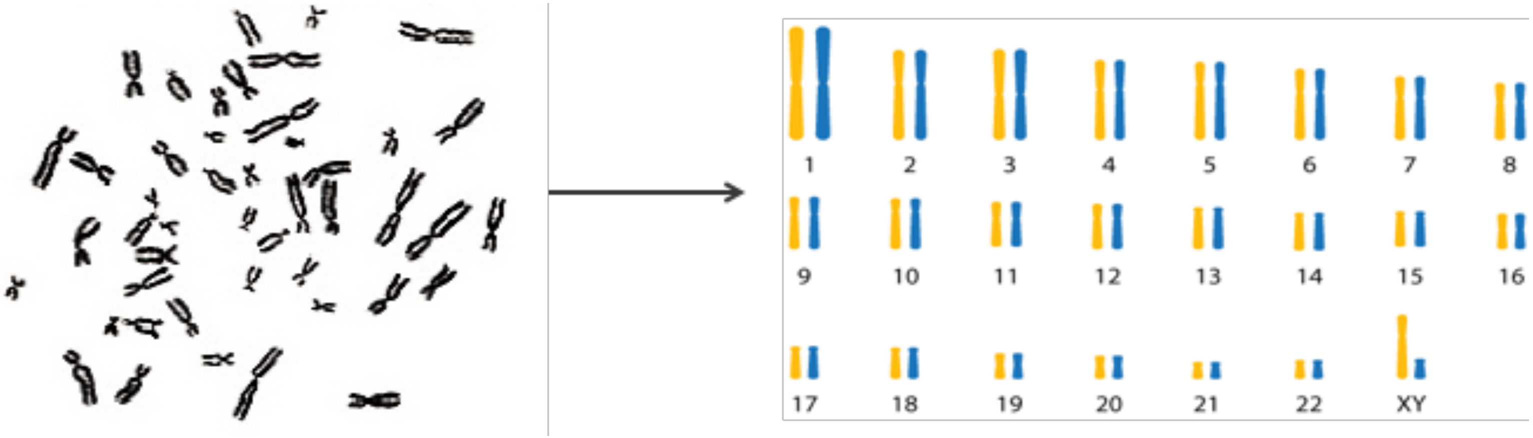
TUDelft



EXISTING PIPELINES DON'T EFFICIENTLY USE COMPUTERS

IDEA

- We use input data segmentation to group data of the same chromosome for parallelization.
- Data of the same chromosome can further be divided to create more segments.



COMPARISON OF BIG DATA TECHNIQUES

GATK Queue

- Slow scheduling
- Hard to setup
- Disk intensive
- Poorly documented

GRID ENGINE

Halvade

- Disk intensive
- Limited memory use
- Static load balancing

hadoop

Our Tool

- ✓ In-memory compute
- ✓ Dynamic load balancing
- ✓ Effective use of disk
- ✓ Easy to setup

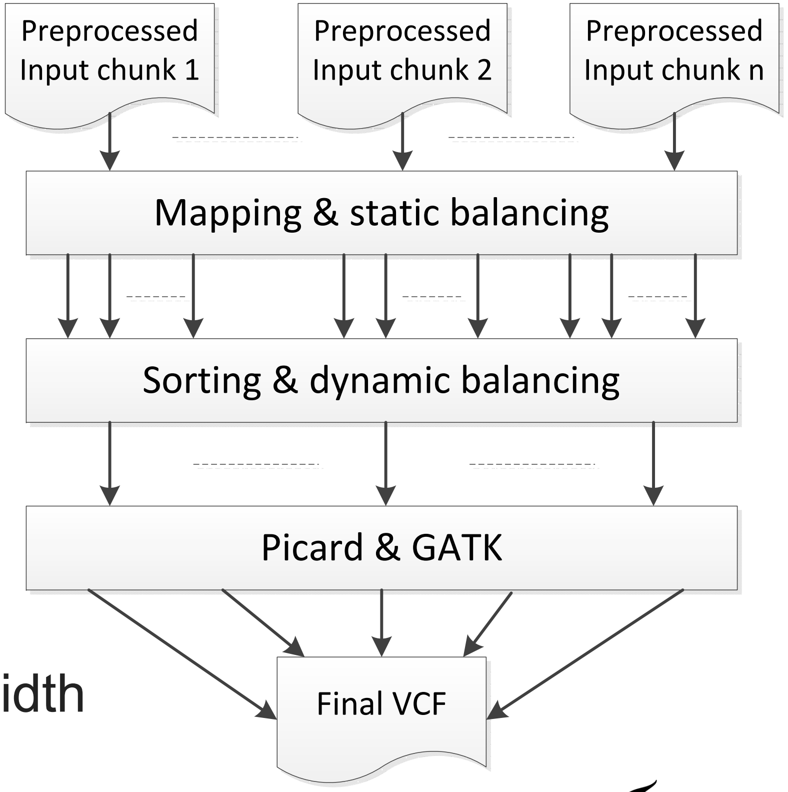
Spark™

ADAM

Lack of genomics verification of the outputs

IMPLEMENTATION

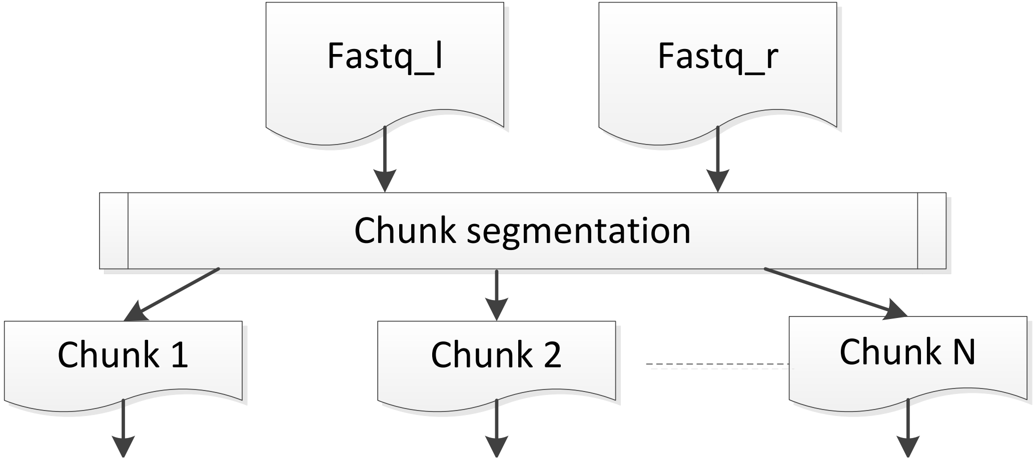
- Input is preprocessed into chunks
- Spark framework divided into 3 stages
  1. Mapping & static load balancing
  2. Sorting & dynamic load balancing
  3. Picard & GATK
- Challenges in memory capacity, processor utilization and network bandwidth


Preprocessed Input chunk 1
Preprocessed Input chunk 2
Preprocessed Input chunk n
Mapping & static balancing
Sorting & dynamic balancing
Picard & GATK
Final VCF

PREPROCESSING INPUT

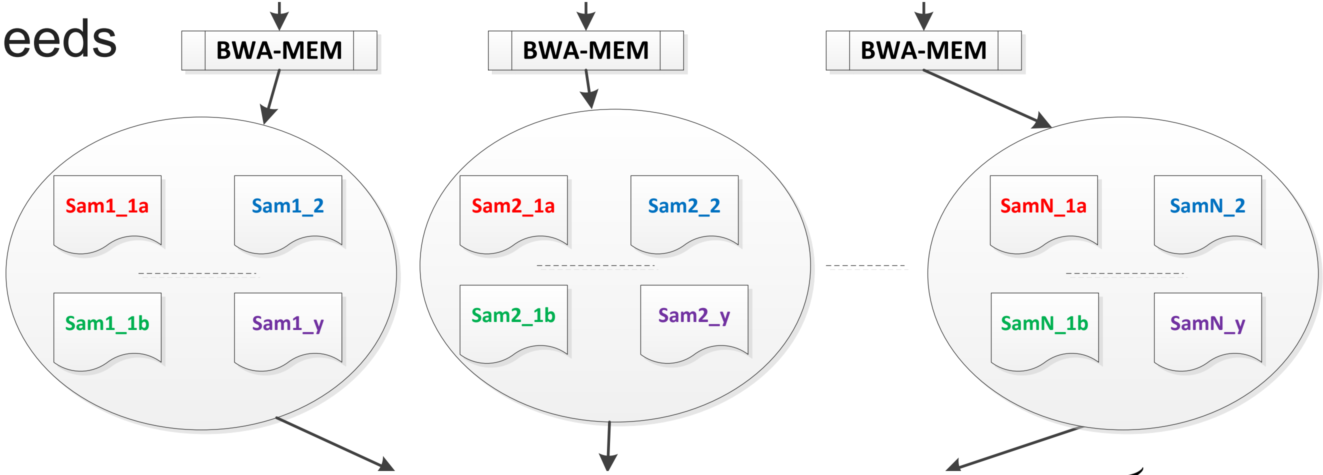
- Two input files are merged
- Divided into chunks
- No. of chunks depends on

  cluster size
- Performed once before data is processed
- Not included into pipeline timing calculation



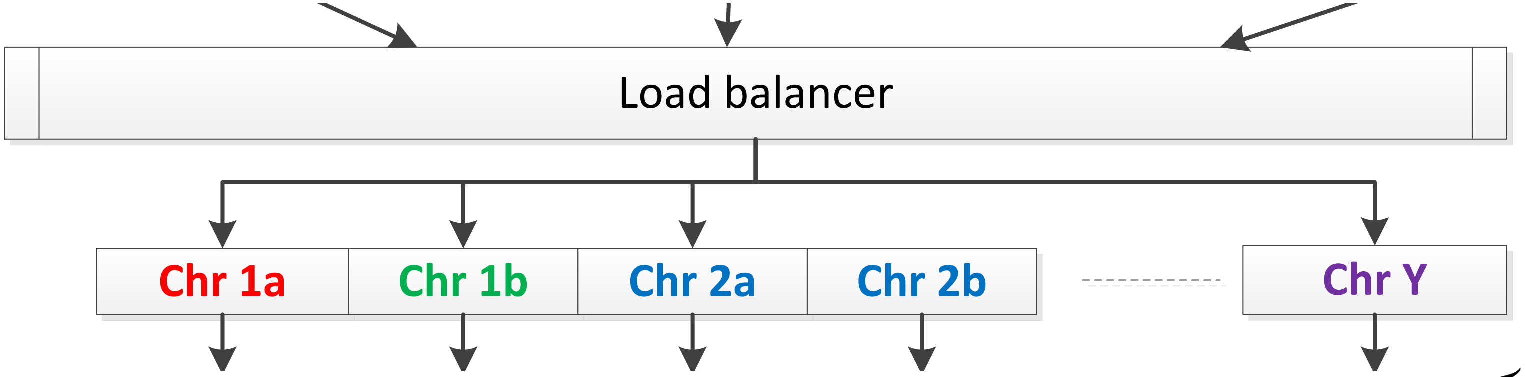
1. MAPPING & STATIC LOAD BALANCING

- Chunks mapped w/ BWA => easily scalable to all cores
- Output divided into chromosomal regions
- Reduces memory needs for dynamic load balancing



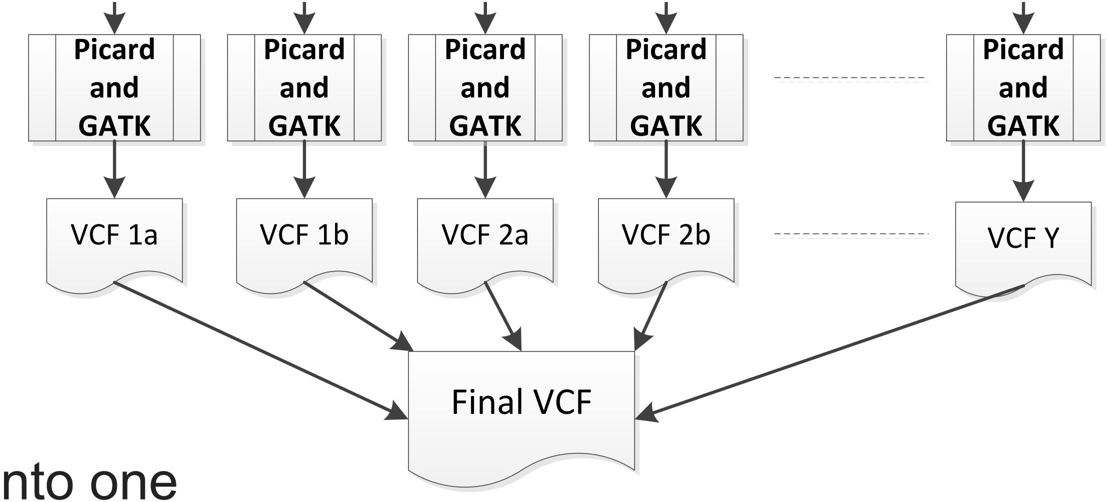
2. SORTING & DYNAMIC LOAD BALANCING

- Files of chromosomal regions are combined
- Files subdivided depending on # reads to ensure optimal load balancing



3. PICARD DEDUPLICATION & GATK VARIANT CALLING

- Many mini GATK pipelines run on region files
- # Spark containers depends on efficient core utilization
- Output VCF files combined into one output VCF file



EXPERIMENT SETUP

POWER8 Cluster

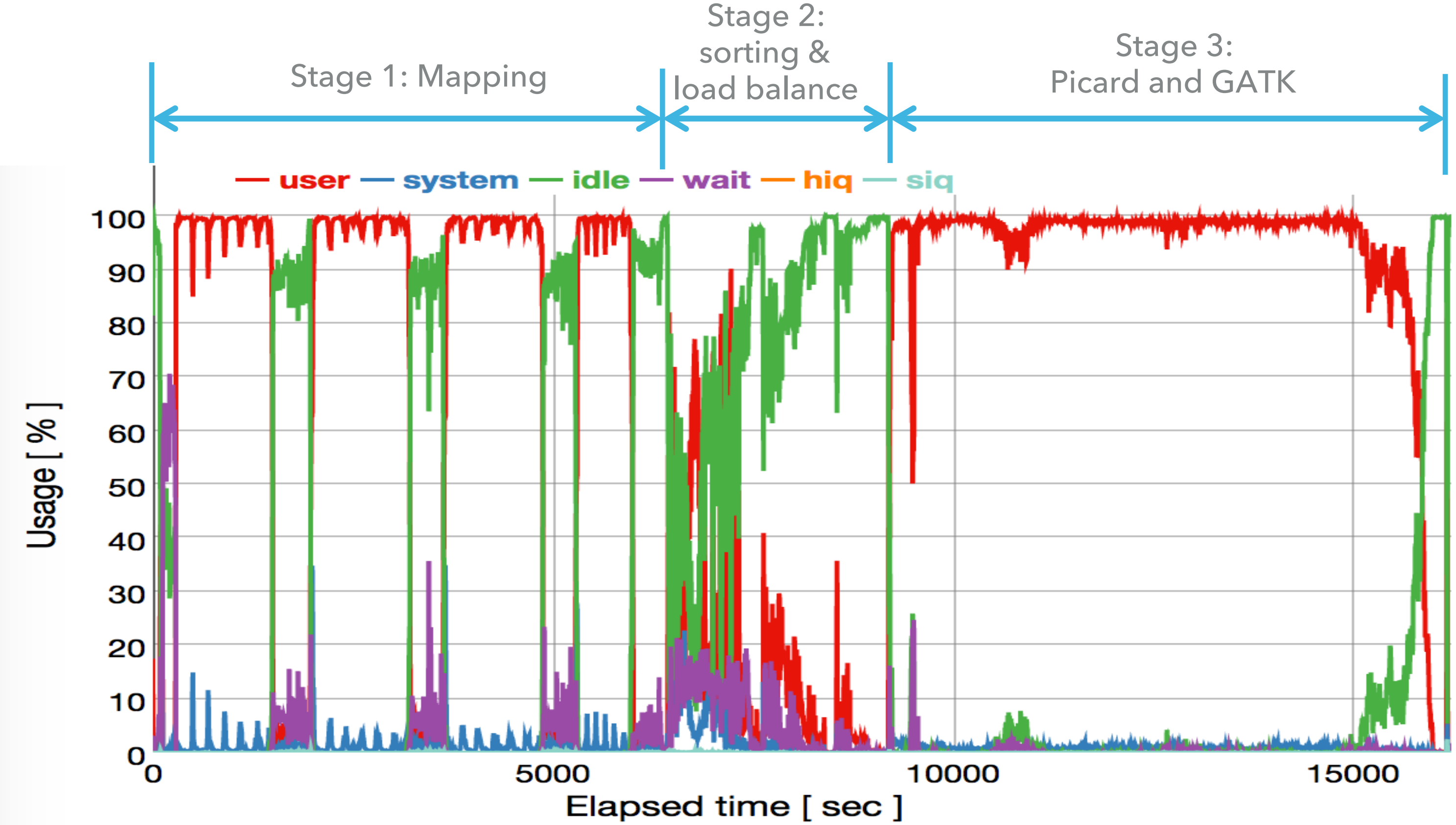
- 20 POWER8 S822LC nodes, bare metal
- 2x SCM 10-core, SMT8; 160 HW threads per node
- 512 GB of RAM per node
- Mellanox Infiniband EDR ConnectX-4 adapters (100Gb/s)
- IBM General Parallel File System (GPFS)

- Red Hat EL 7.2
- Spark 1.5.1, openJDK 1.8
- IBM LSF for resource management
- Configured as 1 master + 19 slaves

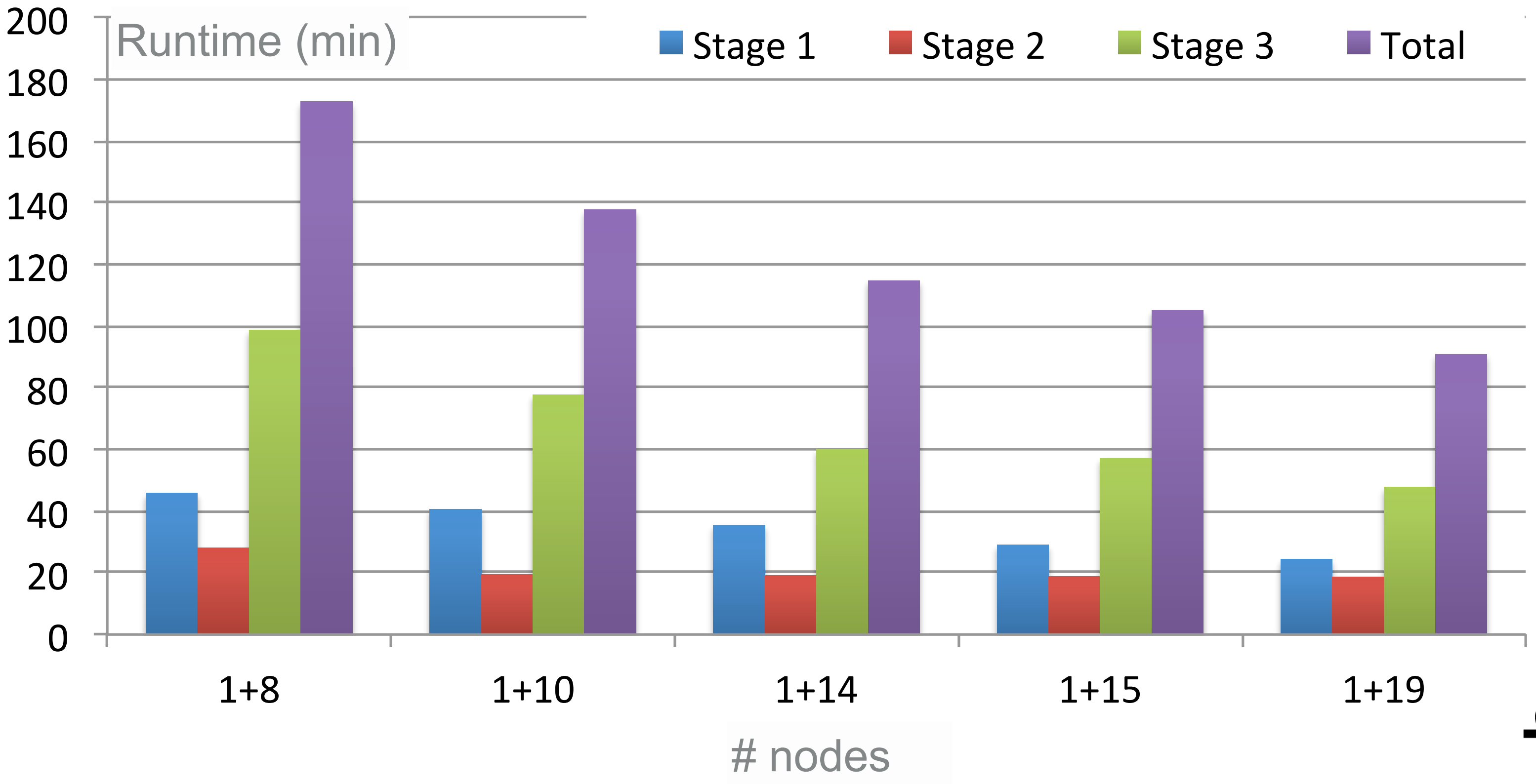
Data Used:

- Raw reads: G15512.HCC1954 (whole human genome, 400GB uncompressed)
- Reference: human_g1k_v37

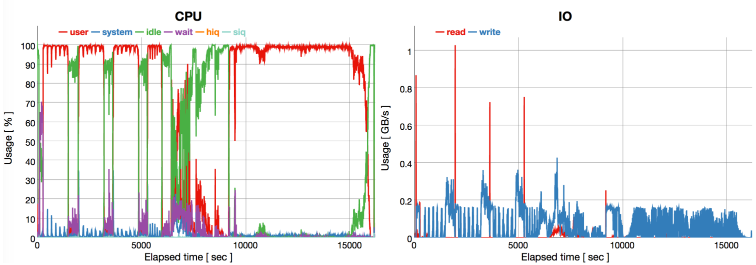
CPU UTILIZATION ON SMALLER CLUSTER

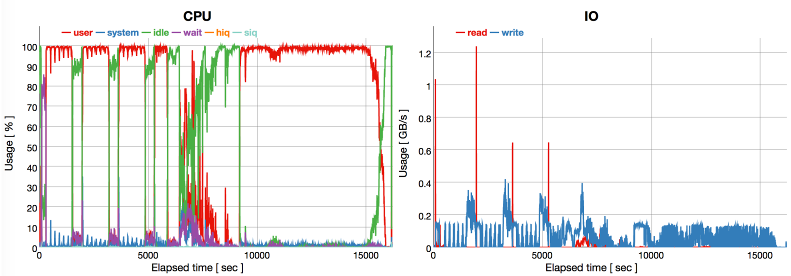


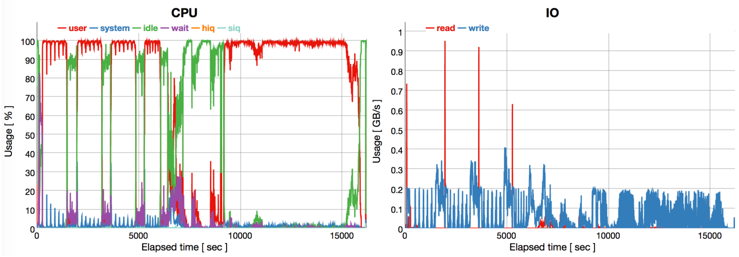
RUNTIME SCALABILITY ON 20-NODE POWER8 CLUSTER

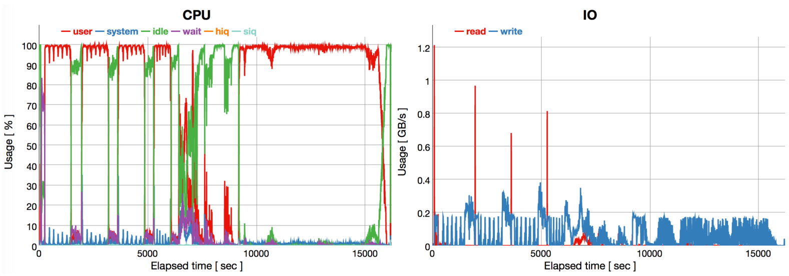


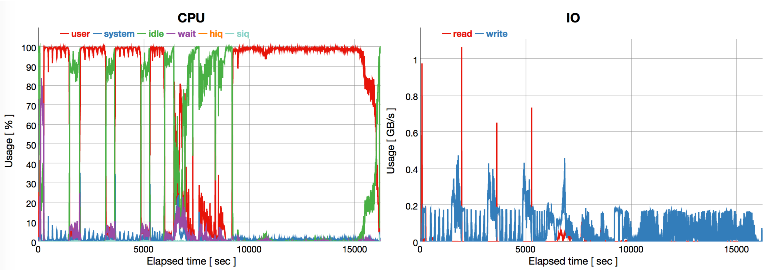
UNIFORM CPU UTILIZATION AND IO AMONG SIX NODES

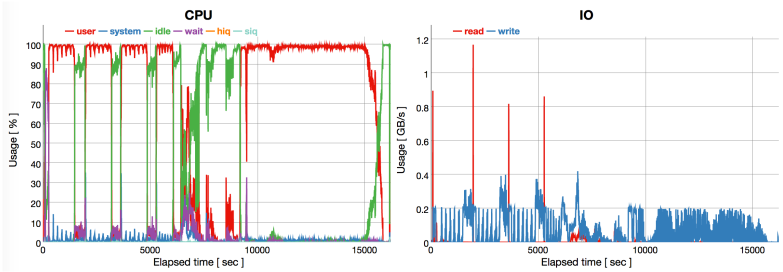






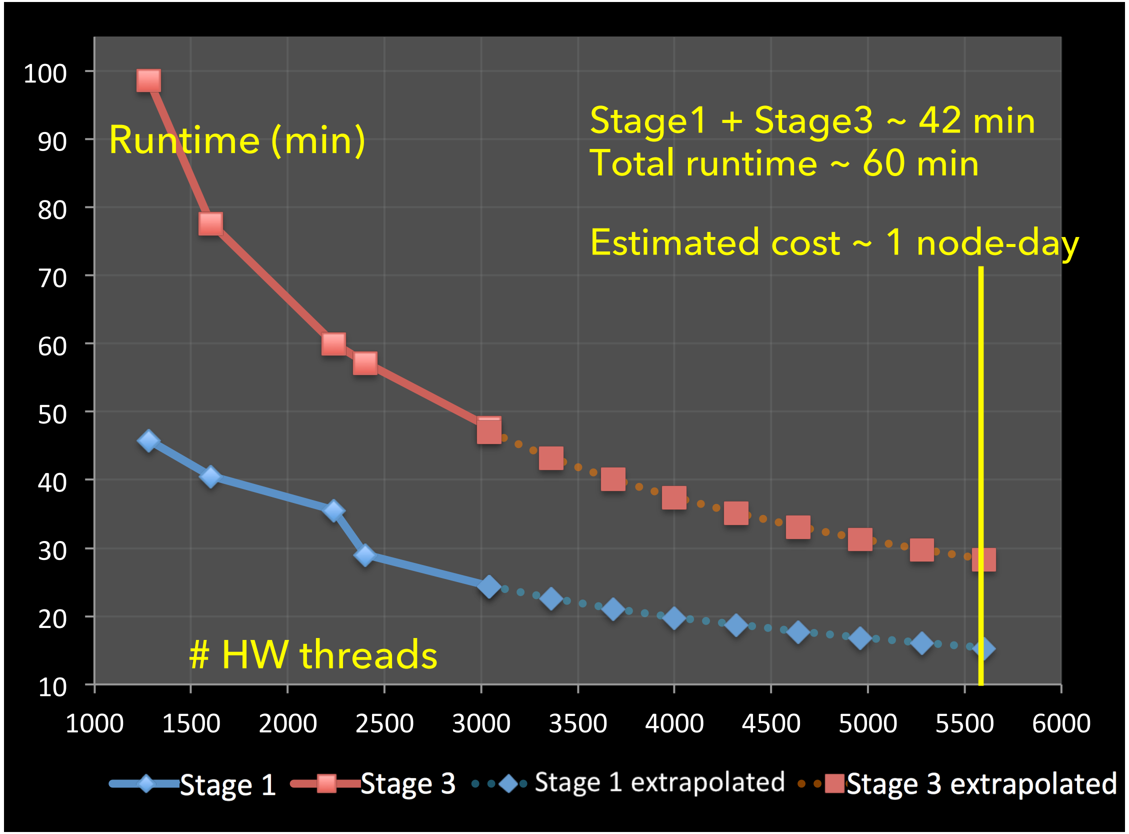




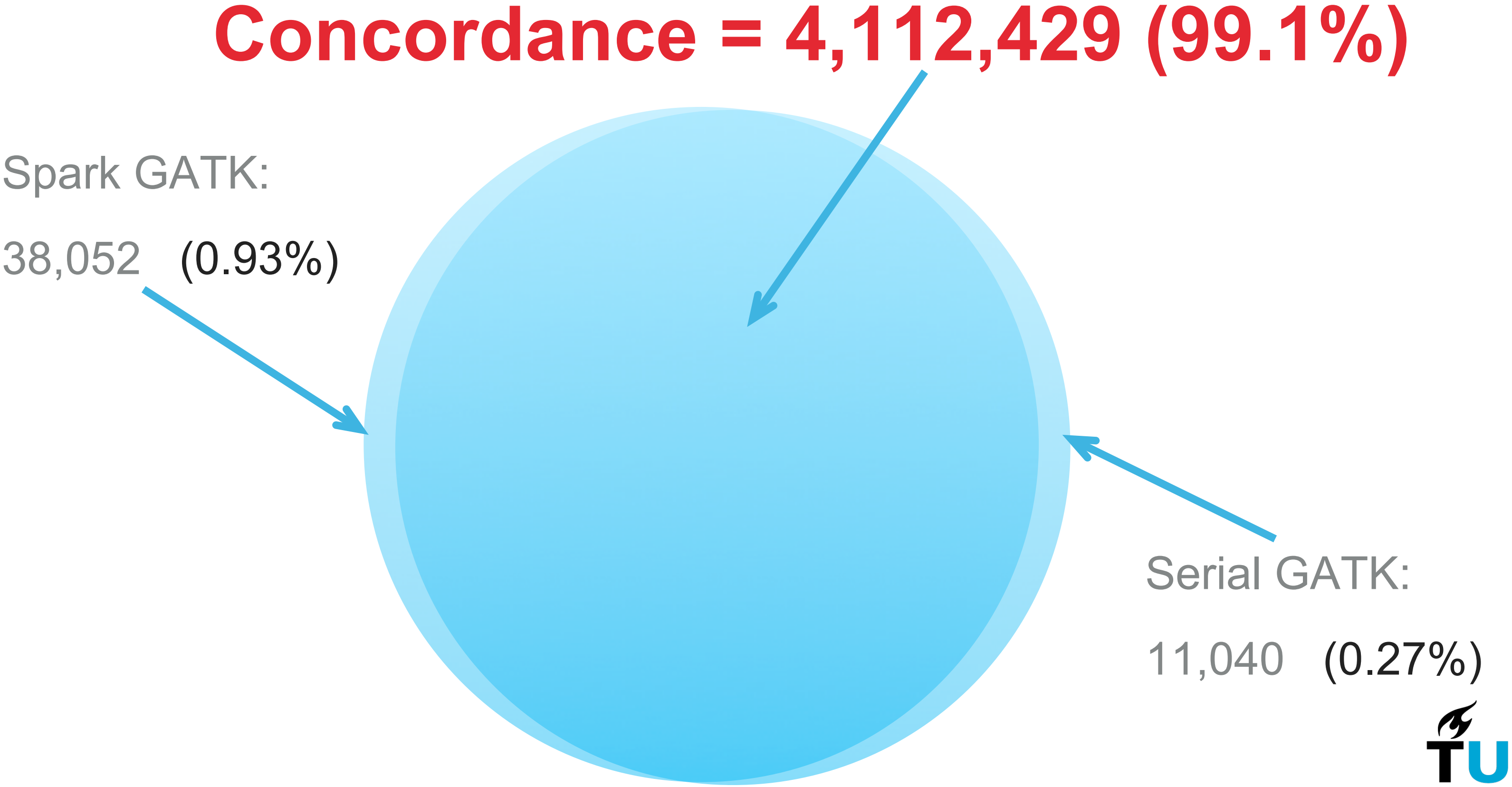


SCALABILITY ANALYSIS FOR LARGER CLUSTERS

- 90 min runtime on 20-node cluster
- Stage 1 & 3 are scalable
- Runtime scales down to 1 hour for 35 node cluster



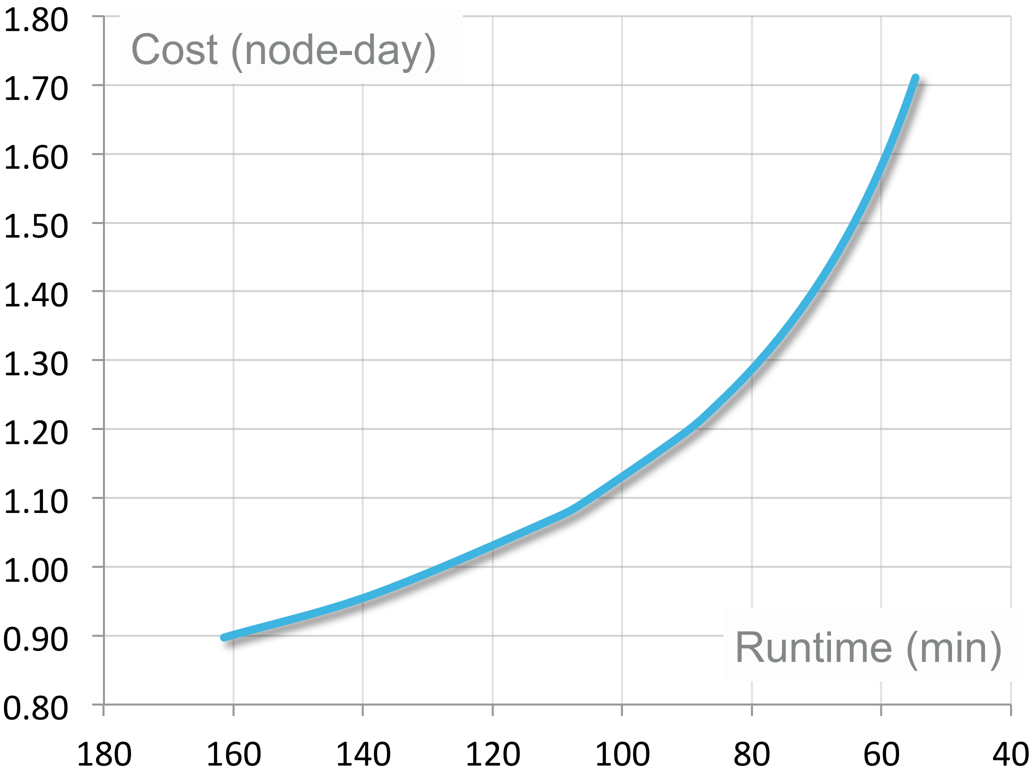
ACCURACY



Concordance = 4,112,429 (99.1%)
Spark GATK:
38,052 (0.93%)
Serial GATK:
11,040 (0.27%)

COST ESTIMATE

- GATK Spark allows efficient (i.e., cheap) CPU utilization
- 60 minute runs on 35-nodes cluster
- POWER nodes cost <$100 a day ($30-$70 in SoftLayer)



CONCLUSIONS

- GATK pipeline runs on Apache Spark framework
- Solution shows good scalability without sacrificing accuracy
- 400GB WGS data scales to under 1 hour for 35 nodes
- Price point is below $100 for POWER8 cluster
- Future work
  - We are accelerating our solution further using FPGAs through the IBM POWER8's CAPI interface
  - Examples: compression & math-heavy kernels like pairHMM

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THANK YOU.

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