Medical Big Data Analysis System to Discover Associations between Genetic Variants and Diseases

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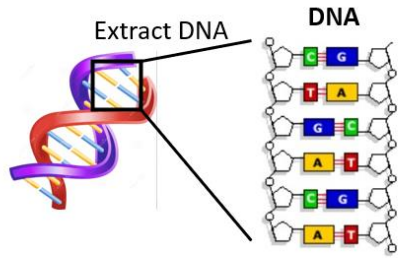
Background

- Health Record Data
  - Used to record data on patients
  - Biological measurements
    - Disease Diagnoses
    - Medical procedures
- Genetic Data
  - Retrieved from DNA in blood samples
- Marshfield Clinic Research Institute (MCRI)
- Genome-Wide Association Studies (GWASs)
  - Find genetic variants for certain diseases
  - Phenotype-to-genotype approach
- Phenome-Wide Association Studies (PheWASs)
  - Explore multiple diseases relevant to genetic variant
  - Genotype-to-phenotype approach

Name: Jane Doe
Medical History #: 111111
DOB: 01/01/1950
Weight: 150 lbs
Height: 5'5''
Address: 1000 N. Oak Street

Diagnosis & Procedure
(ICD9 codes):
250 = Diabetes
493.1 = Intrinsic Asthma
474.00 = Chronic Tonsillitis
28.2 = Tonsillectomy

Prescriptions:
Antibiotics
Albuterol
Metformin



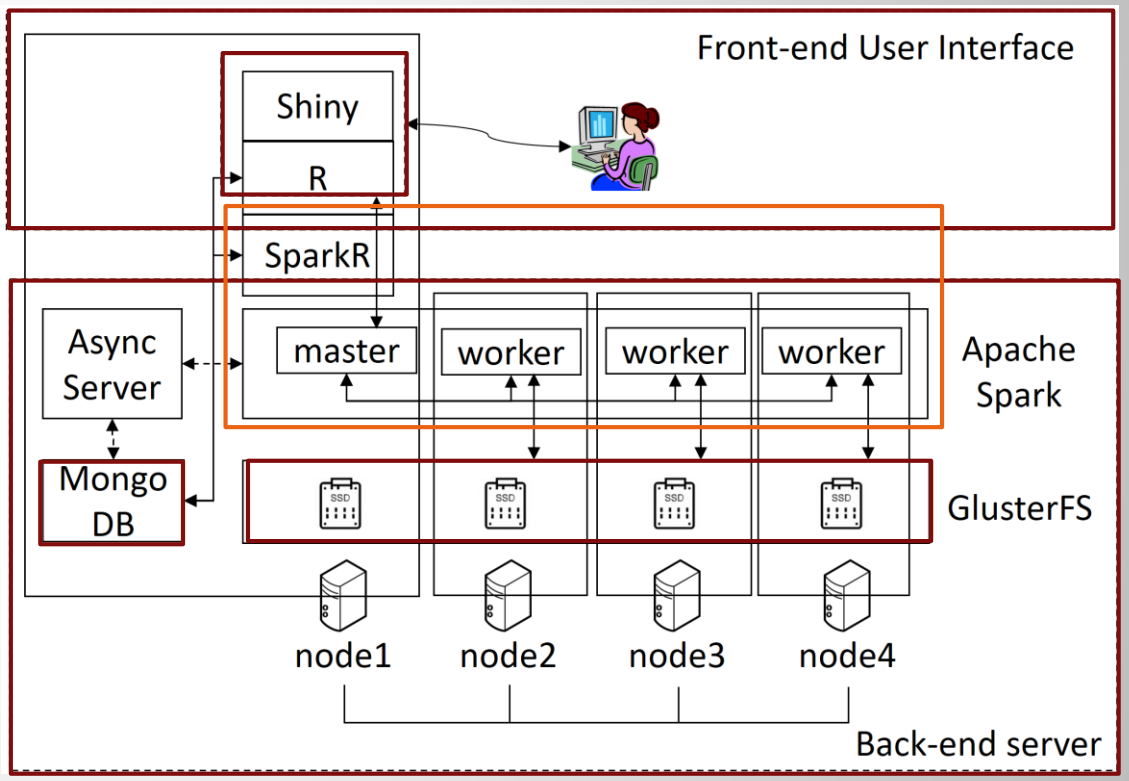
Person1: A–G–T–C–A–A–G
Without disease

SNP

Person2: A–G–T–A–A–A–G
With disease

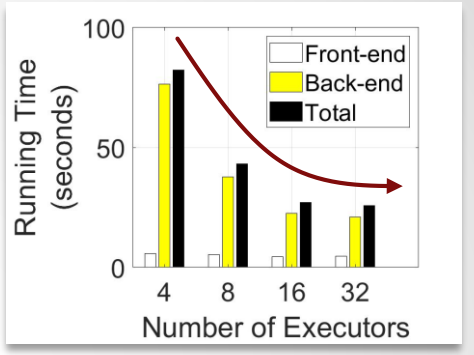
Architecture

- Web Query System architecture
  - Front-end user interface
    - R Shiny
  - Back-end server
    - GlusterFS, Spark, MongoDB, Java daemon
- Each node runs on
  - Dell PowerEdge R710
  - 2U rack sever (144GB)
  - 2 Intel Xeon 5660
- Each node has
  - 2 TB SSD

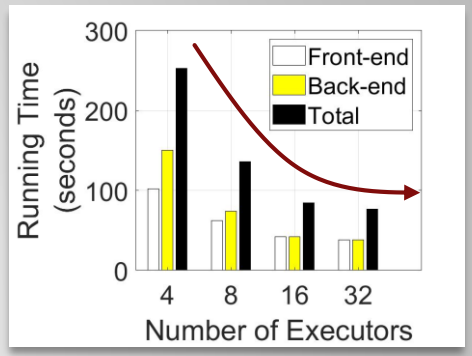


Evaluation

- SparkR (front-end), Spark-submit (back-end)
- Measured running time of:
  - Front-end & back-end operations
- Each executor: 2 CPU cores, 16 GB
- Varying the number of executors to 4, 8,16 and 32
- Running time for disease / genome data
  - Running time becomes faster with more executors on parallel processing
  - Running time of front-end is much less than back-end processing
  - Running time with 16 and 32 executors is similar, indicating the existence of upper bounds

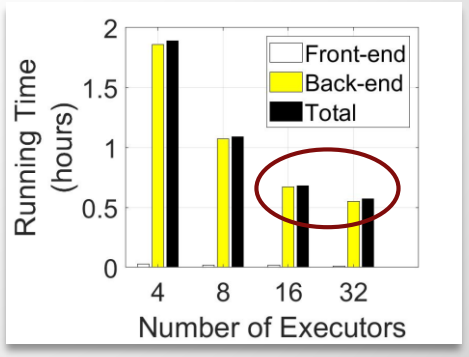


Chromosome 22

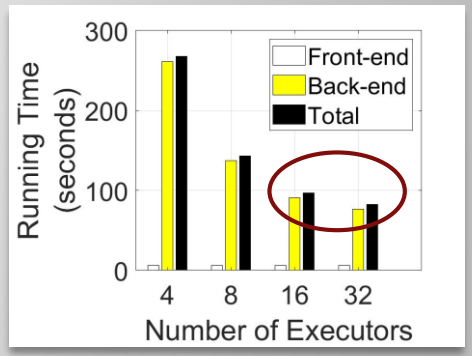


All chromosomes

Search disease



Chromosome 22



All chromosomes

Search genome

Thank you

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