Using Impala For Variant Pipelines

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11-19-2015

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Connect!

- GitHub tutorials and PowerPoint:

https://github.com/summerela/impala_training/

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Agenda

I. What is Impala?
   I. Use Cases
   II. System Structure
   III. What's available?
II. Connecting to Impala
   I. Hue Web Interface
   II. Impala Shell
   III. R
   IV. Python
III. Creating Queries
IV. Sample Pipelines
V. Getting Help

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Use Cases: Impala in the real world

- Locate and annotate variants based on:
  - Pathogenicity
  - Gene region
  - Allele frequency
- Clarity2 Challenge
  - ACMG actionable genes
  - Pathogenic
- Candidate Script
  - Centralized annotation of candidate variants
- Newborn Screening Genes
  - Rare variants
  - Pathogenic

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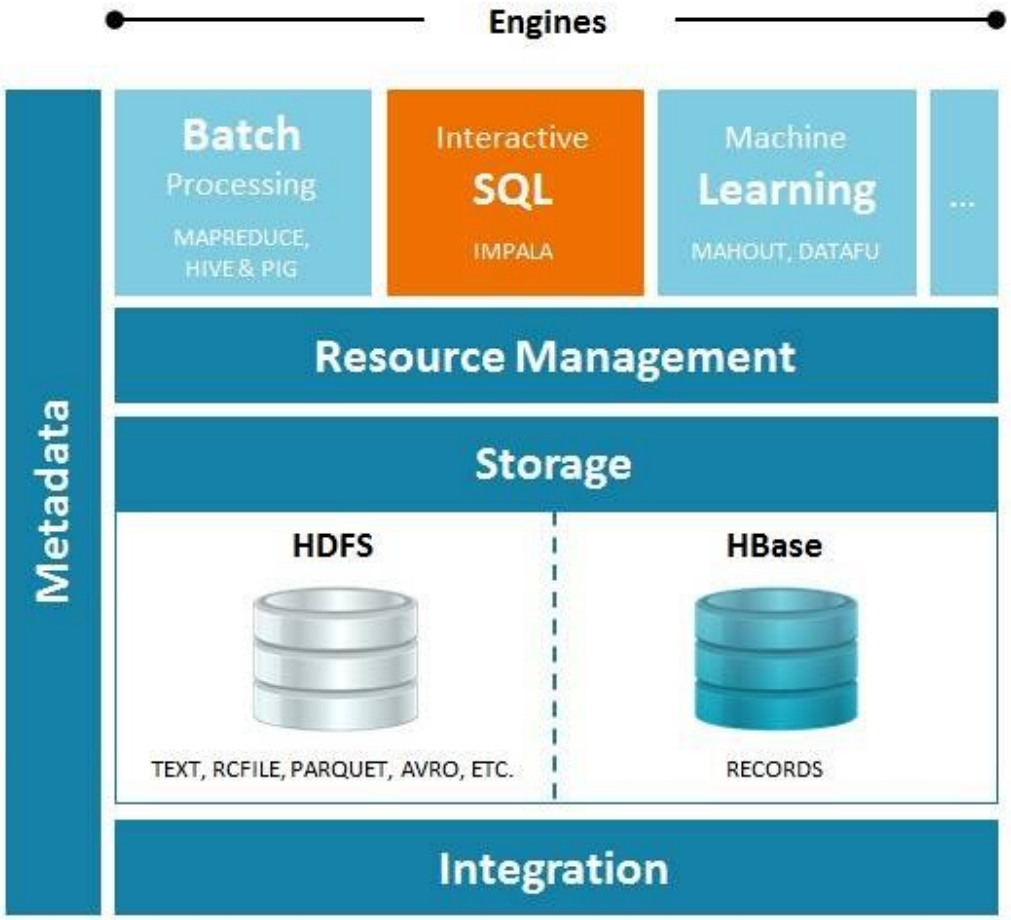
What exactly is impala?



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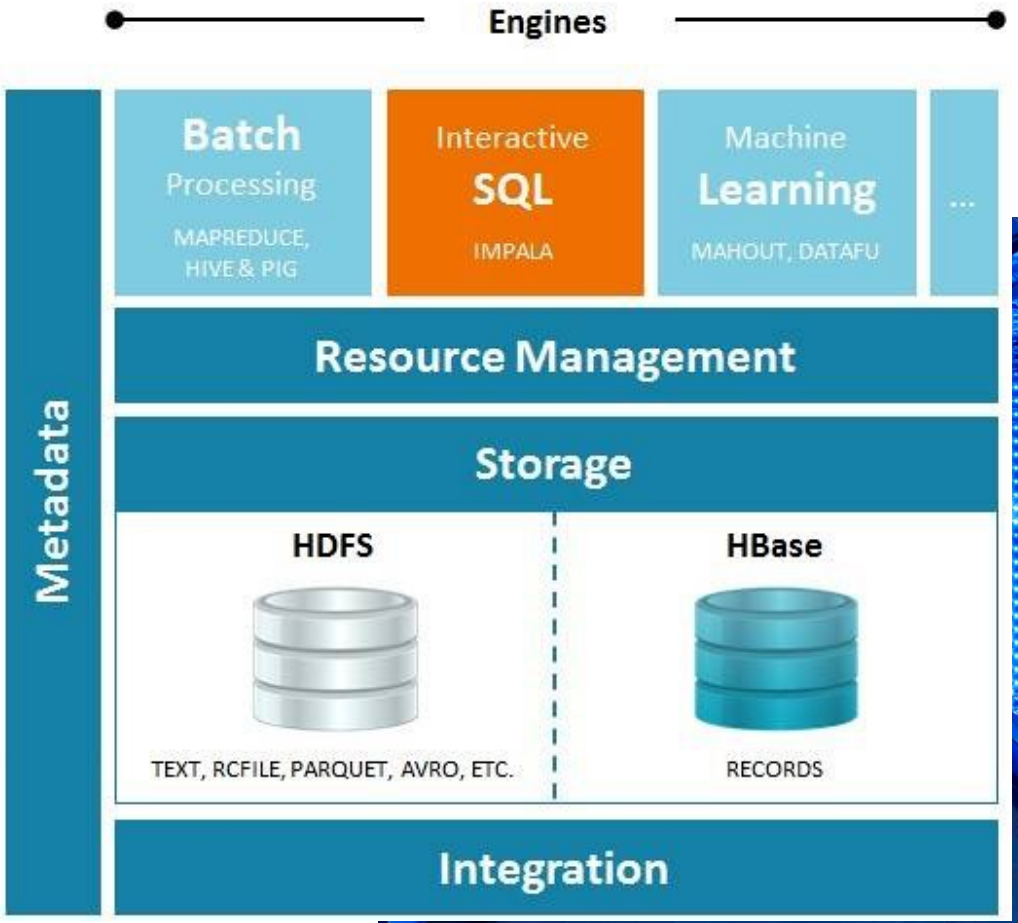

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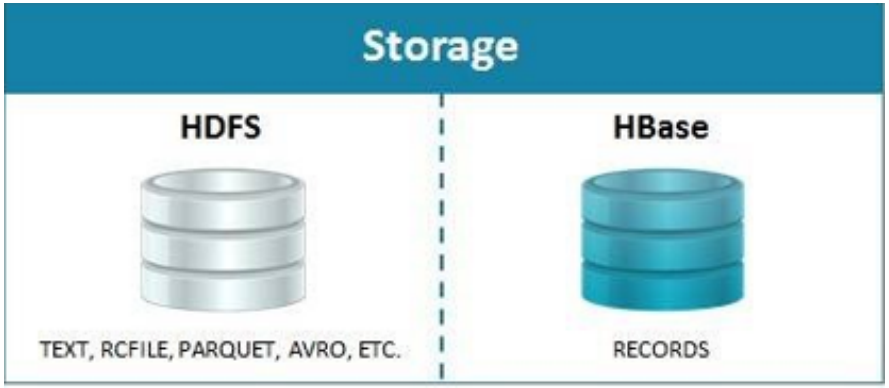

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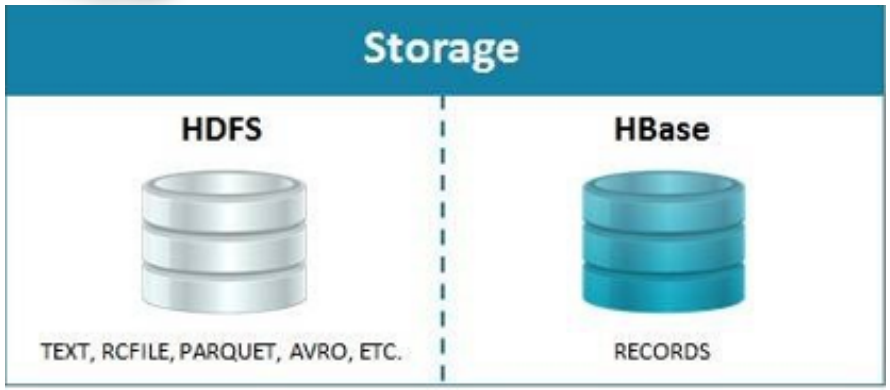
What exactly is impala?

Store Files:

- Variants
- Reference DB's
- Analysis results



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What exactly is impala?

Raw VCF file transformed and uploaded to HDFS:

```
##fileformat=VCFv4.1
##fileDate=20150819
##source=bin/makeVCF.pl
##reference=file:///proj/famgen/resources/Kaviar-150812-ISB/bin/../tabixedRef/hg19.gz
##version=<Kaviar-150812 (hg19)>
##kaviar_url=<http://db.systemsbiology.org/kaviar>
##publication=<Glusman G, Caballero J, Mauldin DE, Hood L and Roach J (2011) KAVIAR: an accessible system for testing SNV novelty. Bi
oinformatics, doi: 10.1093/bioinformatics/btr540>
##INFO=<ID=AF,Number=A,Type=Float,Description="Allele Frequency">
##INFO=<ID=AC,Number=A,Type=Integer,Description="Allele Count">
##INFO=<ID=AN,Number=1,Type=Integer,Description="Total number of alleles in data sources">
##INFO=<ID=END,Number=.,Type=Integer,Description="End position">
##INFO=<ID=DS,Number=A,Type=String,Description="Data Sources containing allele">
#CHROM  POS     ID      REF     ALT     QUAL    FILTER  INFO
1       10001   .       T       C       .       .       AF=0.0000384;AC=1;AN=26028;DS=SS6004475
1       10002   .       A       C,T     .       .       AF=0.0001153,0.0000384;AC=3,1;AN=26028;DS=HGDP00927|HGDP00998|HGDP01284,HGDP0
1029
1       10002   .       A       AT      .       .       AF=0.0000384;AC=1;AN=26028;DS=HGDP00521
1       10003   .       A       C,T     .       .       AF=0.0000384,0.0000768;AC=1,2;AN=26028;DS=HGDP01284,HGDP00521|HGDP00927
1       10004   .       C       A       .       .       AF=0.0000384;AC=1;AN=26028;DS=HGDP01284
1       10018   .       C       T       .       .       AF=0.0000384;AC=1;AN=26028;DS=HGDP00998
1       10019   rs775809821     TA      T       .       .       AF=0.0000384;AC=1;AN=26028;END=10020;DS=Malay
```

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What exactly is impala?

Read as a table by impala:

Data sample for kaviar

View in Metastore Browser

chrom	pos	stop	rs_id	ref	alt	qual	filter	allele_freq	allele_cnt	allele_num	sources
10	98917931	None	rs75578462	C	T	None	None	0.053711399436	1398	26028	ADNI|GMIAK9|GS000009930|GS0000117
10	98917981	None	rs545362632	C	T	None	None	0.000230499994359	6	26028	phase3-MSL
10	98917985	None	rs17112469	T	C	None	None	0.106577500701	2774	26028	!Gubi|ADNI|Desmond Tutu|GMIAK6|GMIA
10	98918018	None	rs746271737	G	A	None	None	3.84000013582e-05	1	26028	UK10K
10	98918073	None	rs756450057	T	C	None	None	3.84000013582e-05	1	26028	UK10K

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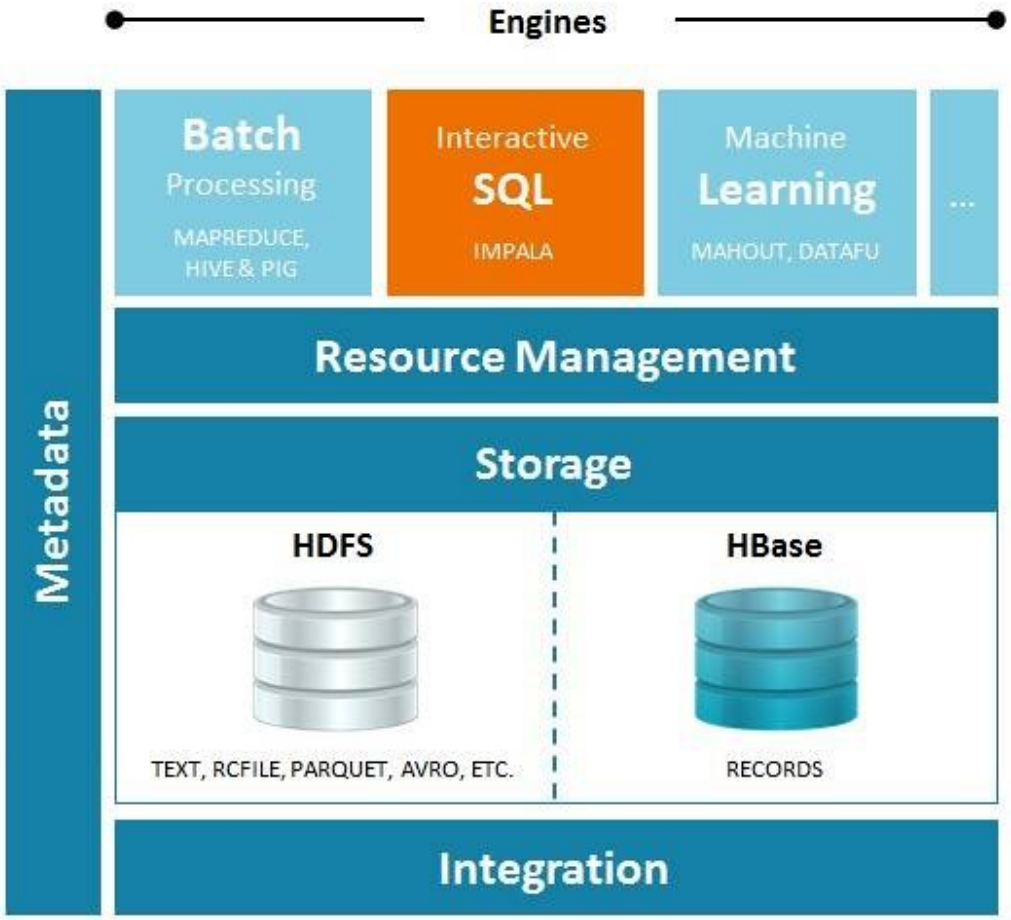

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What exactly is impala?

What exactly is impala?

SQL – Structured query language

SELECT columns
FROM database.table_name
WHERE parameters

What exactly is impala?

Interact with impala to:

- Filter
- Locate
- Annotate

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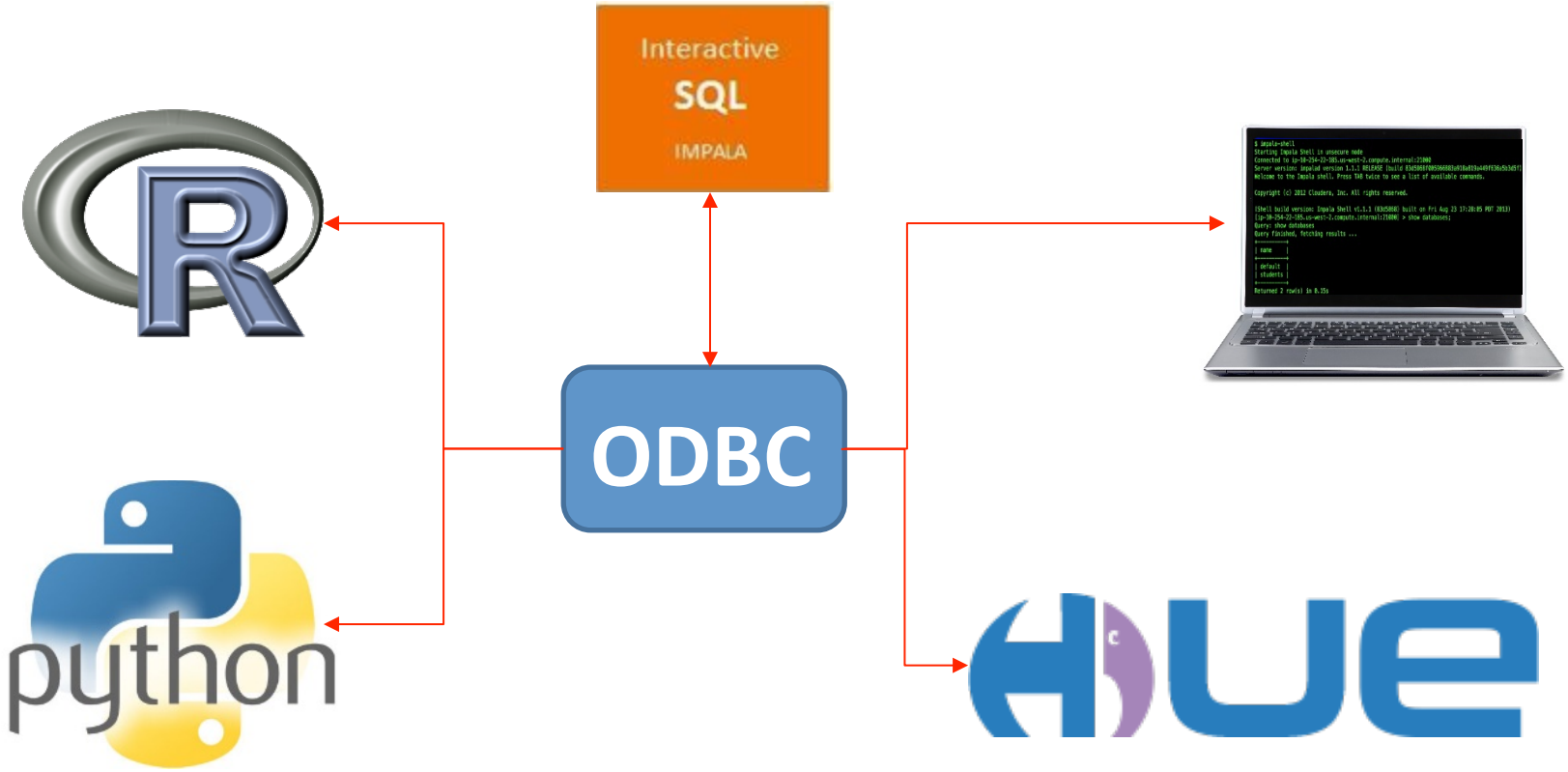
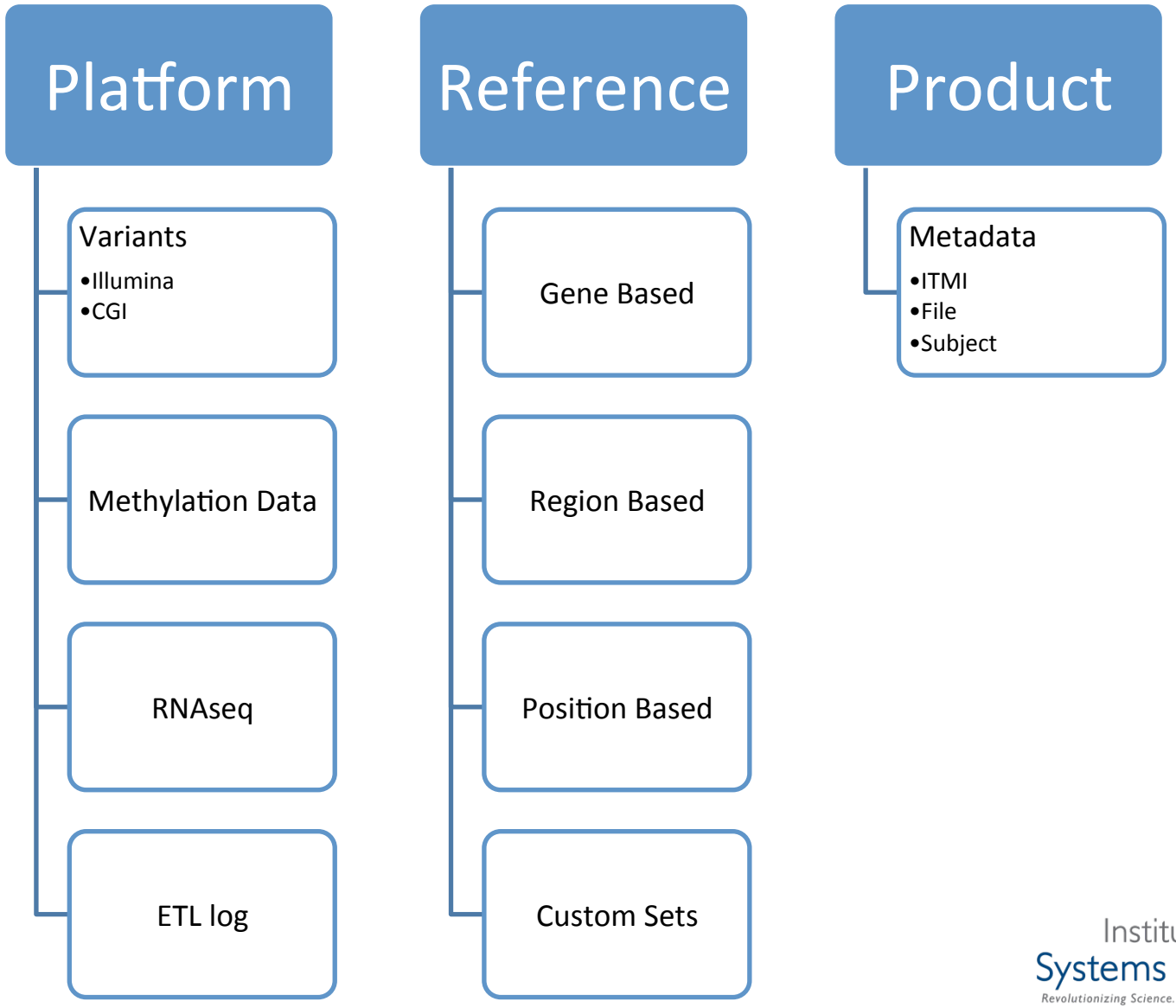
What exactly is impala?



Database Structure



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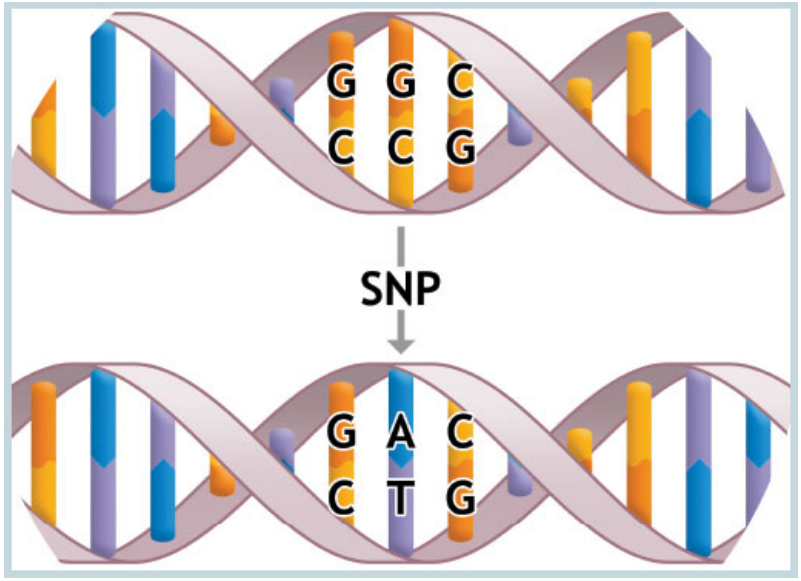
Benefits

- Consistency
  - Normalized Data
  - 1-based coordinates
  - Universal Column Names
- Speed
  - Quickly run queries across thousands of genomes

What's Available: Variants

Complete genomics

illumina®



101

102

103

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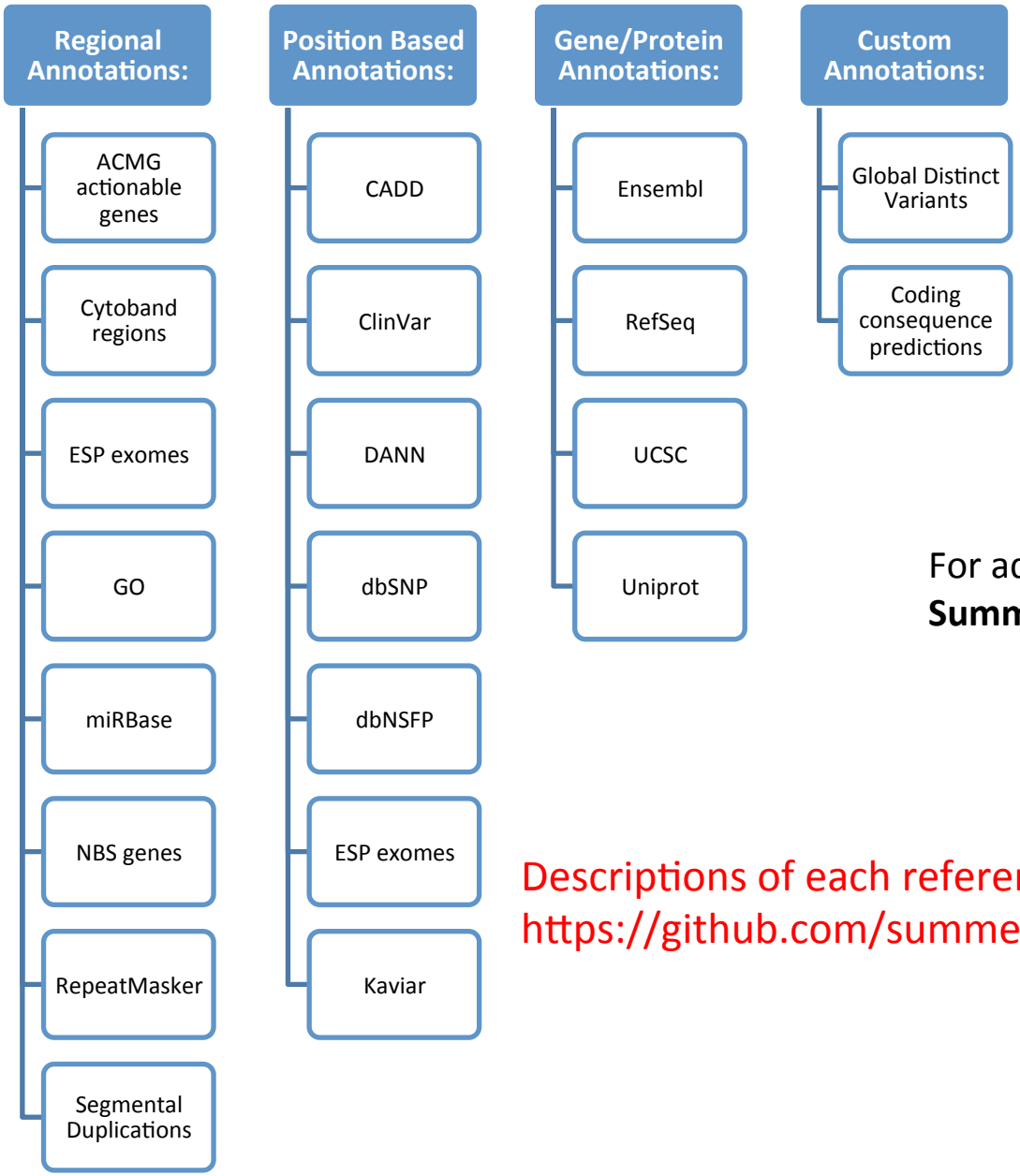
Reference Sources



For additional annotations email:
Summer.Elasady@systemsbiology.org

Descriptions of each reference:
https://github.com/summerela/external-reference-data-catalog

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Accessing Impala

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Accessing Impala

cloudera® IMPALA

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Accessing Impala

cloudera® IMPALA

Pros:

- Easy, web based, quick
- Easy to do query testing and EXPLAIN plan

HUE

Cons:

- Cuts off results inconsistently at 100,000 rows
- AJAX connection issues:
  - Connection drops
  - Canceling queries

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Accessing Impala

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Accessing Impala

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Pros:

- Fast
- Efficient

Cons:

- Less intuitive and friendly interface

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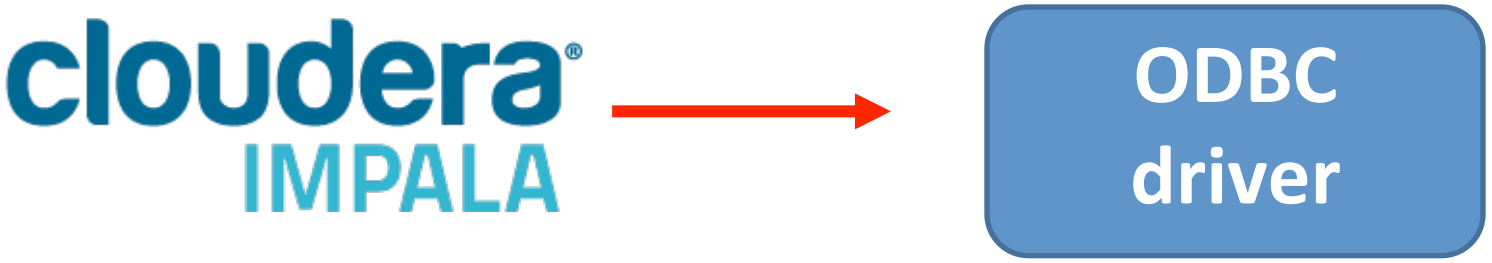

Accessing Impala

cloudera IMPALA → **ODBC driver**

RODBC

Impyla
Ibis

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Hue Interface

ISB Impala Login:

- Login to your impala web interface

Hue Tutorial:

https://github.com/summerela/impala_training/blob/master/using_hue.pdf

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Connecting with impala-shell

- Tutorial:

  https://github.com/summerela/impala_training/blob/master/impala_shell.ipynb

Connecting with Python

- Python:

  https://github.com/summerela/impala_training

  – Click on the ‘launch binder’ icon

  

  – Click on connect_python.ipynb

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Connecting with R

- R:
  - Launch R:
    - Launch your R web interface
  - Tutorial:

    https://github.com/summerela/impala_training/blob/master/connect_with_R.md
  - Script:
    https://github.com/summerela/impala_training/blob/master/connect_R.R

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Building Queries

- Choose your interface
- Follow along (and copy/paste queries):

  https://github.com/summerela/impala_training/blob/master/building_queries.md

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Basic Pipeline

1. Connect to impala
2. Input genes of interest
3. Input subject id's of interest
4. Annotate
5. Filter
6. Export results

https://github.com/summerela/impala_training/blob/master/variant_pipeline_python.ipynb

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