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Genomic Studies at Inova

Inova Translational Medicine Institute
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April 1, 2015

INOVA® HEALTH SYSTEM

Organization

- Background
- Data management
- Challenges \ Lessons Learned

Background:
The Healthcare System

Six hospital + ambulatory healthcare system

- Largest healthcare system in Northern VA
- Two million patient visits/year
- 20,000 deliveries/year

Main hospital

- 1,000 beds including Children's hospital
- 10,000 deliveries/year

Background: The Research Institute

- Started: 2010
- Overall goal: research on integration of genomic information into the practice of medicine
- Staffing: 1/3 clinical, 1/3 bioinformatic/IT, 1/3 laboratory

Background: ITMI Studies

Themes

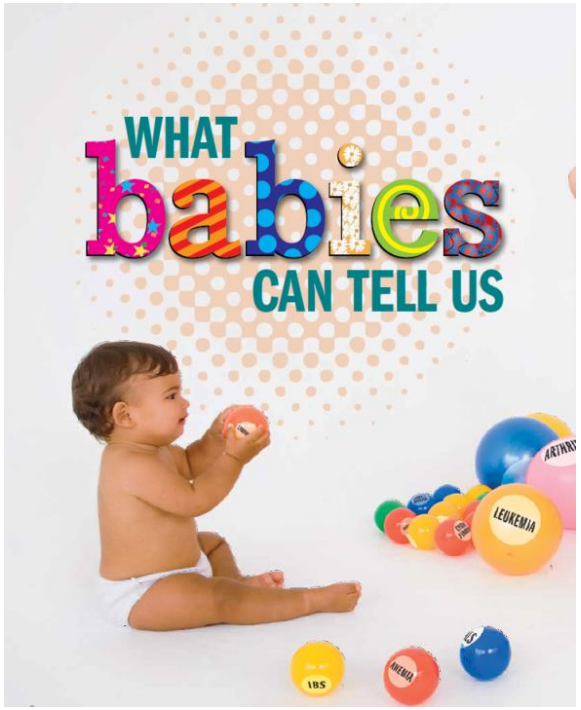
- Trio-based WGS
- Other ‘omics
- Comprehensive clinical data
- Integrated Laboratory
- Unified database

Preterm Birth Study (2011)

- Molecular associations with preterm birth
- ~500 PTB trios, ~500 FT trios
- WGS + ‘omics + clinical data
- Specimens: blood, saliva, cord blood, placenta

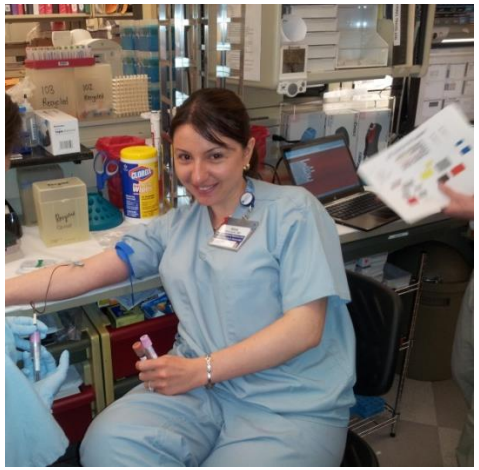
Longitudinal Study (2012)

- **WGS + 'omics + clinical data on 5,000 → 10,000 <u>trio-based</u> families**
- Longitudinal study (≥18 yrs)
- Blood, saliva, urine, cord blood, placenta
- DNA, RNA, protein, epigenetic + clinical data



Congenital Disorders Study (2012)

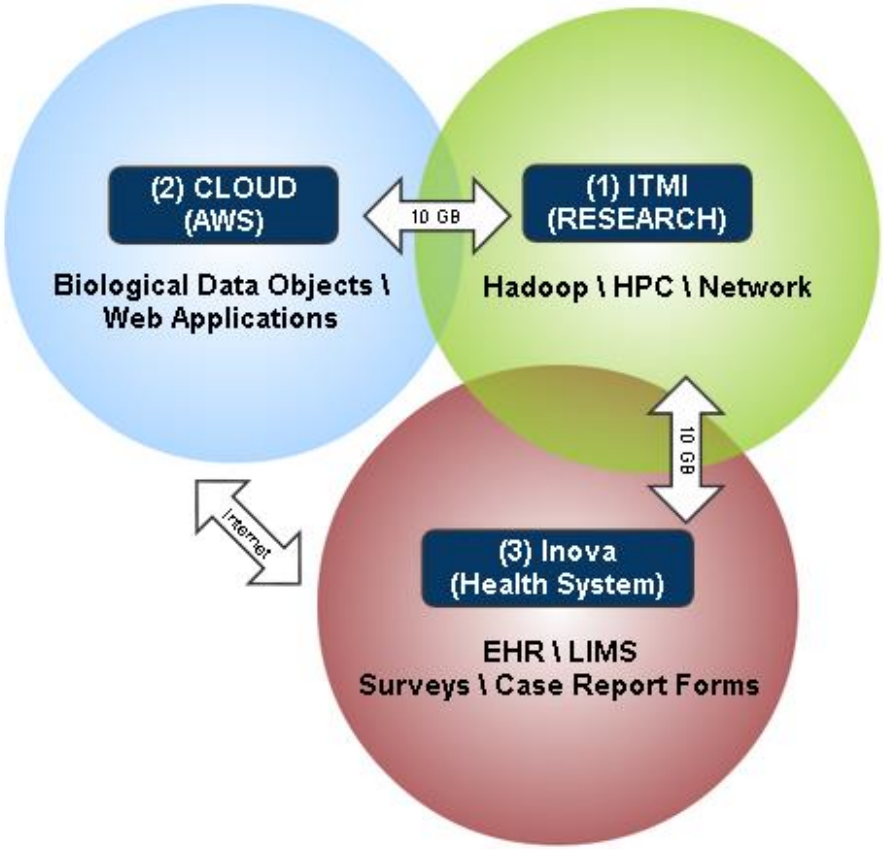
- Mostly NICU-based
- Any other patient with a "congenital/genetic" disorder
- ~2-3 families/week
- Trio-based WGS, etc.



Data Management

Infrastructure: Hybrid Cloud

Cloud



On-Premises

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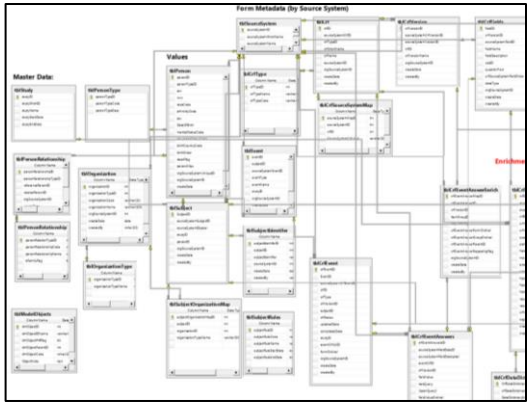
Database

Yes, a Hybrid

Relational Databases
Inova Infrastructure



Object Store
AWS

Storage & Content Delivery

S3
Scalable Storage in the Cloud

Storage Gateway
Integrates On-Premises IT Environments with Cloud Storage

Glacier
Archive Storage in the Cloud

NoSQL \ Hadoop
Value pair \ Graph
Research Network

```
{
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 "name": "HandshakeRequest",
 "namespace":"org.apache.avro.ipc",
 "fields": [
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  {"name": "clientProtocol", "type": ["null", "string"]},
  {"name": "serverHash", "type": "MD5"},
  {"name": "meta", "type": ["null", {"type": "map", "values": "bytes"}]}
 ]
}

{
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 "name": "HandshakeResponse",
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      "symbols": ["BOTH", "CLIENT", "NONE"]}},
  {"name": "serverProtocol","type": ["null", "string"]},
  {"name": "serverHash",
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 ]
}
```

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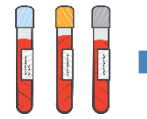
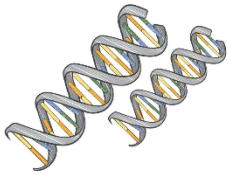
ITMI Data Collection



Bio Materials



Omic Data

Reporting
& Analysis

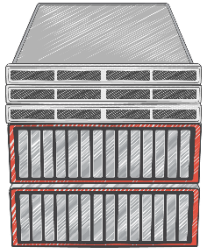
De-identified Data

Meta data

Only As Needed

Hybrid Storage Sync

Consent Patient



On-premises Epic &
Laboratory &
Clinical &
Datawarehouse

External
Collaborators

On-premises Inova
Research staff

On-premises HPC Cluster & Storage

1024 Cores
1.2 PB storage
16 TB Ram

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The Stats

- **Participants**
  - *9,750* participants (enrolled), *> 110* Different Countries of Birth
- **Files and data**
  - Manage 3+ Petabytes of storage capacity (between cloud and on-premise)
  - ~10,000,000 million files
  - File Sizes Range from Kilobytes to 100+ GB per file
- **Specimens**
  - *>450,000* specimens
- **Whole Genomes Sequences**
  - *7,300 +*
  - *~36,500,000,000 variants!*
  - *Also have epigenetic data*
- **Clinical**
  - *55,000+* Patient Diagnosis (Longitudinal)
  - *110,000+*Surveys and Case Report Forms
- **Labs Results**
  - *2,000,000+* discrete lab results
  - *2,200,000+* discrete variables from Case Report Form and Surveys

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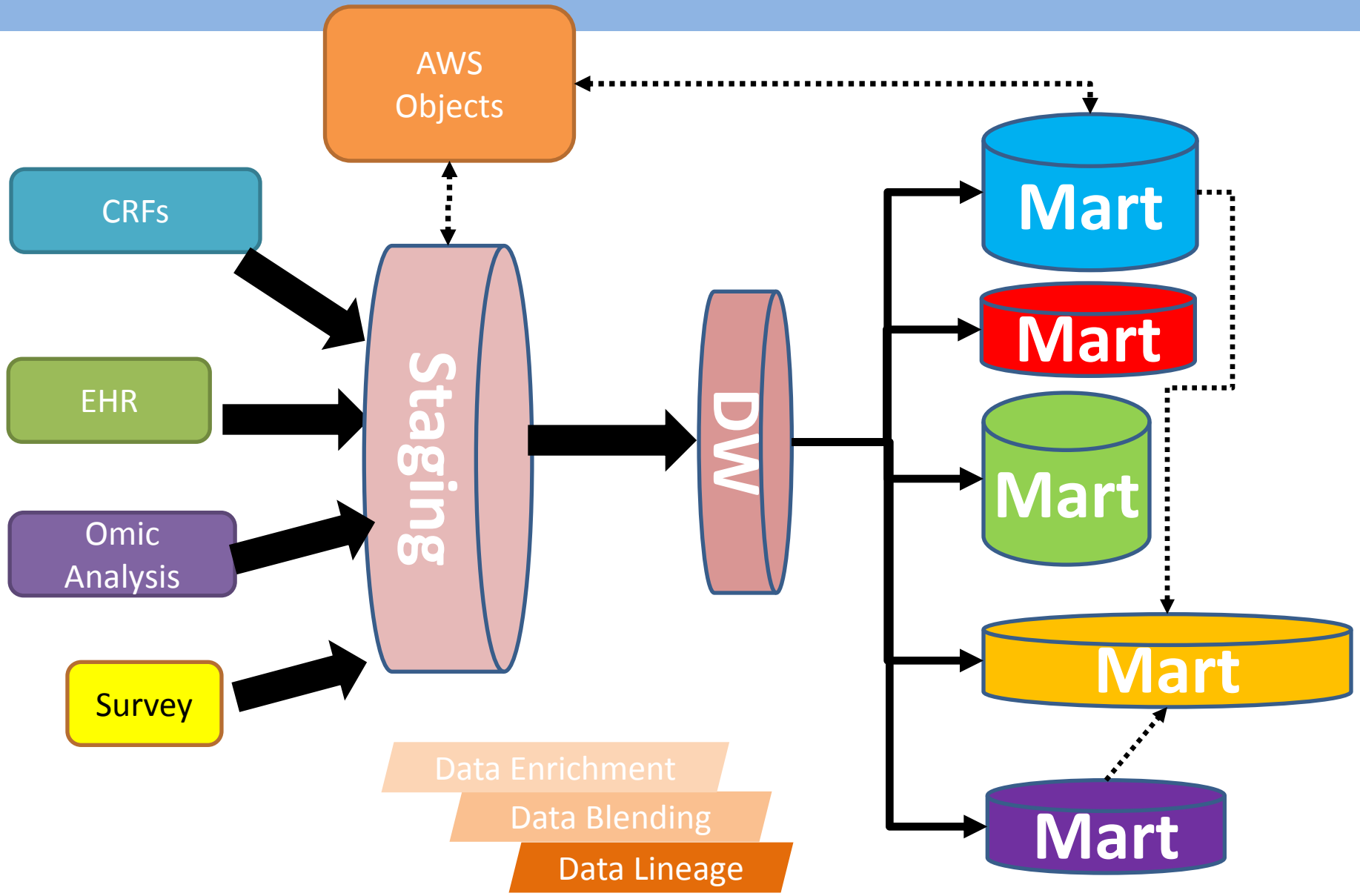
General Data Process



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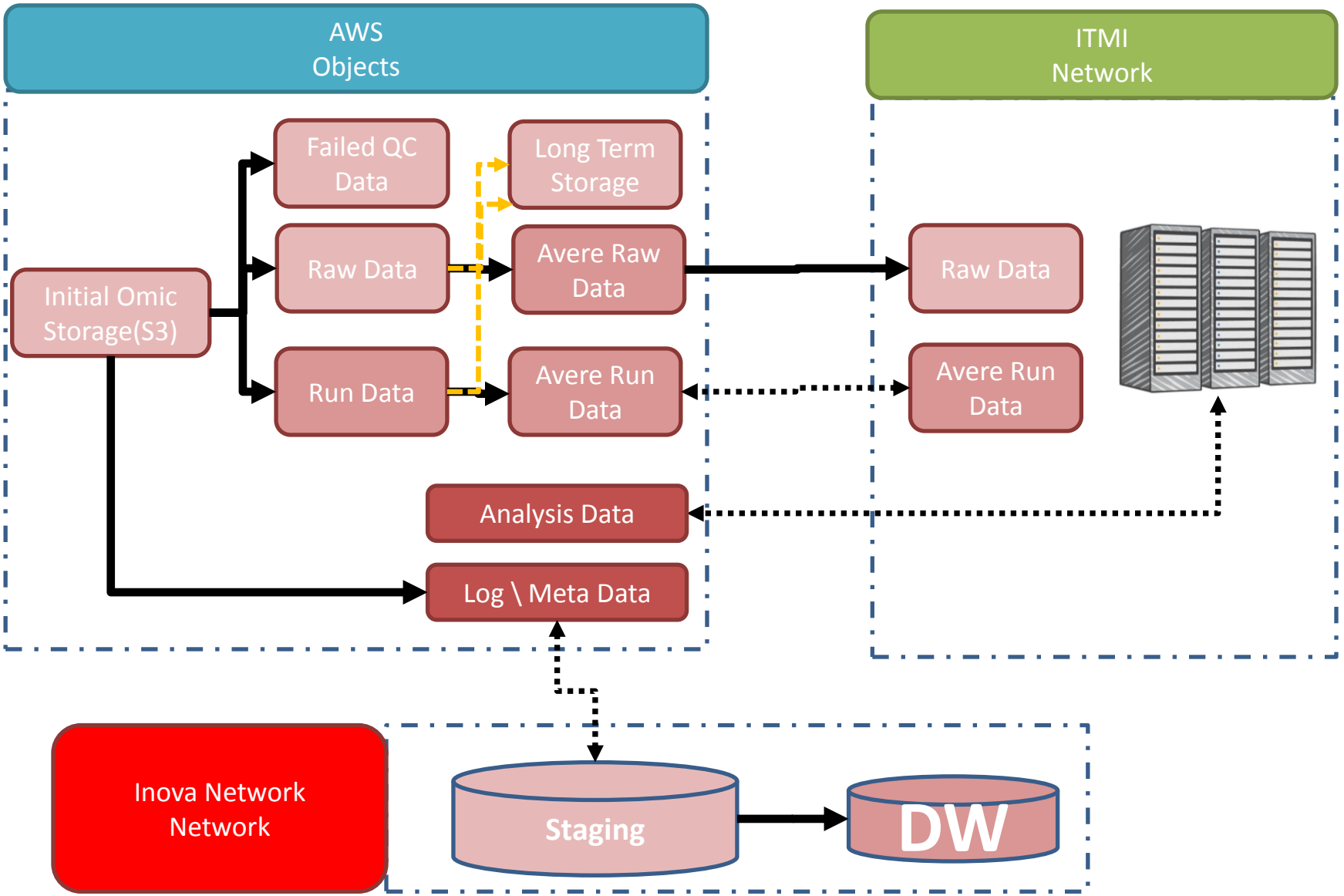
Omic Data Process



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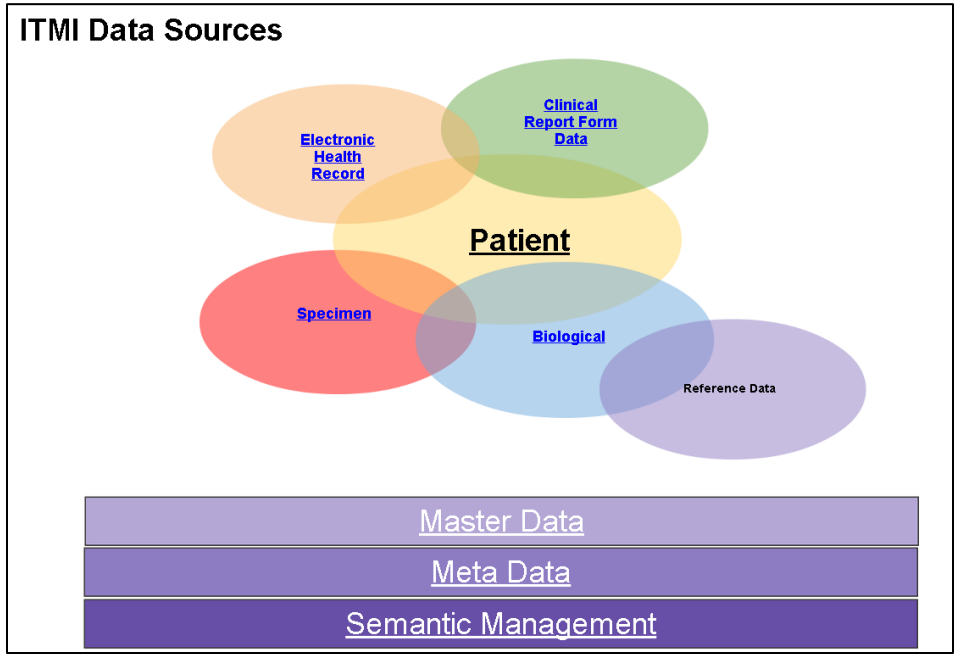
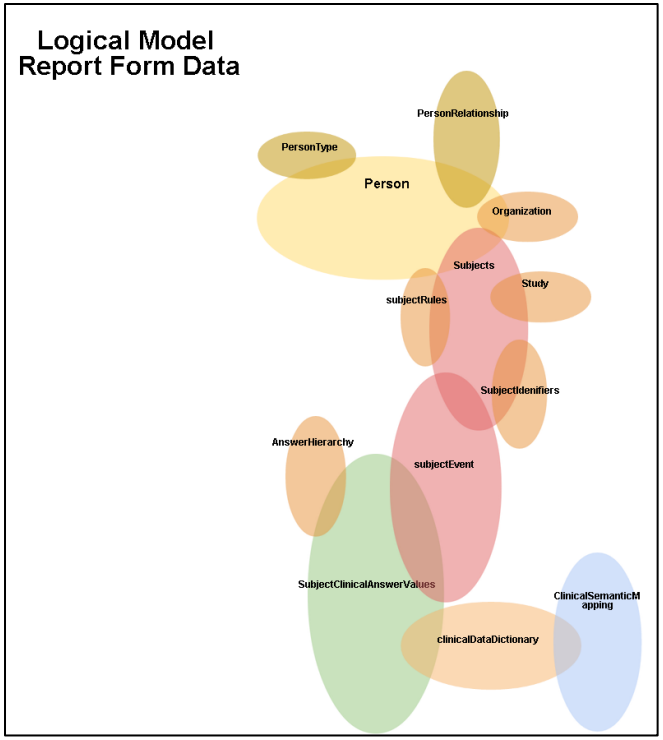
Data Model

(1) Logically Model



(2) Break down each



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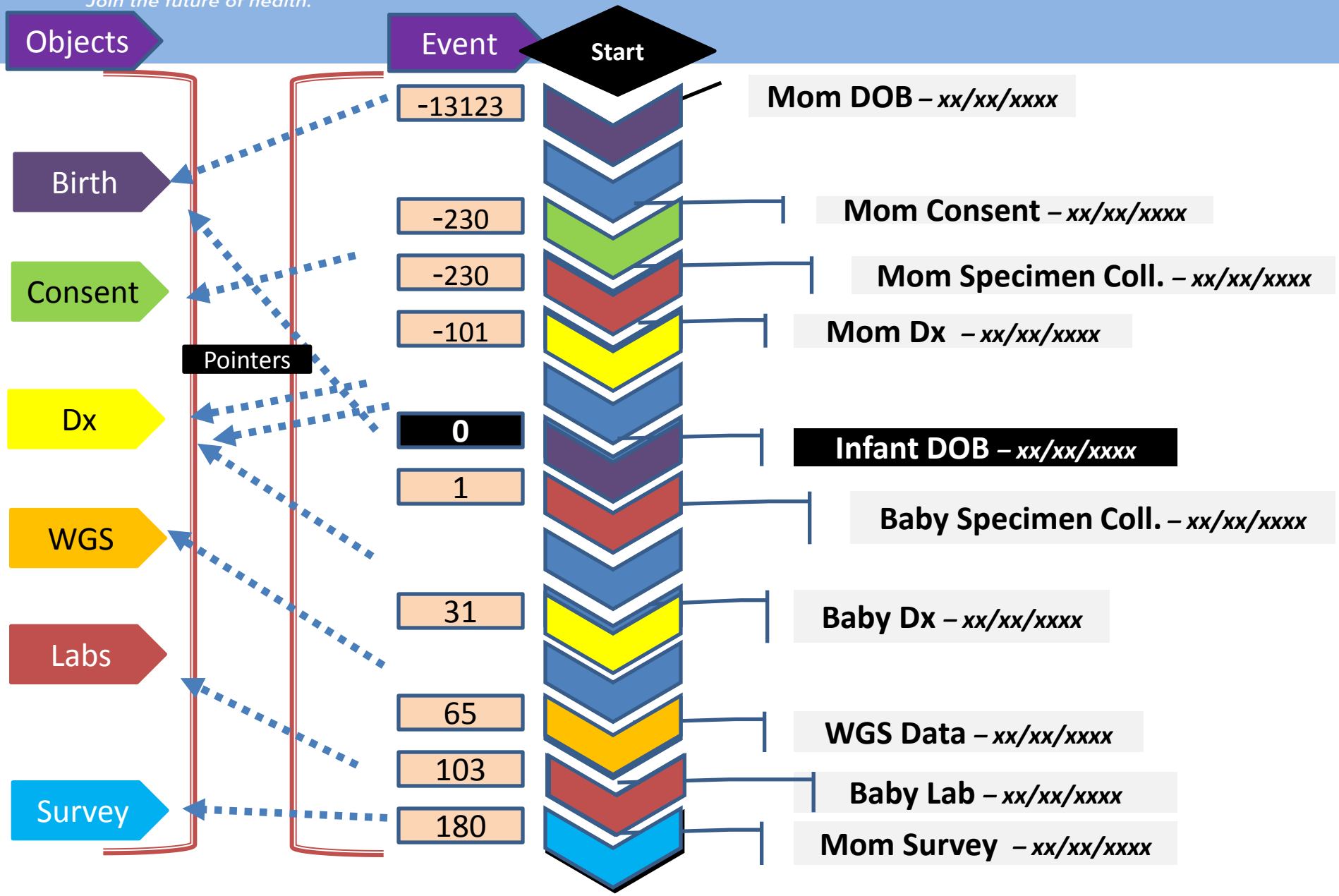
LONGITUDINAL DATA



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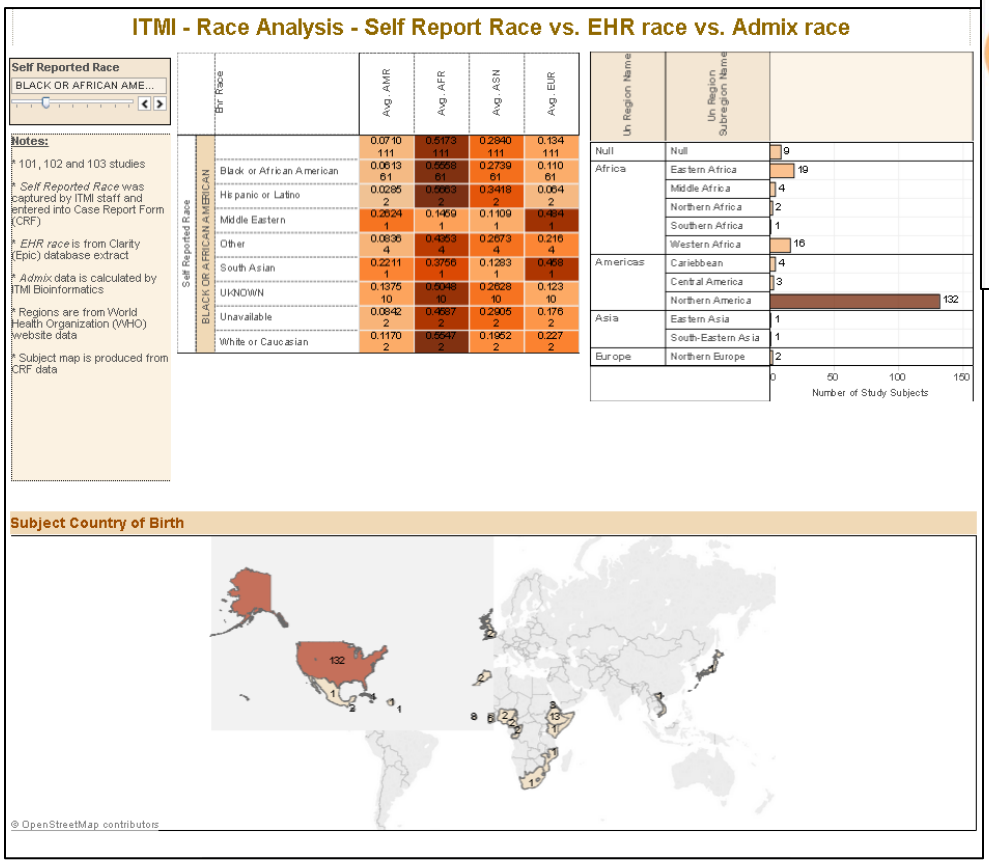
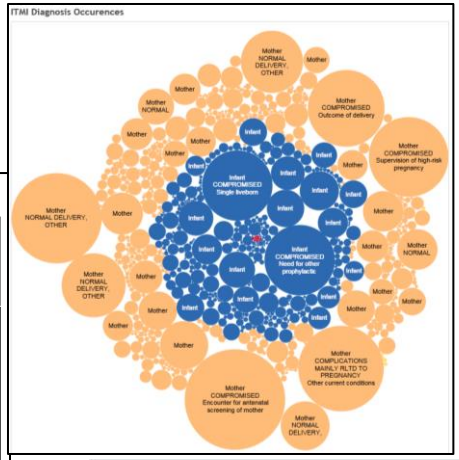
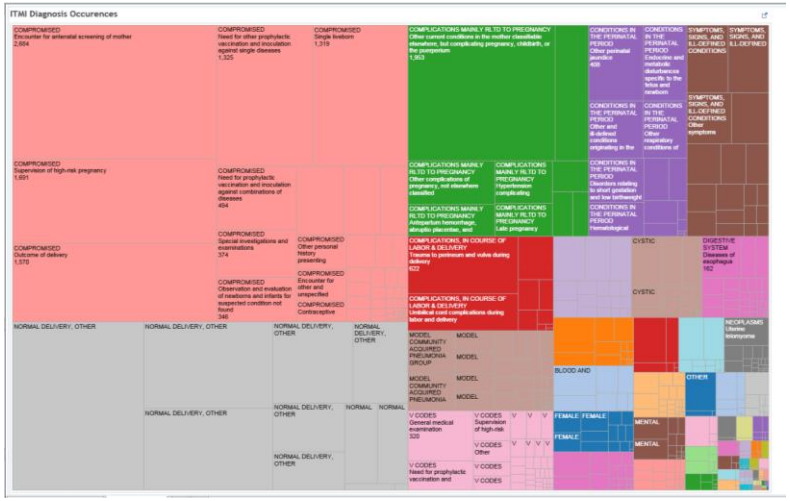
Visualization







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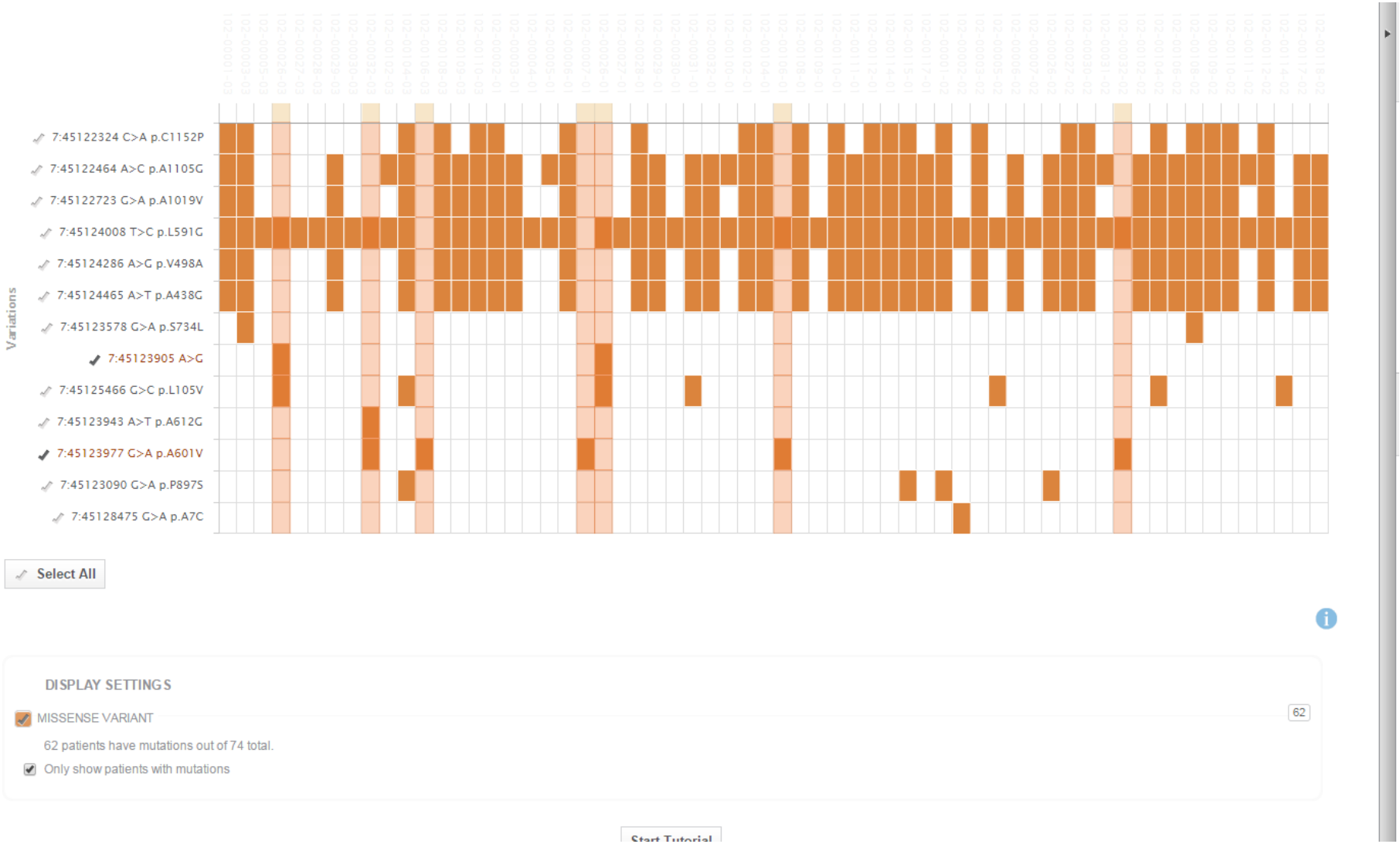
Web Portal



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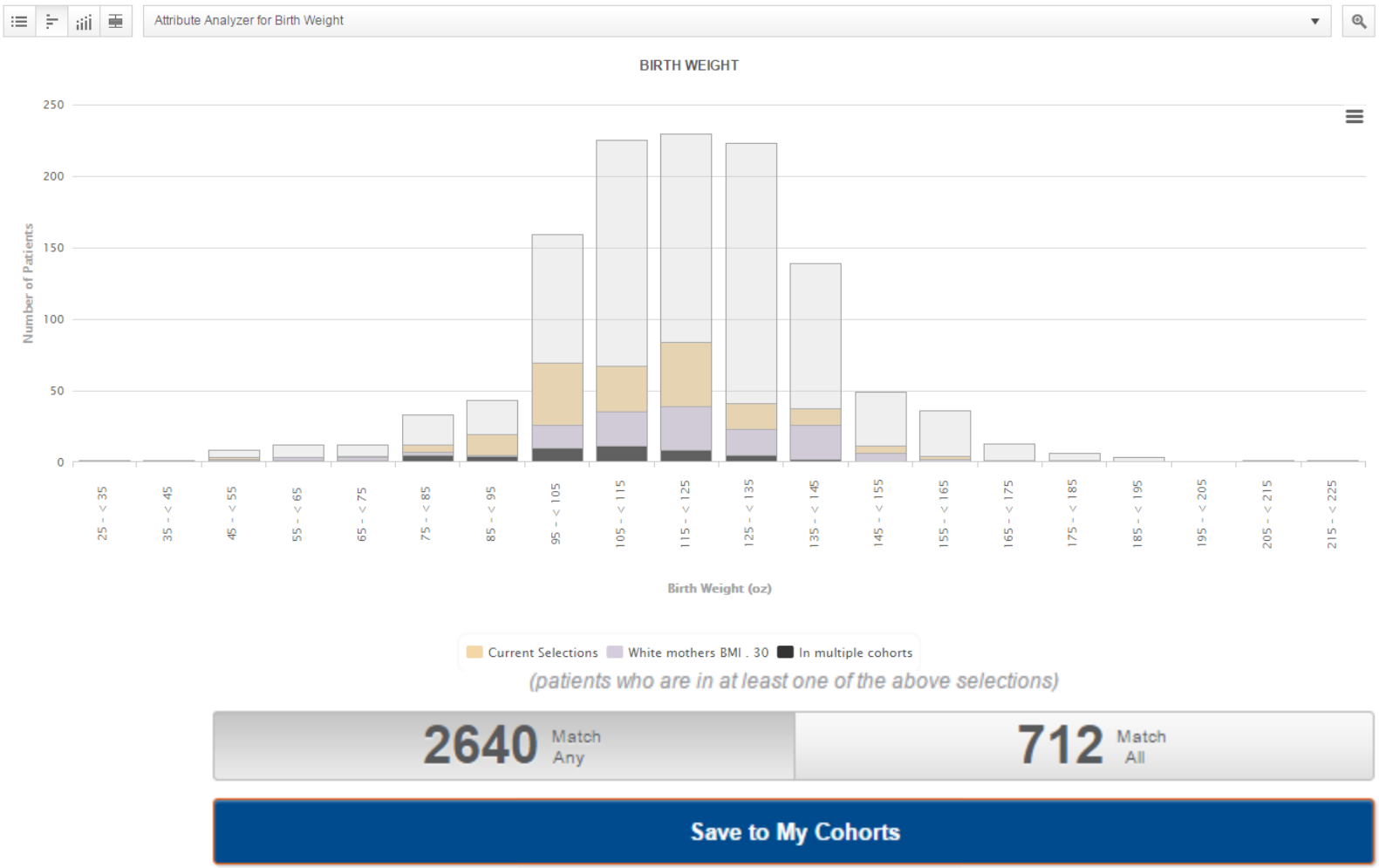
Web Portal



III. Challenges \ Lessons Learned

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Challenges

- Scalability
  - Storage
  - Compute
- Data Movement
- IT Standards
  - Data
  - Programming

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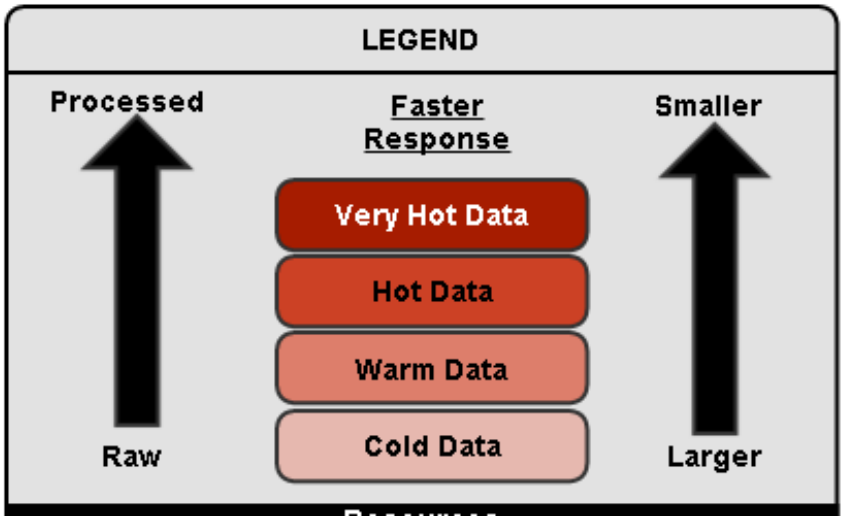
Lessons Learned

- Build infrastructure around data
  - Network is bottleneck
  - Hidden costs
- Manage Data Tiers



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Lessons Learned

- Spend time to model
  - Build strong metadata layer
  - Make it understandable to your team
  - Use best practices
- IT Partners that understand the business

Acknowledgements

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