Genomics and the EHR

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Overview

- EHR from Commercial Perspective
- What can be done TODAY?
- What could be done TOMORROW?
- What are some of the challenges?
- Change management

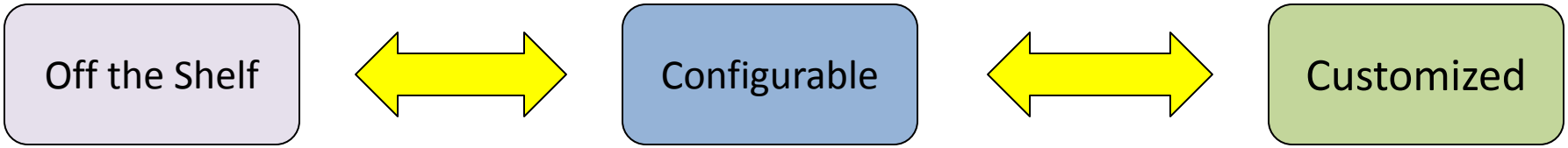
EHR

- Links diagnostic, treatment, procedural, workflow, administrative information in a machine readable framework
- The **LEGALLY BINDING MEDICAL RECORD**
  - Information must be recoverable during legal proceedings
  - Requires ability to reconstruct information available to clinician **AT THE TIME THAT A DECISION WAS MADE**
    - Strict versioning, date/time stamp
    - Updates to interpretations must be clearly identified
- **Enables** privacy protection through:
  - Audit trail of access
  - Role and organization security

How private is this?

Many Facets of an EHR

Many types of “EHR”



- Common build
- Easy start up
- Quick implementation
- Limited flexibility
- Data can be easily exchanged, limited depth

- Common architecture
- Release variability
- Modularity
- Implementations vary widely between sites
- Most data can be exchanged

- Custom architecture
- Highly flexible
- Significant effort to implement
- Limited ability to deploy innovations beyond “parent” institution

Meaningful Use – some important lessons

- Incentive based – enables providers to purchase and install specific EHR capabilities

- Focuses on **functionality**, does not prescribe *how* to deliver functionality
  - Commercial systems have significant investments in their platforms

What can we do TODAY to support genomics in the EHR?

Non-exceptionalist perspective:

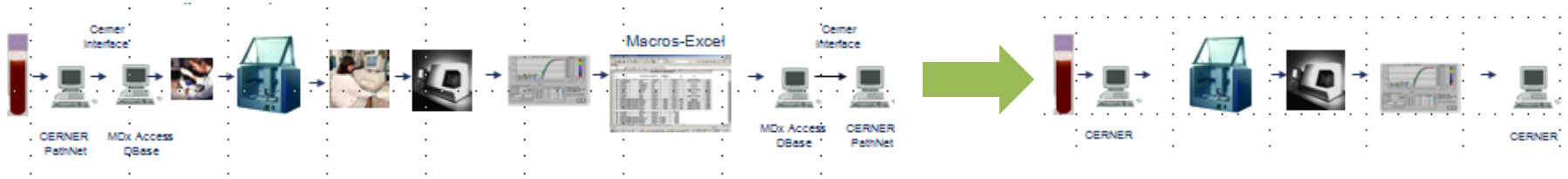
- Any genetic or genomic test can be ordered from an EHR via CPOE
- Any genetic test report can be included in the EHR as a text document

Some EHR systems (or LIS modules within an EHR):

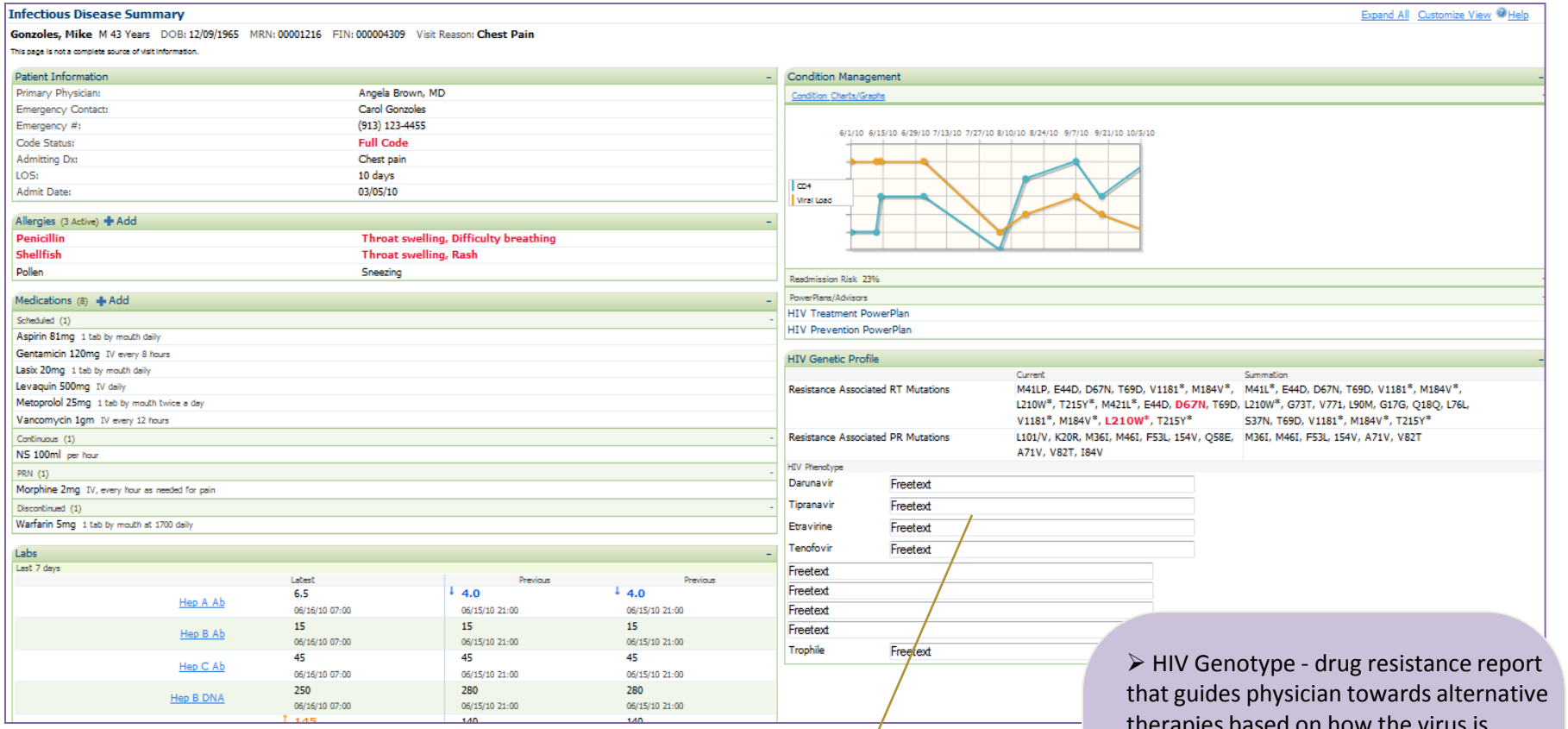
- Support storage of discrete genetic findings (variants, quantitative results, cytogenetic abnormalities, karyotypes)
- Support the workflow in molecular diagnostic laboratories
- Provide ISCN syntax checking
- Enable automated interactions with molecular diagnostic devices (DNA extractions, RT-PCR etc.)
- Can configure decision support rules that utilize discrete genetic information

Current capabilities

- Streamline laboratory process



- Codify discrete results
  - LOINC for orders
  - CBO: becoming *de facto* standard – 17 diagnostic labs in US, Canada and Egypt, VA and 15+ other organizations in process
    - Provides rich semantic structure
- Enable decision support with discrete results
- Publish interpretative reports to EHR
- Works within the most widely used HL7 framework (2.x)

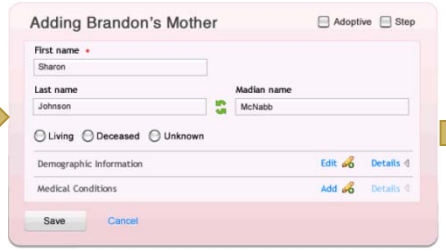


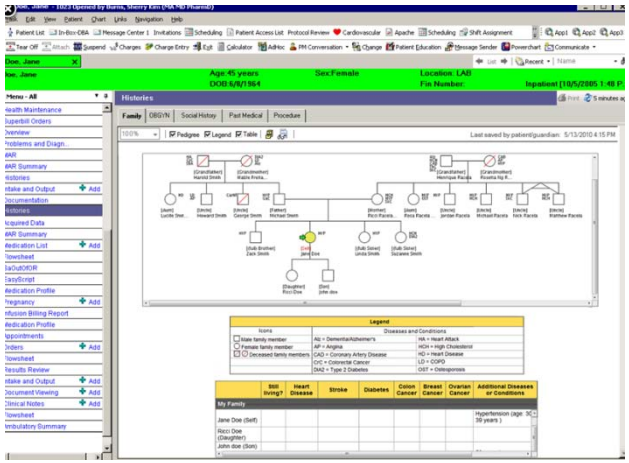
HIV Genetic Profile

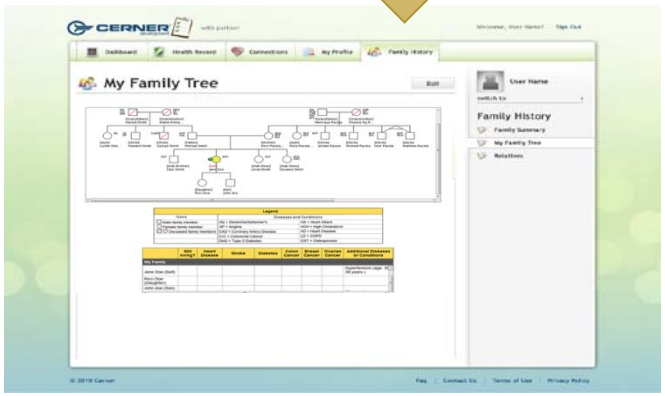
	Current	Summation
Resistance Associated RT Mutations	M41LP, E44D, D67N, T69D, V1181*, M184V*, L210W*, T215Y*, M421L*, E44D, **D67N**, T69D, V1181*, M184V*, **L210W***, T215Y*	M41L*, E44D, D67N, T69D, V1181*, M184V*, L210W*, G73T, V771, L90M, G17G, Q18Q, L76L, S37N, T69D, V1181*, M184V*, T215Y*
Resistance Associated PR Mutations	L101/V, K20R, M36I, M46I, F53L, 154V, Q58E, A71V, V82T, I84V	M36I, M46I, F53L, 154V, A71V, V82T

- HIV Genotype - drug resistance report that guides physician towards alternative therapies based on how the virus is evolving through course of treatment.
  - Current: displays recent mutations for patient based on current therapy. Red indicates new mutations identified.
  - Summation: displays all mutations found for the patient through course of treatment

- Consumer-driven – Cerner Health PHR
- Accessible – web-based Cerner Health PHR
- Portable – one Cerner Health connection to multiple providers
- Time Saving – Completed prior to seeing clinician
- Data Sharing – Copy tree and share with family members
- Standardized – HL7 compliant and SNOMED enabled
- Clinically Relevant – Visual representation for clinicians to track familial conditions using NSCG pedigree standards







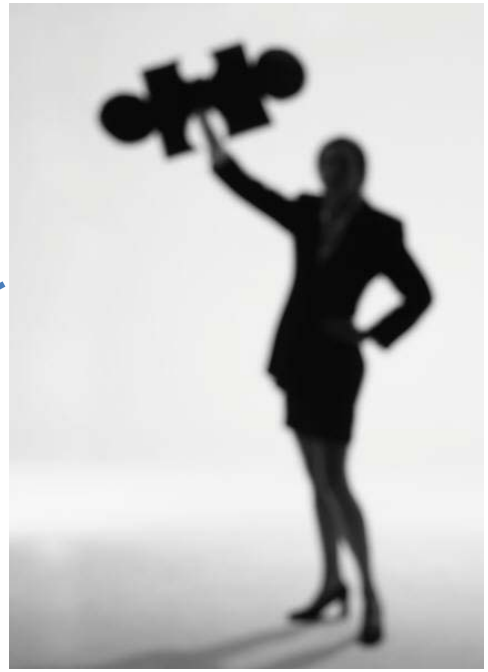
What could be done TOMORROW?

- Link storage of DNA sequencing output to EHR
  - Some lessons from large image files that are stored in archive approach
  - Can be compressed by storing differences relative to a reference sequence
- Assist diagnostic expert with interpretation of DNA sequence results (current or NextGEN)
  - Highlight variants of known significance
  - Highlight variants with likely impact despite lack of known signficance (stop, frameshift)
  - Document and archive variants of unknown significance for future interpretation
- Periodically reassess results of unknown significance as new findings become available

The problem with using a fixed set of codes

- There's always that extra piece…
- Difficult for curated content to keep up with the pace of science

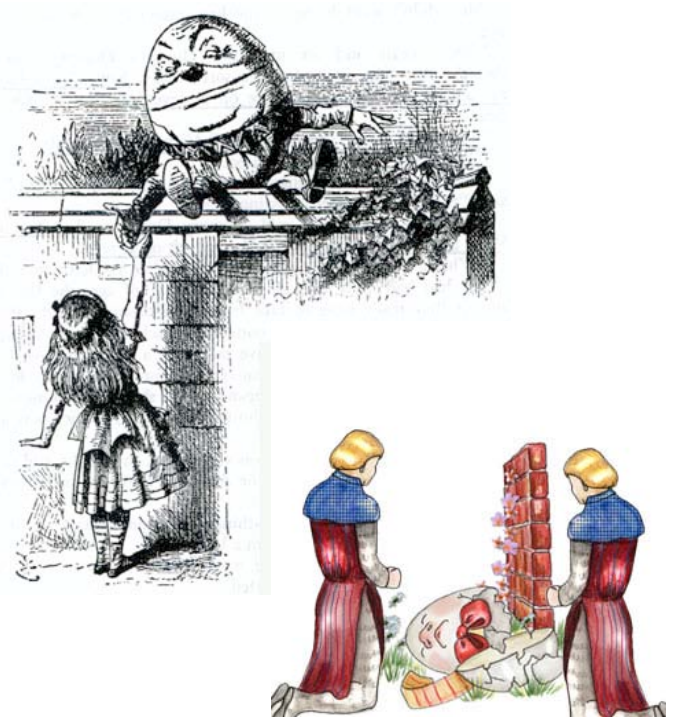
CTTCTCCGTGTCCACCTTGCGCAACTTGGG
CCTGGGCAAGAAGTCGCTGGAGCAGTGG
GTGACCGAGGAGGCCGCCTGCCTTTGTGC
CGCCTTCGCCGACCAAGCCGG



The problem with codifying after the result is captured….

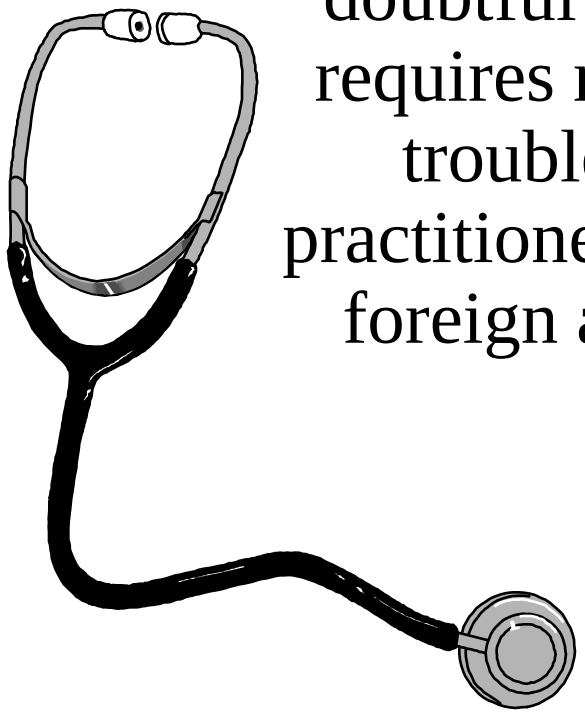
• Codification depends on the rules that are implemented at the time that a result is captured

•If (when) the logical rules change, the codification of a variant can vary due to minor modification of the rules. Querying these codes will be challenging.



CTTCTCCGTGTCCACCTTGCGCAACTTGGG
CCTGGGCAAGAAGTCGCTGGAGCAGTGG
GTGACCGAGGAGGCCGCCTGCCTTTGTGC
CGCCTTCGCCGACCAAGCCGG

Change Management - Scenario 1



"That it will ever come into general use, notwithstanding its value, is extremely doubtful because its beneficial application requires much time and gives a good bit of trouble, both to the patient and to the practitioner because its hue and character are foreign and opposed to all our habits and associations."

from The London Times in 1834

Commenting on ...

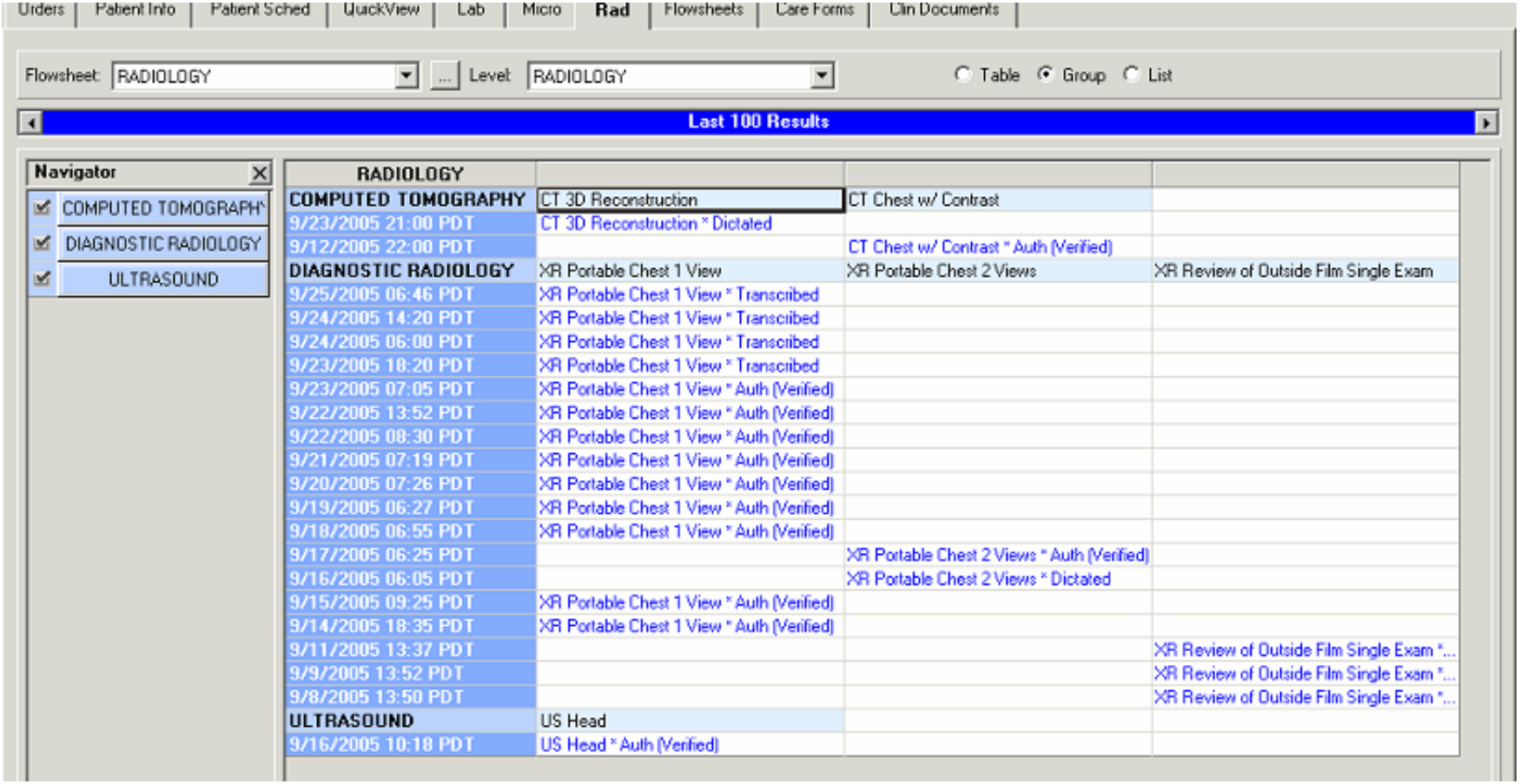
the "stethoscope"

Change Management - Scenario 2

- The iPad was immediately utilized in clinical practice

- Users saw immediate value and did not seek permission or regulatory approval

Imaging – some similar issues

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Edit
View
Time Sca
Document Viewer - MDDEMOONLY, ONE - 1000450-5
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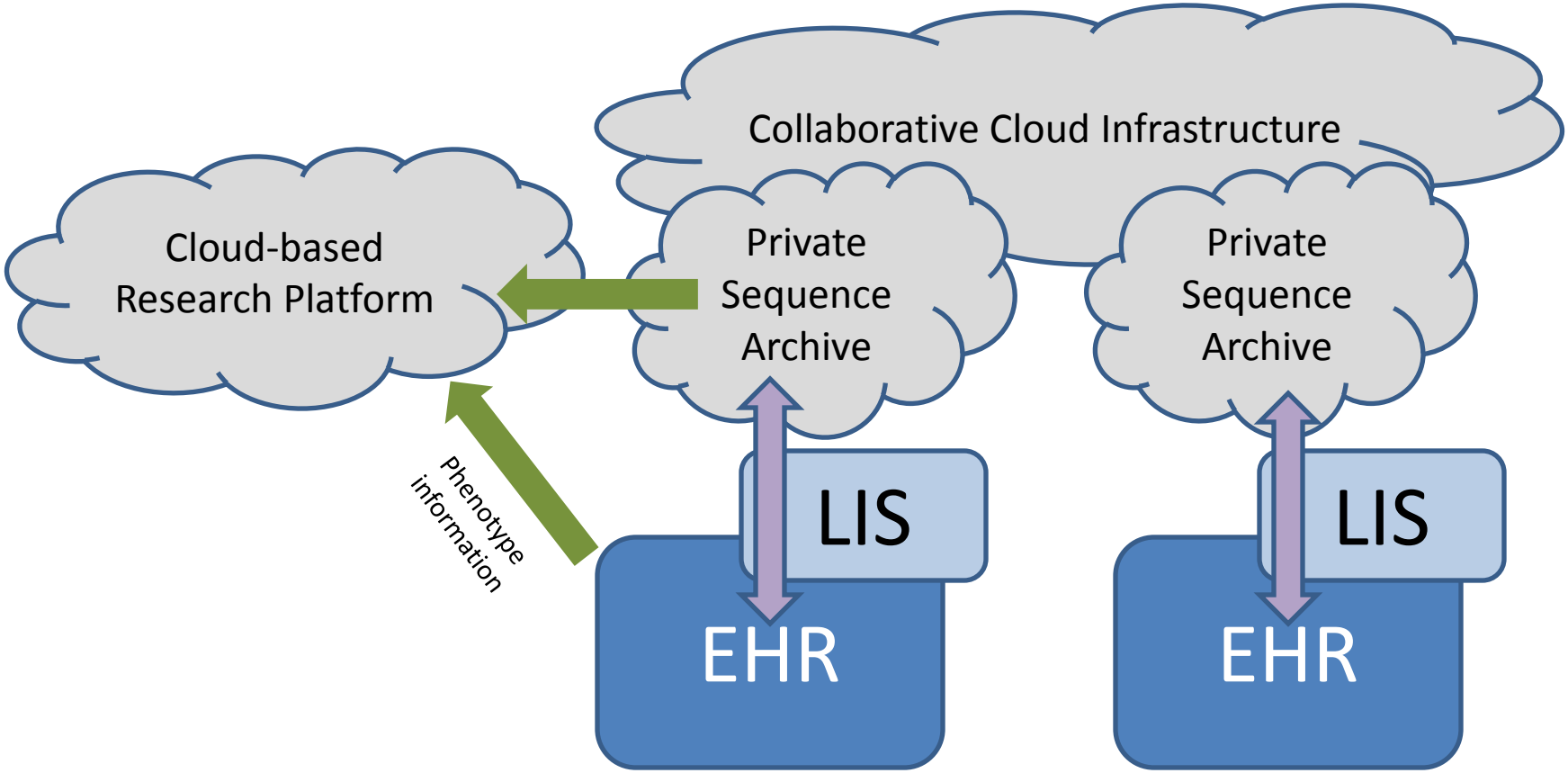
HAHNJ logged in on Thursday, March 10, 2005 9:17:43 PM
0.0
516/1032
<no presentation state>
Orders
Flows
Nav
Viewport
Series
Study
All
Patient info
Exam info
Report
Worklist

Some lessons and questions from imaging

- Reports and orders are within EHR context
- Large image files are stored in archive, with links to EHR
- Would the $10 MRI mean that everybody would and should have an MRI? Are there enough radiologists to support the interpretation of the results?

Architecture



Scenario 3

Here is my
DNA sequence!

Courtesy: A. Gonzalez

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