

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

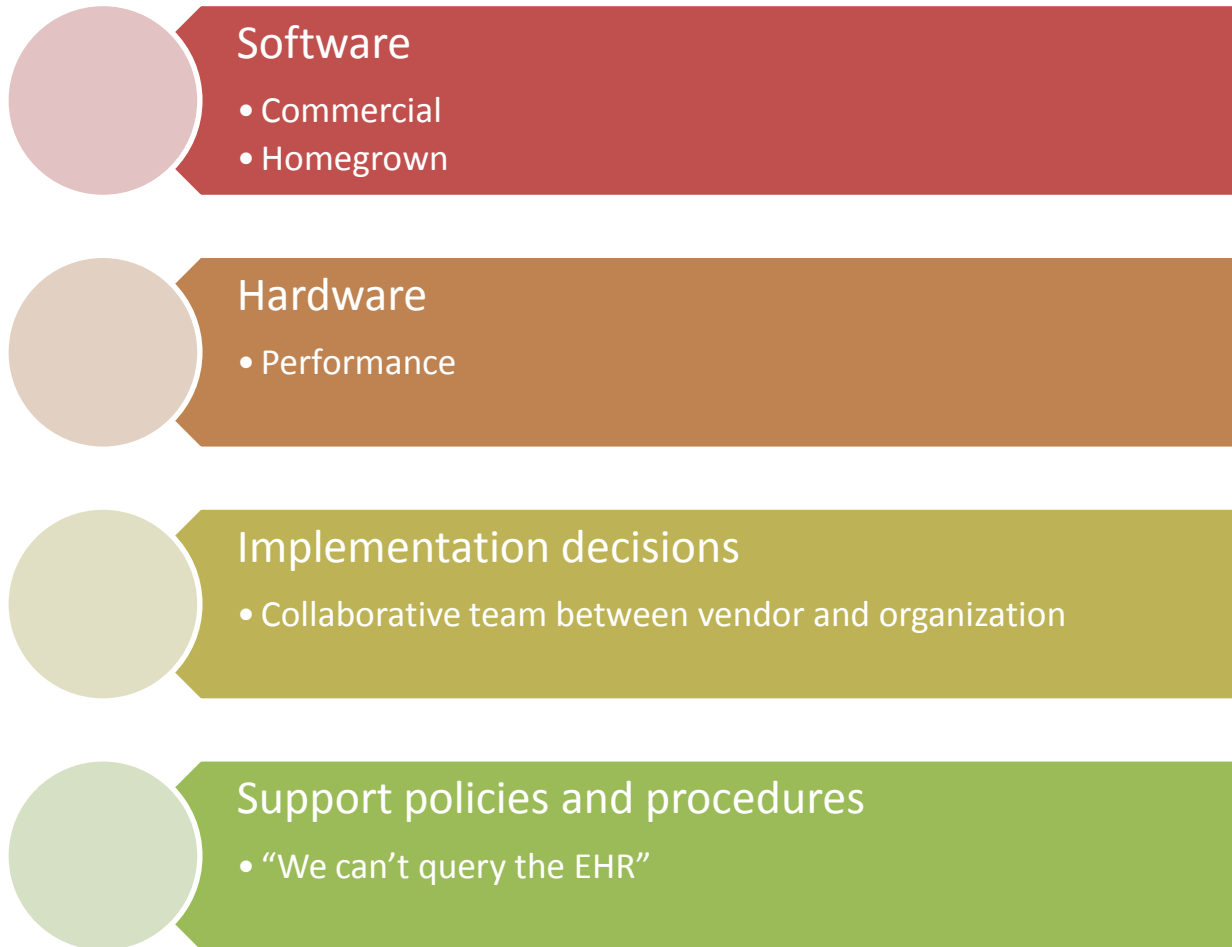
EHR

- 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

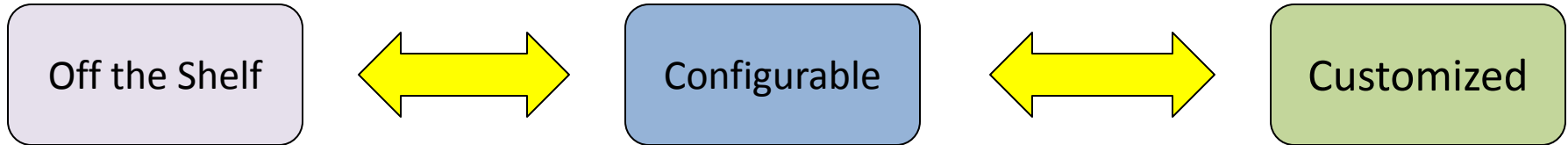
How private is this?



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:

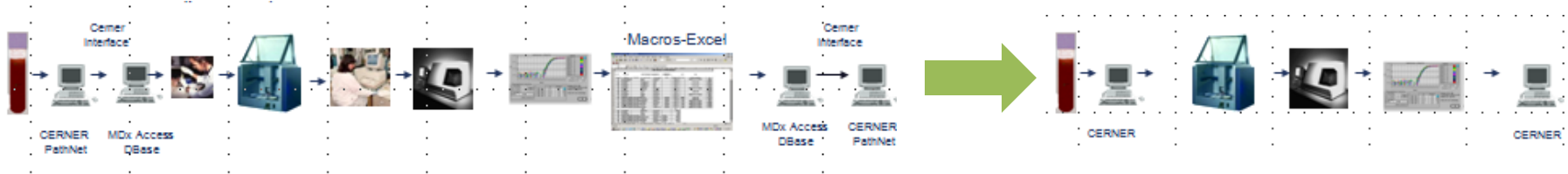
- Any genetic or genomic test can be ordered from an EHR via CPOE
- Any genetic test report can be included in the EHR as a text document

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)

Infectious Disease Summary

Gonzales, Mike M 43 Years DOB: 12/09/1965 MRN: 00001216 FIN: 000004309 Visit Reason: **Chest Pain**

This page is not a complete source of visit information.

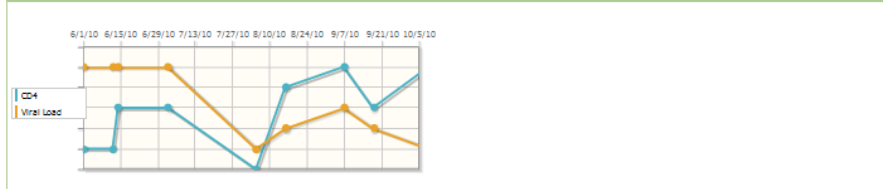
| Patient Information | |
|---------------------|------------------|
| Primary Physician: | Angela Brown, MD |
| Emergency Contact: | Carol Gonzales |
| Emergency #: | (913) 123-4455 |
| Code Status: | Full Code |
| Admitting Dx: | Chest pain |
| LOS: | 10 days |
| Admit Date: | 03/05/10 |

| Allergies (3 Active) + Add | |
|----------------------------|---------------------------------------|
| Penicillin | Throat swelling, Difficulty breathing |
| Shellfish | Throat swelling, Rash |
| Pollen | Sneezing |

| Medications (8) + Add | |
|-----------------------|-----------------------------------|
| Scheduled (1) | |
| Aspirin 81mg | 1 tab by mouth daily |
| Gentamicin 120mg | IV every 8 hours |
| Lasix 20mg | 1 tab by mouth daily |
| Levaquin 500mg | IV daily |
| Metoprolol 25mg | 1 tab by mouth twice a day |
| Vancomycin 1gm | IV every 12 hours |
| Continuous (1) | |
| NS 100ml | per hour |
| PRN (1) | |
| Morphine 2mg | IV, every hour as needed for pain |
| Discontinued (1) | |
| Warfarin 5mg | 1 tab by mouth at 1700 daily |

| Labs | | | | |
|---------------------------|-----------------------|-------------------------|-------------------------|----------|
| Last 7 days | Latest | Previous | Previous | Previous |
| Hep A Ab | 6.5 06/16/10 07:00 | ↓ 4.0 06/15/10 21:00 | ↓ 4.0 06/15/10 21:00 | |
| Hep B Ab | 15 06/16/10 07:00 | 15 06/15/10 21:00 | 15 06/15/10 21:00 | |
| Hep C Ab | 45 06/16/10 07:00 | 45 06/15/10 21:00 | 45 06/15/10 21:00 | |
| Hep B DNA | 250 06/16/10 07:00 | 280 06/15/10 21:00 | 280 06/15/10 21:00 | |

Condition Management



| | |
|---------------------|---|
| Readmission Risk | 23% |
| PowerPlans/Advisors | HIV Treatment PowerPlan HIV Prevention PowerPlan |

| 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 Phenotype | |
|---------------|----------|
| Danavir | Freetext |
| Tipranavir | Freetext |
| Etravirine | Freetext |
| Tenofovir | Freetext |
| Freetext | |
| Freetext | |
| Freetext | |
| Freetext | |
| Trophile | Freetext |

➤ 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

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 |

- 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



Adding Brandon's Mother

Adoptive Step

First name

Last name **Median name**

Living Deceased Unknown

Demographic Information [Edit](#) [Details](#)

Medical Conditions [Add](#) [Details](#)

Adding Brandon's Mother

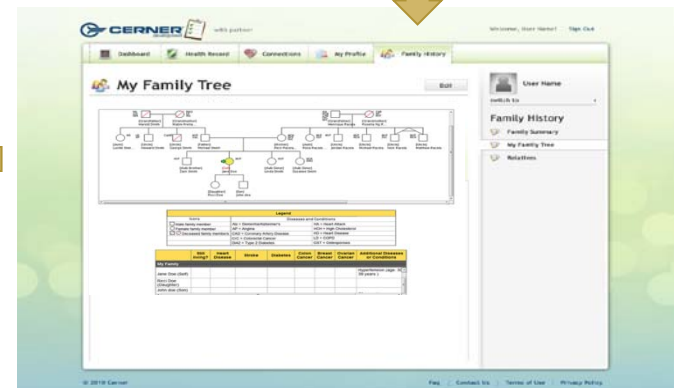
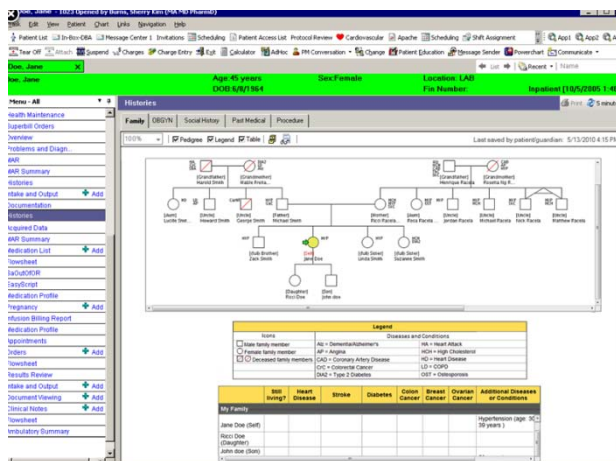
Adoptive Step

Identify the Type of Condition

heart Lung Kidney Gastro Diabetes Cancer Neuro Pregnancy Other

Choose a Condition
Congenital Heart Defects

Age at Onset of Condition
Date of Birth



What could be done TOMORROW?

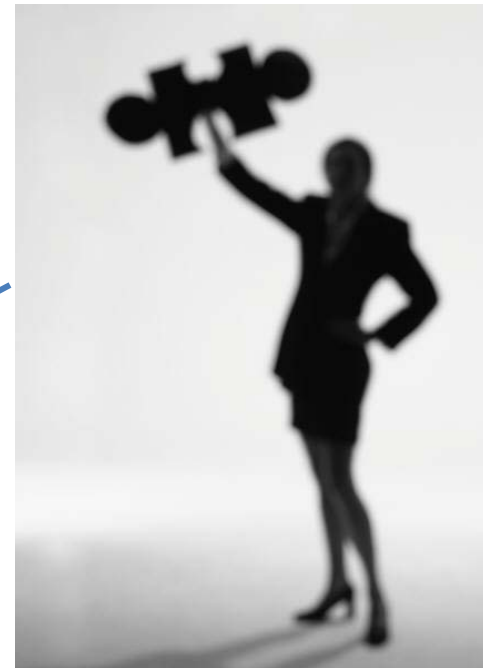
- Link storage of DNA sequencing output to EHR
 - Some lessons from large image files that are stored in archive approach
 - Can be compressed by storing differences relative to a reference sequence
- Assist diagnostic expert with interpretation of DNA sequence results (current or NextGEN)
 - Highlight variants of known significance
 - Highlight variants with likely impact despite lack of known significance (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

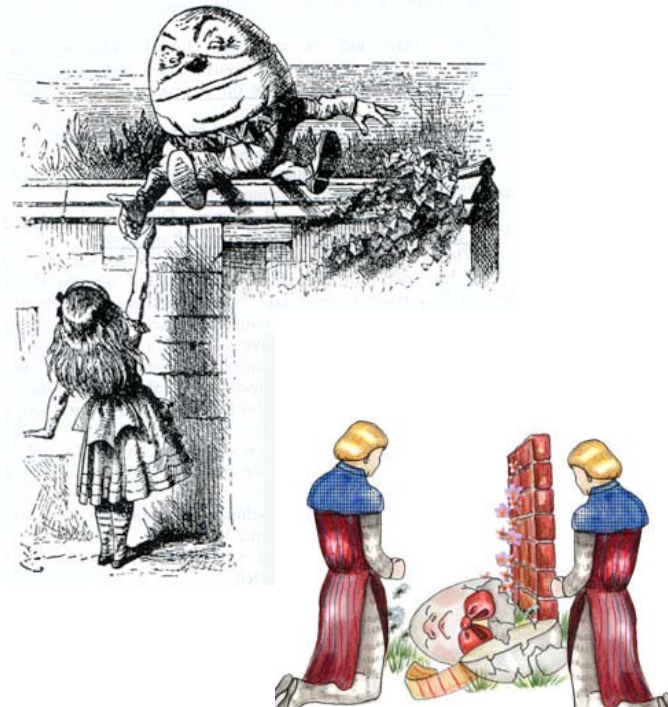
```
CTTCTCCGTGTCCACCTTGCGCAACTTGGG  
CCTGGGCAAGAAGTCGCTGGAGCAGTGG  
GTGACCGAGGAGGCCGCCTGCCTTTGTGC  
CGCCTTCGCCGACCAAGCCGG
```



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

Orders | Patient Info | Patient Sched | QuickView | Lab | Micro | **Rad** | Flowsheets | Care Forms | Clin Documents

Flowsheet: RADIOLOGY Level: RADIOLOGY Table Group List

Last 100 Results

| Navigator | RADIOLOGY | | | |
|--|-----------------------------|--|---|--|
| <input checked="" type="checkbox"/> COMPUTED TOMOGRAPHY | COMPUTED TOMOGRAPHY | CT 3D Reconstruction | CT Chest w/ Contrast | |
| | 9/23/2005 21:00 PDT | CT 3D Reconstruction * Dictated | | |
| <input checked="" type="checkbox"/> DIAGNOSTIC RADIOLOGY | 9/12/2005 22:00 PDT | | CT Chest w/ Contrast * Auth (Verified) | |
| <input checked="" type="checkbox"/> ULTRASOUND | DIAGNOSTIC RADIOLOGY | XR Portable Chest 1 View | XR Portable Chest 2 Views | XR Review of Outside Film Single Exam |
| | 9/25/2005 06:46 PDT | XR Portable Chest 1 View * Transcribed | | |
| | 9/24/2005 14:20 PDT | XR Portable Chest 1 View * Transcribed | | |
| | 9/24/2005 06:00 PDT | XR Portable Chest 1 View * Transcribed | | |
| | 9/23/2005 18:20 PDT | XR Portable Chest 1 View * Transcribed | | |
| | 9/23/2005 07:05 PDT | XR Portable Chest 1 View * Auth (Verified) | | |
| | 9/22/2005 13:52 PDT | XR Portable Chest 1 View * Auth (Verified) | | |
| | 9/22/2005 08:30 PDT | XR Portable Chest 1 View * Auth (Verified) | | |
| | 9/21/2005 07:19 PDT | XR Portable Chest 1 View * Auth (Verified) | | |
| | 9/20/2005 07:26 PDT | XR Portable Chest 1 View * Auth (Verified) | | |
| | 9/19/2005 06:27 PDT | XR Portable Chest 1 View * Auth (Verified) | | |
| | 9/18/2005 06:55 PDT | XR Portable Chest 1 View * Auth (Verified) | | |
| | 9/17/2005 06:25 PDT | | XR Portable Chest 2 Views * Auth (Verified) | |
| | 9/16/2005 06:05 PDT | | XR Portable Chest 2 Views * Dictated | |
| | 9/15/2005 09:25 PDT | XR Portable Chest 1 View * Auth (Verified) | | |
| | 9/14/2005 18:35 PDT | XR Portable Chest 1 View * Auth (Verified) | | |
| | 9/11/2005 13:37 PDT | | | XR Review of Outside Film Single Exam *... |
| | 9/9/2005 13:52 PDT | | | XR Review of Outside Film Single Exam *... |
| | 9/8/2005 13:50 PDT | | | XR Review of Outside Film Single Exam *... |
| | ULTRASOUND | US Head | | |
| | 9/16/2005 10:18 PDT | US Head * Auth (Verified) | | |

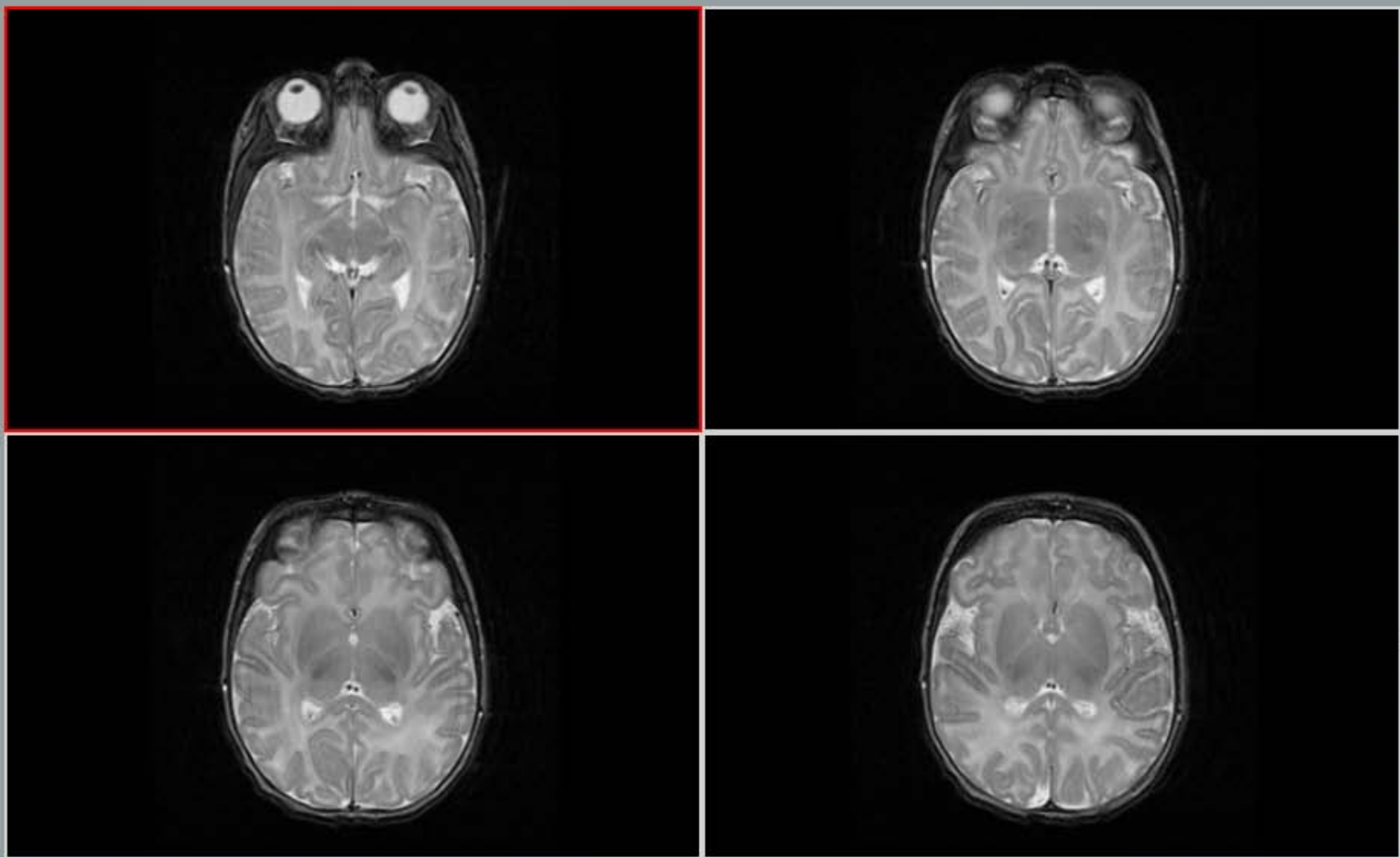
Pictorial +

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Navigation and display controls including zoom (0.0), color calibration, and window management icons.

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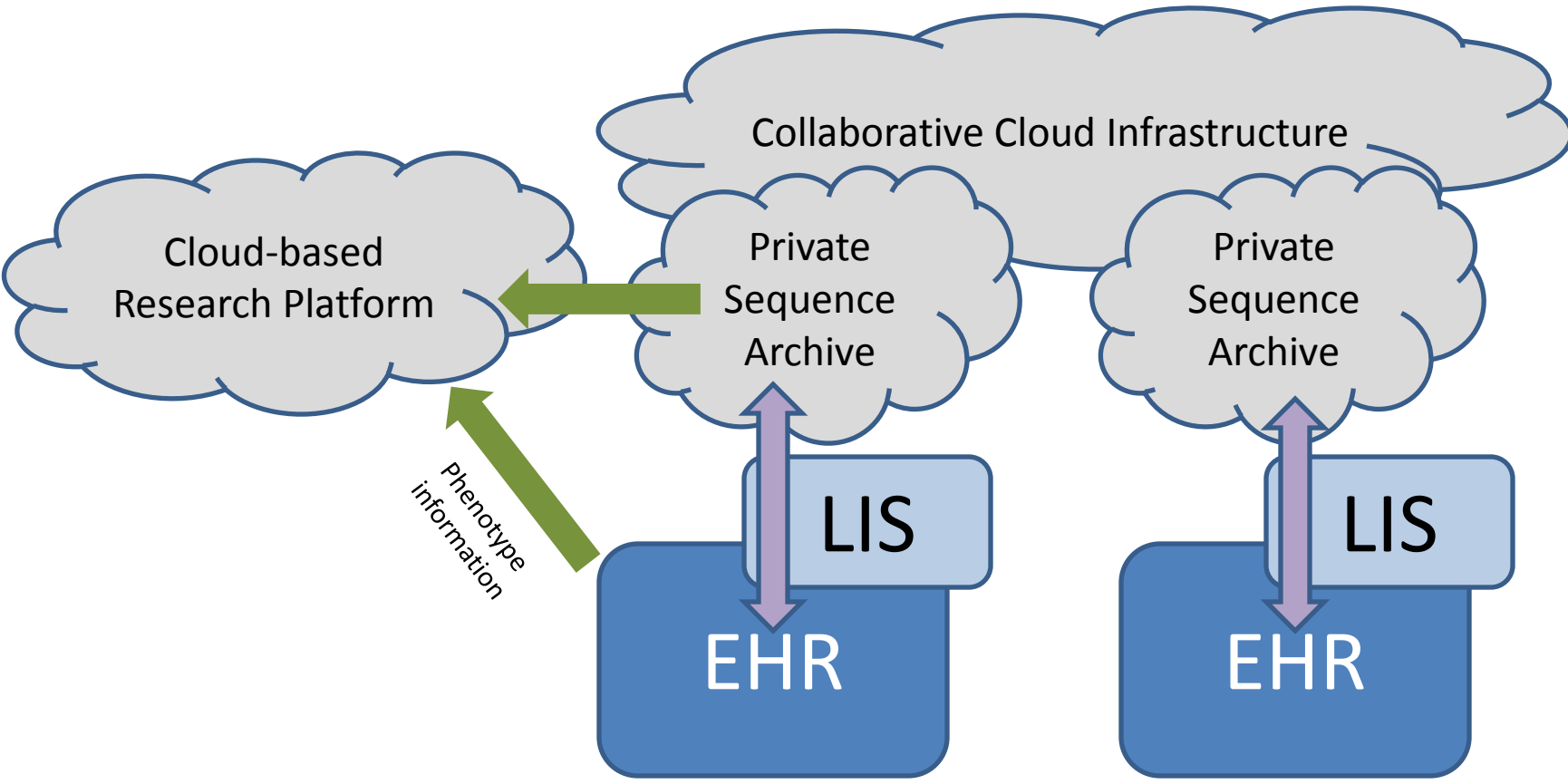


- Viewport
- Series
 - Study
 - All

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!

