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

Software

Commercial

Homegrown

Hardware

• Performance

Implementation decisions

• Collaborative team between vendor and organization

Support policies and procedures

• "We can't query the 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:

- <u>Any genetic or genomic test can be ordered from an EHR via CPOE</u>
- <u>Any</u> 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

LOS:

Labs

Gonzoles, 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.

Expand All Customize View Help

Patient Information Condition Management Angela Brown, MD Primary Physician: Condition Charts/Graphs Emergency Contact: Carol Gonzoles (913) 123-4455 Emergency #: 6/1/10 6/15/10 6/29/10 7/13/10 7/27/10 8/10/10 8/24/10 9/7/10 9/21/10 10/5/10 Code Status: Full Code Admitting Dx: Chest pain 10 days 004 Admit Date: 03/05/10 Viral Load Allergies (3 Active) 🕂 Add Penicillin Throat swelling, Difficulty breathing Shellfish Throat swelling, Rash Pollen Sneezing Readmission Risk 23% Medications (8) + Add PowerPlans/Advisors HIV Treatment PowerPlan Scheduled (1) HIV Prevention PowerPlan Aspirin 81mg 1 tab by mouth daily Gentamicin 120mg IV every 8 hours **HIV Genetic Profile** Lasix 20mg 1 tab by mouth daily Current Summation Levaquin 500mg IV daily Resistance Associated RT Mutations M41LP, E44D, D67N, T69D, V1181*, M184V*, M41L*, E44D, D67N, T69D, V1181*, M184V*, Metoprolol 25mg 1 tab by mouth twice a day L210W*, T215Y*, M421L*, E44D, D67N, T69D, L210W*, G73T, V771, L90M, G17G, Q18Q, L76L, V1181*, M184V*, L210W*, T215Y* Vancomycin 1gm IV every 12 hours 537N, T69D, V1181*, M184V*, T215Y* Resistance Associated PR Mutations L101/V, K20R, M36I, M46I, F53L, 154V, Q58E, M36I, M46I, F53L, 154V, A71V, V82T Continuous (1) A71V, V82T, I84V NS 100ml per hour HIV Phenotype PRN (1) Darunavir Freetext Morphine 2mg IV, every hour as needed for pain Tipranavir Freetext Discontinued (1) Warfarin 5mg 1 tab by mouth at 1700 daily Etravirine Freetext Tenofovir Freetext Last 7 days Freetext Latest Previou Previous 4.0 4.0 Freetext 6.5 Hep A Ab 06/16/10 07:00 06/15/10 21:00 06/15/10 21:00 Freetext 15 15 15 Freetext Hep B Ab 06/16/10 07:00 06/15/10 21:00 06/15/10 21:00 Trophile Freetext 45 45 45 HIV Genotype - drug resistance report Hep C Ab 05/16/10 07:00 06/15/10 21:00 06/15/10 21:00 that guides physician towards alternative 250 280 280 Hep B DNA 06/16/10 07:00 06/15/10 21:00 06/15/10 21:00 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		V
	Current	Summation
Resistance Associated RT Mutations	M41LP, E44D, D67N, T69D, V1181*,	M41L*, E44D, D67N, T69D, V1181*,
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	M421L*, E44D, D67N, T69D,	L90M, G17G, Q18Q, L76L, S37N,
	V1181*, M184V*, L210W* ,	T69D, V1181*, M184V*, T215Y*
	T215Y*	
Resistance Associated PR Mutations	L101/V, K20R, M36I, M46I, F53L,	M36I, M46I, F53L, 154V, A71V,
	154V, Q58E, A71V, V82T, I84V	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

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What could be done TOMORROW?

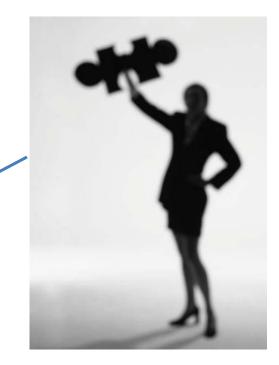
- Link storage of DNA sequencing output to EHR
 - Some lessons from large image files that are stored in archive approach
 - Can be compressed by storing differences relative to a reference sequence
- Assist diagnostic expert with interpretation of DNA sequence results (current or NextGEN)
 - Highlight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



CTTCTCCGTGTCCACCTTGCGCAACTTGGG CCTGGGCAAGAAGTCGCTGGAGCAGTGG GTGACCGAGGAGGCCGCCTGCCTTTGTGC CGCCTTCGCCGACCAAGCCGG

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

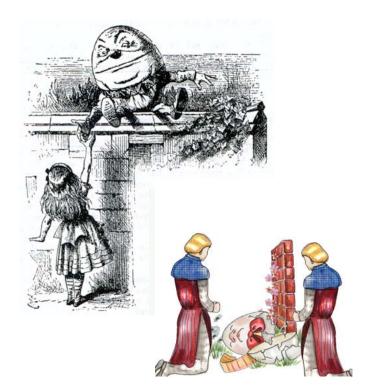


The problem with codifying after the result is captured....

 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

6

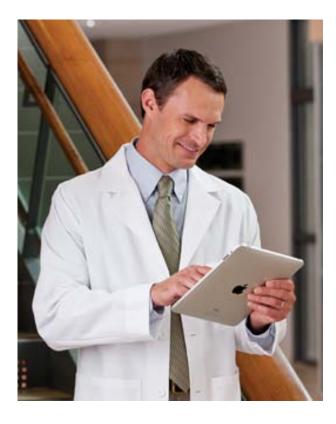
"That it will ever come into general use, notwithstanding its value, is extremely doubtful because its beneficial application requires much time and gives a good bit of trouble, both to the patient and to the practitioner because its hue and character are foreign and opposed to all our habits and associations."

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

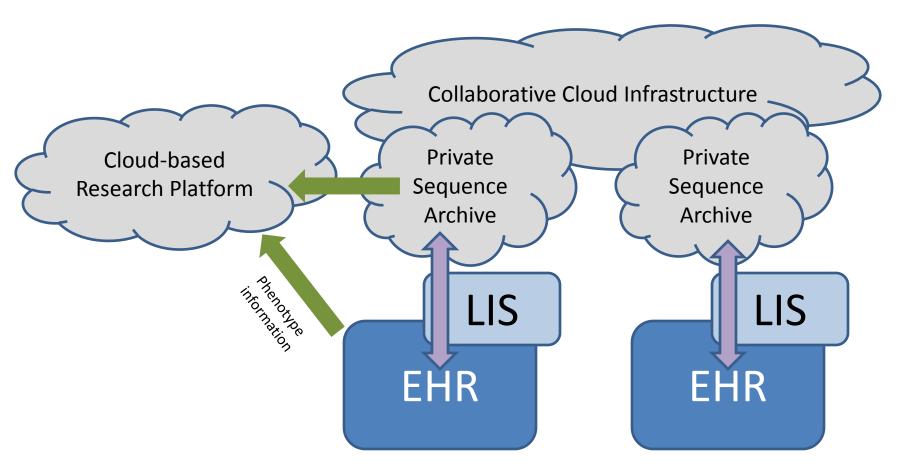
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Some lessons and questions from imaging

- 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!

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