We built it but will they come?

Mark Hoffman, Ph.D.
Chief Research Information Officer
Children’s Mercy Research Institute

@markhoffmankc
Nearly 20 years ago….

- Filed patent on Genomic CDS
  April 20, 2001

- Core concept: Alert user if clinical order is contraindicated by genetic information.
We built it …

- Lab information system (LIS)
- Standardized terminology (Clinical Bioinformatics Ontology)
- Genomics CDS strategy

… They came

- Molecular diagnostics and cytogenetics labs
- Some genetics specialists
- Oncology

… They didn’t

- Most generalists
- Most specialties
Periods of EHR / Genomics

2000-2008
• What is genomics?
• We don't even have an EMR...

2009 - 2015
• Hurray - now we have an EMR!
• But wait - we have to fulfill the MU requirements and can't deal with “this”.

2016 - present
• So many options
• Will this startup survive?
• Still seen as academic
• Patient portal is real
• How will this help with COVID-19?

Lab Systems – automation
Ground up build discrete data
Niche applications
Children’s Mercy Genomic Medicine Center

- **Goal:** 30,000 children + family members
- 100k genomes + health information

### Genomic Answers for Kids project progress

We are working hard to make progress for patients with genetic disease!

- 2,149 families enrolled
- 4,987 individuals enrolled
- 5,624 genomic analyses
- 897,614 gigabases sequenced*
- 213 diagnoses from study

Most important stakeholders
Common architecture: Current state of clinical genomics / informatics

A pdf is a **dead end** for decision support, re-analysis
Optimizing resource allocation

Direction of funding:
• FTE - manual EHR build for discrete storage

Gaps:
• Increase utilization among providers (demand)
• Human factors (!!!)
• Confidence in reimbursement
• FHIR in practice
• Consistent storage of clinically actionable variant data

Direction of funding:
• Cloud storage, analytics
• Next next generation seq (long read etc…)
• 3rd party niche applications

Gaps:
• Standard clinical interpretations
• Policy clarity around automated re-interpretation of results – billable?
Difficult observations from the field

• **FHIR** has a lot of promise and potential as a data standard but still early
• Content standards about variant interpretation still limited
• **Strong reluctance** in health systems to use OS in production systems.
• Labs willing to pay for 3rd party niche applications
• Providers much less willing to pay for EHR-adjacent genomics CDS
How do we rebalance?

• Train providers – they must see the value every day and be comfortable

• **HUMAN FACTORS:** Work on the “experience” side of the equation

• **Content:** CPIC like resource for clinical genetics

• **Economics:** Demonstrate financial benefits of using genomic info to improve care

• EHR vendors respond to their clients: Reward providers for using genomics standards in EHR
I’m still cautiously optimistic!

- Mark Hoffman, Ph.D.
- mhoffman@cmh.edu
- @markhoffmankc