Intermountain Healthcare

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Director, Clinical Genetics Institute
April 27, 2011

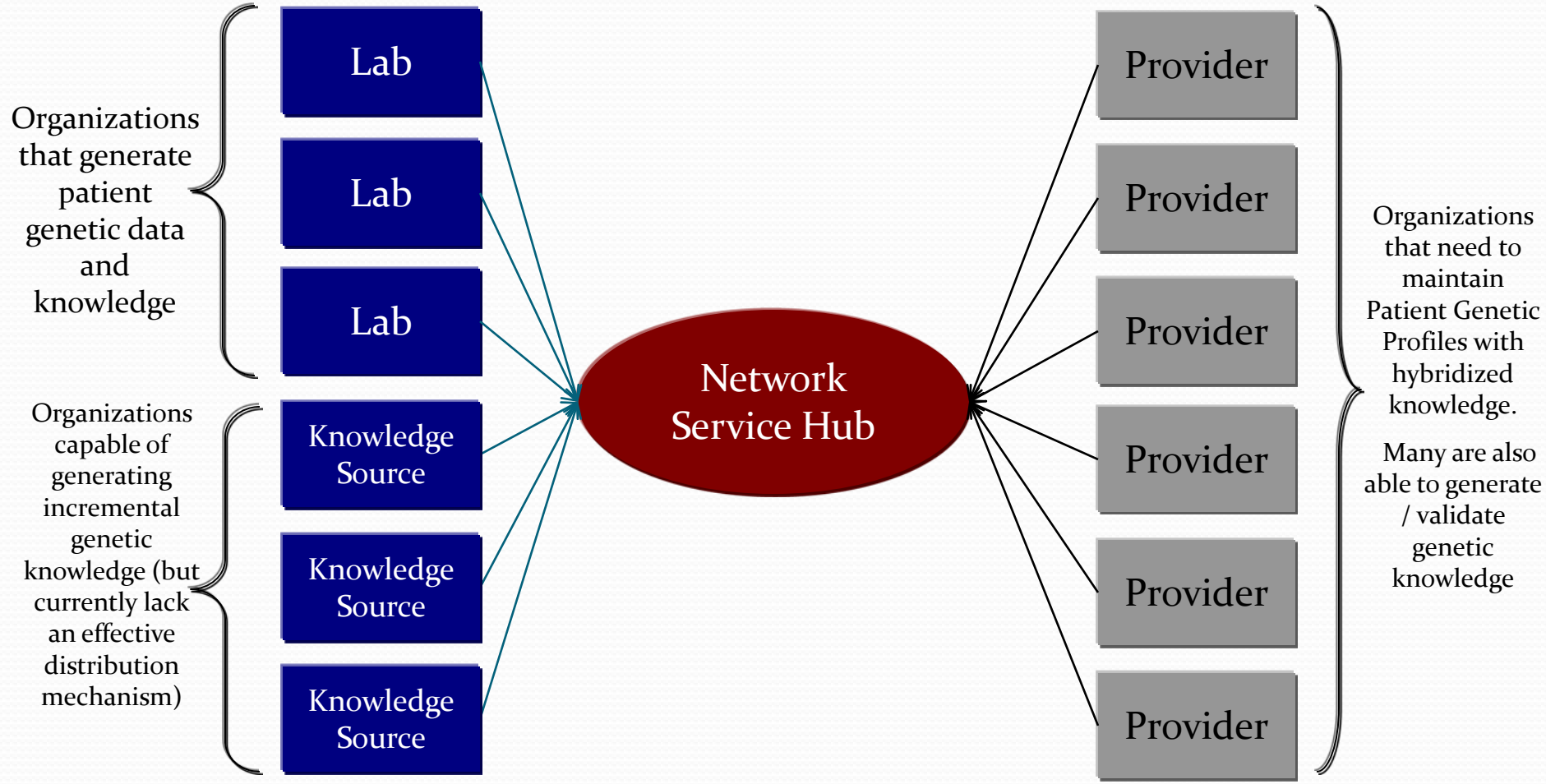
Disclosures

- No financial disclosures
- Will discuss a proprietary product as we are using this in our system
  - Not an endorsement of this particular product

Challenge

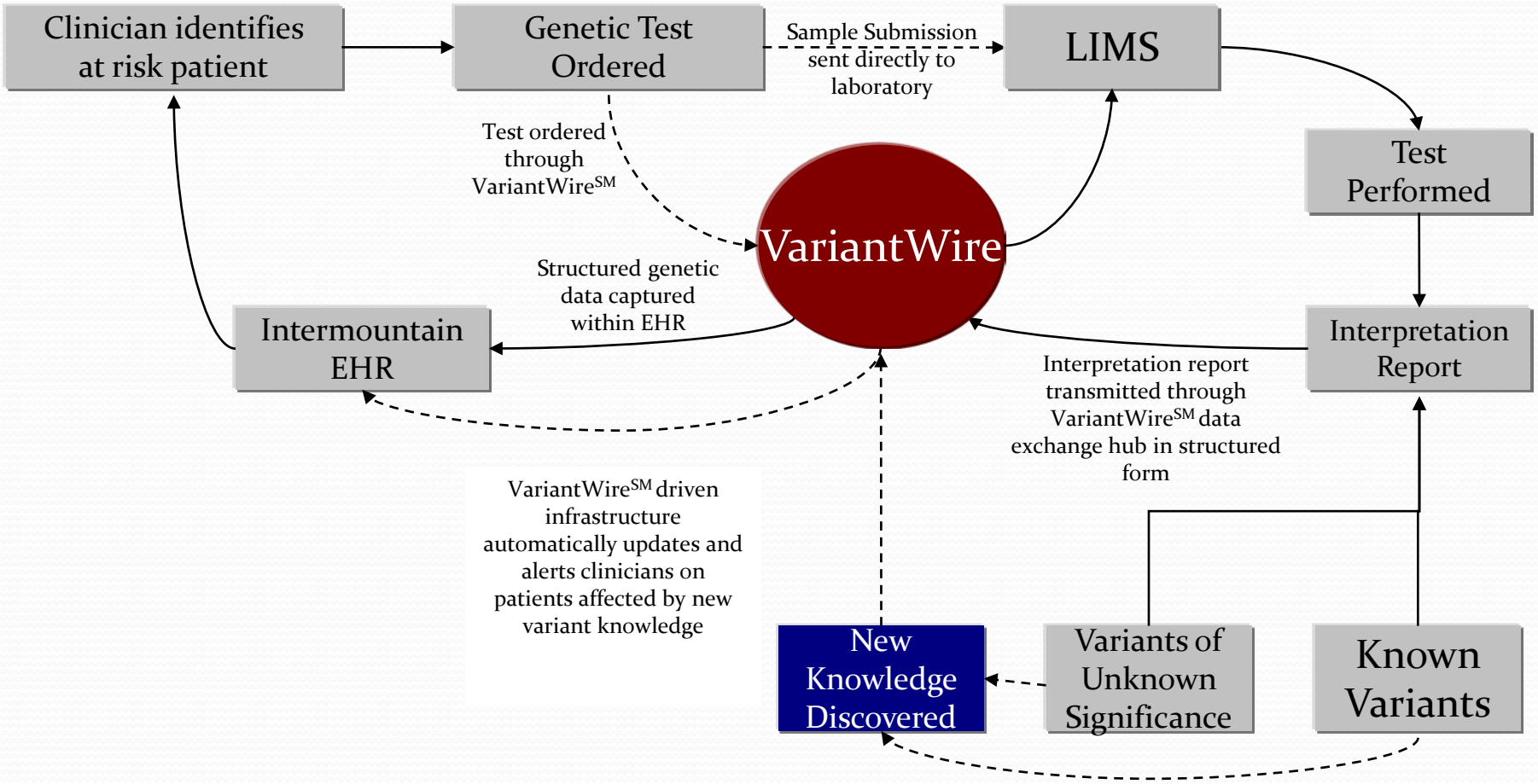
- Genomic test communication
  - Usually text-based or image of report
  - Data not captured in coded and computable format
  - Unable to link interpretive help, clinical decision support to genetic test result
  - How to update interpretation of variants as more knowledge becomes available
  - Linking clinics to laboratories and knowledge repositories
    - Near infinite number of interfaces to build

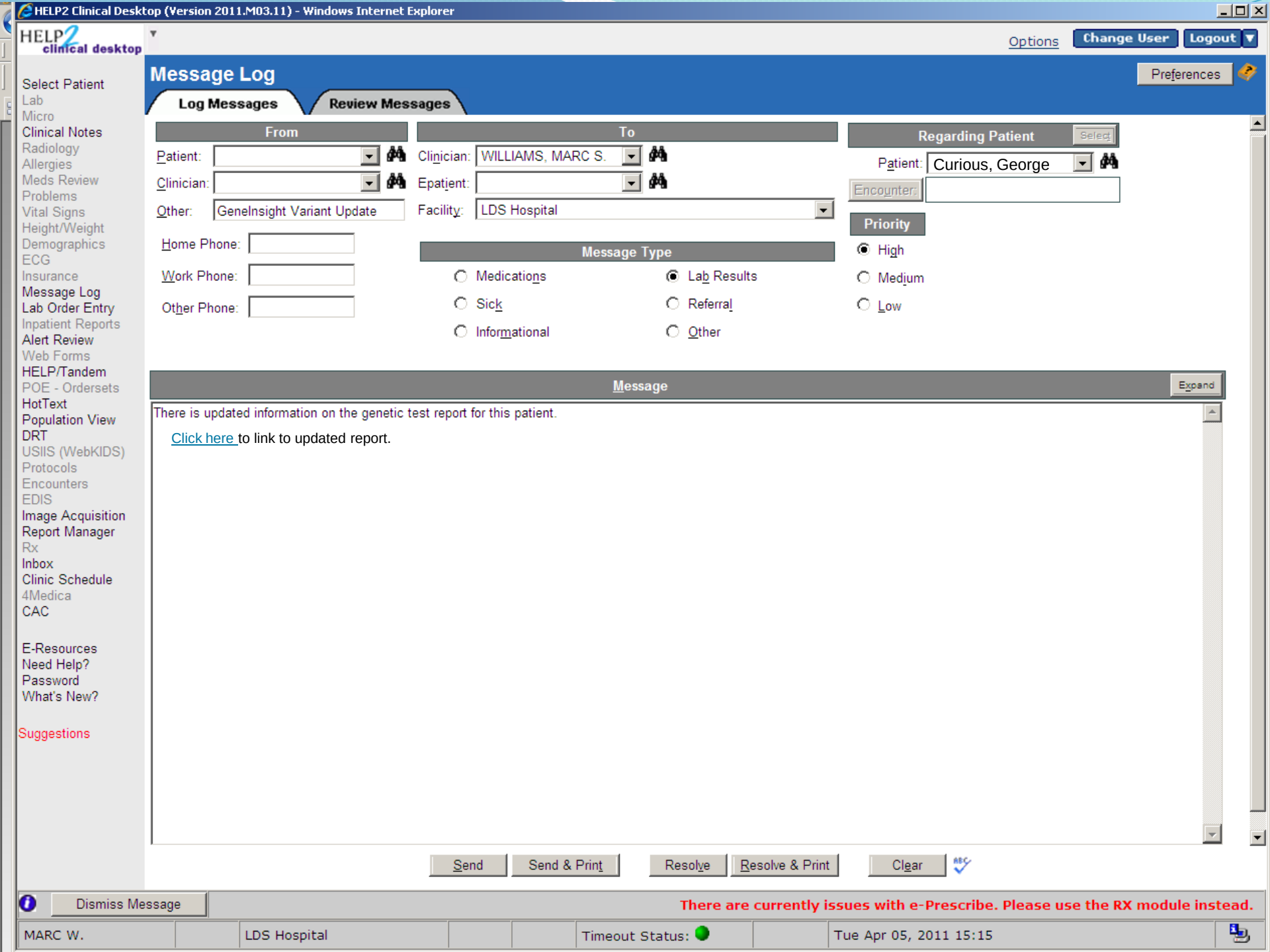
Network Exchange Model



The Harvard Partners Molecular Genetics laboratory electronically transmitted a clinical genomic test result to the Intermountain Healthcare Electronic Data Warehouse in 2009.

Advanced Genetic Testing Workflow




HELP2 Clinical Desktop (Version 2011.M03.11) - Windows Internet Explorer
HELP2 clinical desktop
Options
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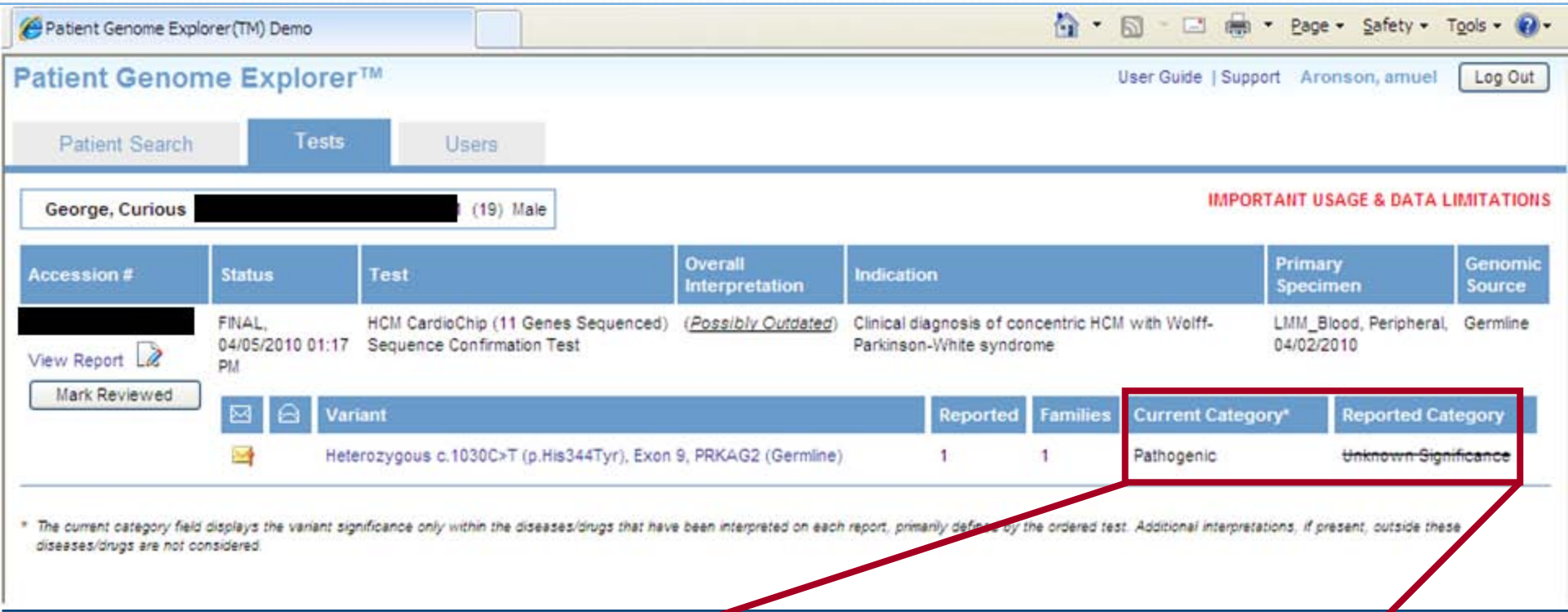
4Medica
CAC
E-Resources
Need Help?
Password
What's New?
Suggestions
Message Log
Preferences
Log Messages
Review Messages
From
Patient:
Clinician:
Other:
GeneInsight Variant Update
Home Phone:
Work Phone:
Other Phone:
To
Clinician:
WILLIAMS, MARC S.
Epatient:
Facility:
LDS Hospital
Message Type
Medications
Lab Results
Sick
Referral
Informational
Other
Regarding Patient
Select
Patient:
Curious, George
Encounter:
Priority
High
Medium
Low
Message
Expand
There is updated information on the genetic test report for this patient.
Click here to link to updated report.
Send
Send & Print
Resolve
Resolve & Print
Clear
Dismiss Message
There are currently issues with e-Prescribe. Please use the RX module instead.
MARC W.
LDS Hospital
Timeout Status:
Tue Apr 05, 2011 15:15

Variant Knowledge Update



Current Category*	Reported Category
Pathogenic	~~Unknown Significance~~

Updated Variant Information

Patient Genome Explorer™

Individual Reported Variant Interpretation History (Variant 1 of 1)

IMPORTANT USAGE & DATA LIMITATIONS

Warning: This page only lists information on a single variant. This is outside of the patient report context and may be insufficient for re-interpretation of the patient report.

Heterozygous c.1030C>T (p.His344Tyr), Exon 9, PRKAG2 (Germline)

Report [redacted] (FINAL, 04/05/2010 01:17 PM), HCM CardioChip (11 Genes Sequenced), Sequence Confirmation Test

Patient George, Curious[redacted] (19) Male

Current Category* Pathogenic (**Reported:** ~~Unknown Significance~~)

Counts Reports (1), Families (1)

Alerts

Status	!	Date	Type	Message
Unreviewed	!	04/06/2010 10:27 AM	Non-incidental Level Change	The category for the PRKAG2 c.1030C>T (p.His344Tyr) association to HCM changed from Unknown Significance to Pathogenic.

Mark Reviewed

Current Knowledge** Approved 04/05/2010 01:22 PM by Matthew Varugheese

Diseases/Drugs	Category	Variant Interpretation
HCM	Pathogenic	The His344Tyr variant has not been reported in the literature nor previously identified in our laboratory. The His344 residue is well conserved from fruitfly to mammals, and the His344Tyr variant occurs within the CBS domain region where all pathogenic PRKAG2 variants have been identified to date. In addition, the presence of concentric HCM and Wolff-Parkinson-White syndrome in the first proband identified with this mutation, which are clinical features consistent with PRKAG2 mutations, as well as follow-up testing showing that the variant arose de novo, provide strong support for this variant being pathogenic.

* *The current category field displays the variant significance only within the diseases/drugs that have been interpreted on each report, primarily defined by the ordered test. Additional interpretations, if present, outside these diseases/drugs are not considered.*

** *The Current Knowledge only includes the following Diseases/Drugs Interpreted on Report: HCM, DCM, LVNC, RCM, Danon disease, myopathy, Fabry disease, ARVD/C, Barth syndrome*

Next Steps

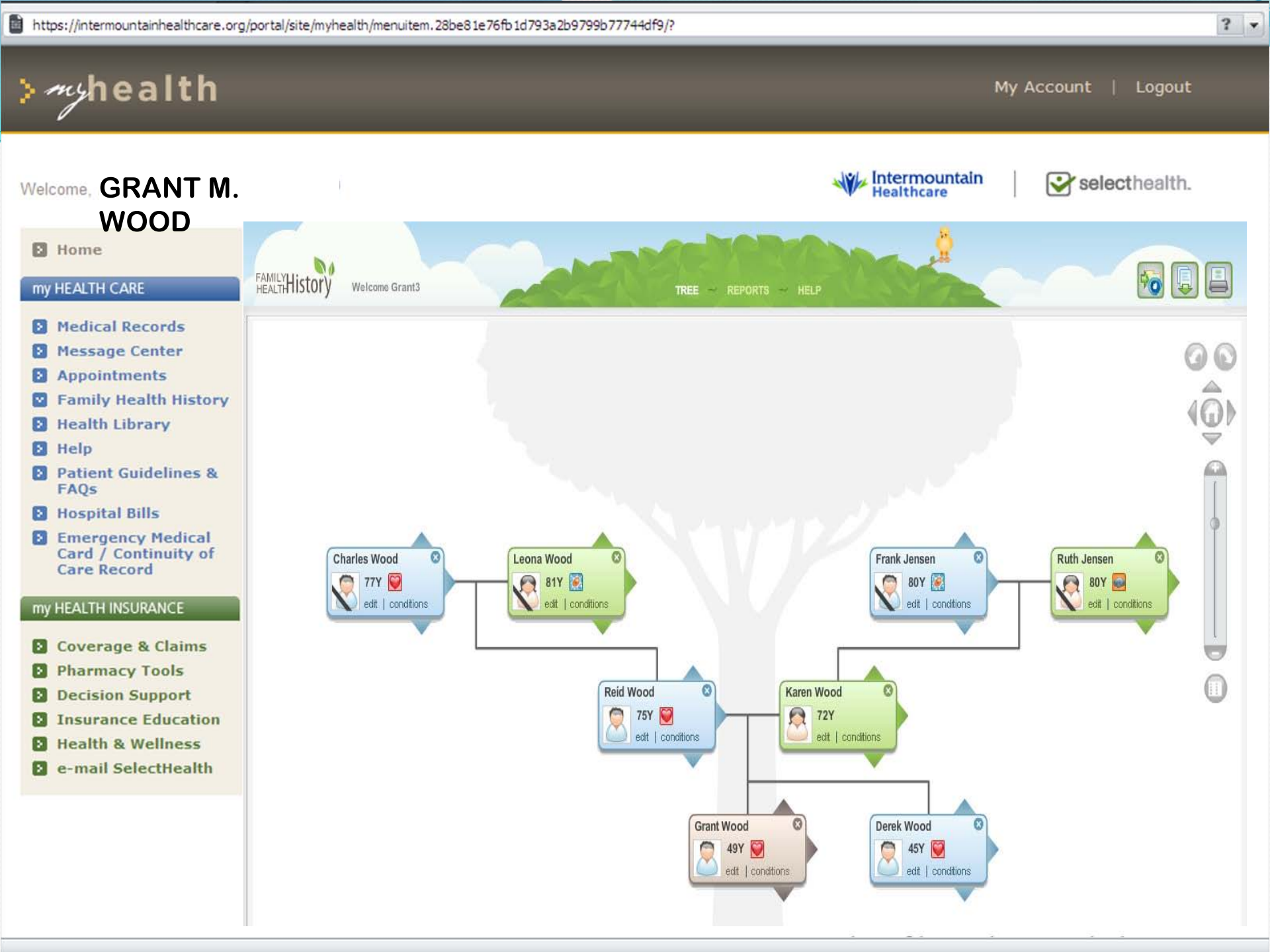
- Primary genetic referral laboratory completing installation of hub
- Will create and test interface with Intermountain EDW
  - HFE gene
- Expand to all genetic tests from this laboratory
- Link information and clinical decision support to test order entry and laboratory results

Challenge

- Optimize use of family history in clinical practice
- Barriers to use
  - Time to collect
  - Patient knowledge about family history
  - How to interpret family history
  - How to use information to change patient care
  - Questions about utility of information
- Potential model—Surgeon General's tool

Empowering Patients-Family History

- How many have completed the Surgeon General's Family History tool?
- How many found out things about their family they didn't know before?
- How many have brought it to their provider to discuss?


https://intermountainhealthcare.org/portal/site/myhealth/menuitem.28be81e76fb1d793a2b9799b77744df9/?
myhealth
My Account | Logout
Welcome, GRANT M. WOOD
Intermountain Healthcare
selecthealth.
Home
my HEALTH CARE
Medical Records
Message Center
Appointments
Family Health History
Health Library
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Patient Guidelines & FAQs
Hospital Bills
Emergency Medical Card / Continuity of Care Record
my HEALTH INSURANCE
Coverage & Claims
Pharmacy Tools
Decision Support
Insurance Education
Health & Wellness
e-mail SelectHealth
FAMILY HEALTH History
Welcome Grant3
TREE ~ REPORTS ~ HELP
Charles Wood 77Y edit | conditions
Leona Wood 81Y edit | conditions
Frank Jensen 80Y edit | conditions
Ruth Jensen 80Y edit | conditions
Reid Wood 75Y edit | conditions
Karen Wood 72Y edit | conditions
Grant Wood 49Y edit | conditions
Derek Wood 45Y edit | conditions

Select Patient
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Image Acquisition
Report Manager
Rx
Inbox
Clinic Schedule

E-Resources
Need Help?
Password
CD Info

Comments

MESSAGE LOG

	Pri	Log Time	Type	To	Patient
☐	H	8/03/2010	FHH	WILLIAMS, MARC S.	WOOD, GRANT
☐	M	11/03/08 08:43	Meds	WILLIAMS, MARC S.	DEVTEST, KARL
☐	L	11/01/08 12:41	Order	WILLIAMS, MARC S.	TESTUSER, CLINT
☐	M	10/31/08 14:40	All	WILLIAMS, MARC S.	PASS, EDWIN
☐	M	10/30/08 10:40	Other	WILLIAMS, MARC S.	PROBAND, JAMES
☐	L	10/28/08 15:22	Rem	WILLIAMS, MARC S.	CLEPT, SYLVIA

Patient: Wood, Grant
MRN#: 232445
Contact#: H: (801) 555-1212 W:(801) 555-1234
Clinician: Williams, Marc S.

>>> >Entered By: OurFamilyHealth 8/3/10 14:41:47 >>>>

A patient has logged in to his/her MyHealth patient portal account, and has completed a family health history record that shows an increased risk for a familial disease. The family history and risk assessment are available for your review.

View as table | View as pedigree

Select Patient
Lab
Micro
Clinical Notes
Radiology
Allergies
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FAMILY HEALTH HISTORY

Upload to Webform

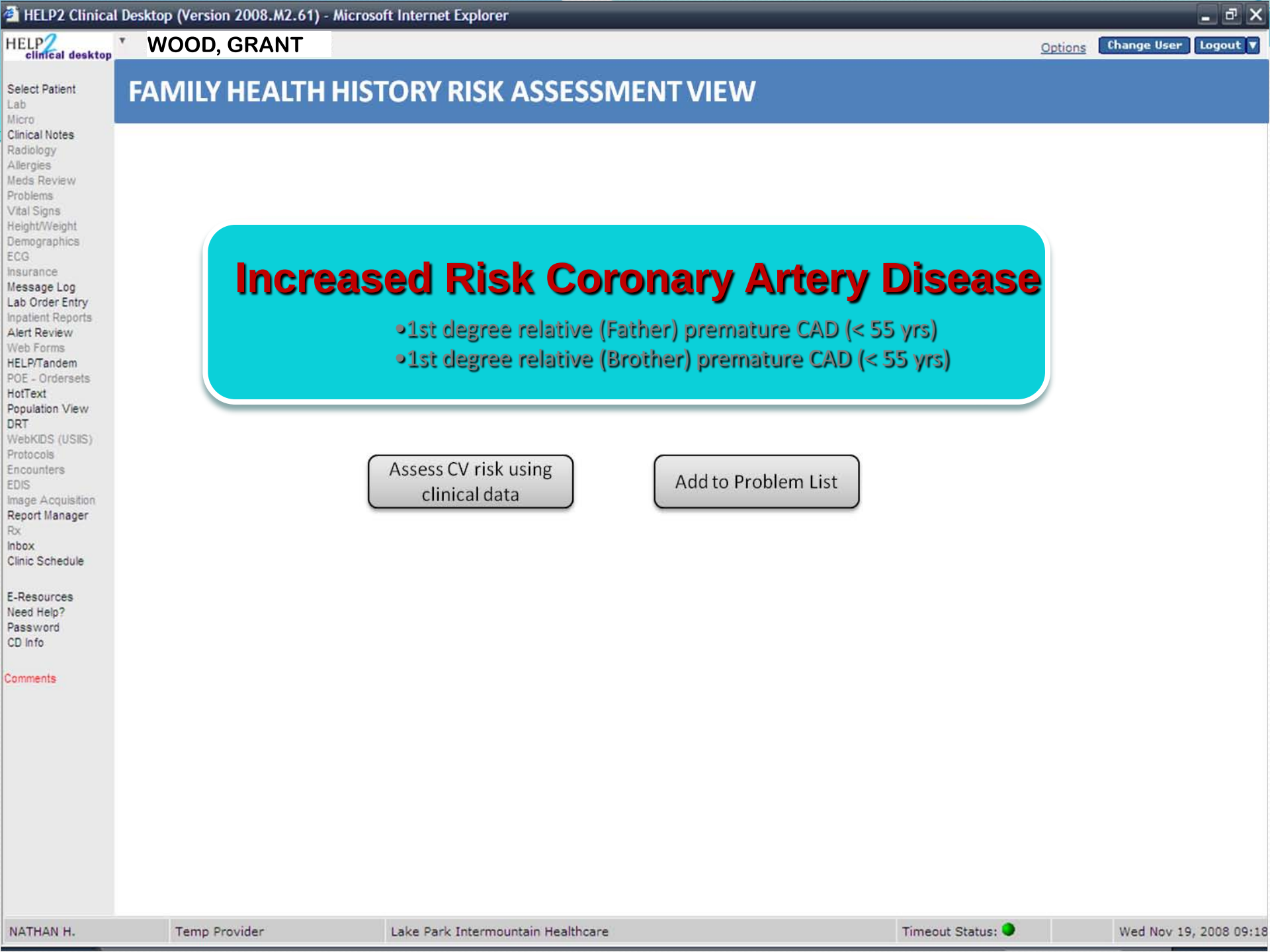
Perform risk assessment

Chart Report
Grant Wood – August 3, 2010

View as pedigree

My Stats: Age: 43

	Still Living?	Heart Disease	Stroke	Diabetes	Colon Cancer	Breast Cancer	Ovarian Cancer	Prostate Cancer	Hypertension	Additional Diseases
Austin Proband [Me]	Yes									
[Mother]	Yes									
[Mother's Mother]	No									
[Mother's Father]	No						N/A	Yes age 65		
[Father]	Yes	Yes age 55	No	No	Yes, 60 and older		N/A	No	No	
[Father's Mother]	No									
[Father's Father]	No		Yes age 77				N/A			
[Daughter]	Yes									
[Son]	Yes						N/A		Yes, At age 30-39	
[Sister]	Yes									
[Sister]	Yes			Yes, At age 40-49						
[Brother]	Yes	Yes age 41					N/A			


HELP2 Clinical Desktop (Version 2008.M2.61) - Microsoft Internet Explorer
HELP2 clinical desktop
WOOD, GRANT
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FAMILY HEALTH HISTORY RISK ASSESSMENT VIEW
Increased Risk Coronary Artery Disease
•1st degree relative (Father) premature CAD (< 55 yrs)
•1st degree relative (Brother) premature CAD (< 55 yrs)
Assess CV risk using clinical data
Add to Problem List
NATHAN H.
Temp Provider
Lake Park Intermountain Healthcare
Timeout Status:
Wed Nov 19, 2008 09:18

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FAMILY HEALTH HISTORY RISK ASSESSMENT VIEW

Recommendation for use of inflammatory marker (IM) (Hs-CRP and Lp-PLA_2) testing

ASSESS CV RISK

ATP III Risk Factors See ATP III Report

- Cigarette smoking
- **Hypertension 140/90 on 1/18/2010**
- **Low HDL cholesterol (male <40mg/dL, female <50mg/dL) 37 mg/dL on 11/25/2009**
- **Family History of premature CAD**
- Age (men less than 55 years, women less than 65 years)

CAD Risk Equivalents

- Other clinically manifest forms of atherosclerotic disease (peripheral arterial disease, abdominal aortic aneurism, and carotid artery disease, [eg. TIA or stroke])
- Diabetes
- Chronic kidney disease
- Ankle-brachial index <0.9
- >50% carotid stenosis

Low CV Risk	**Moderate CV Risk**	High CV Risk	Very High CV Risk
0-1 risk factors	**2+ risk factors**	CAD, or CAD risk equivalent	

Consider inflammatory marker test to re-classify risk

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INFLAMMATORY MARKER (IM) TEST

Hs-CRP >2 mg/L or Lp-PLA_2>200 ng/mL in individuals with moderate or high risk warrants reclassification See references

TREAT To LDL-C Goal

- Intensify treatment of non-lipid risk factors
- Therapeutic lifestyle change See references

Low CV Risk 0-1 risk factors	**Moderate CV Risk 2+ risk factors**	High CV Risk CAD, or CAD risk equivalent	Very High CV Risk

Schedule patient for appointment

Last seen on: 7/18/2010

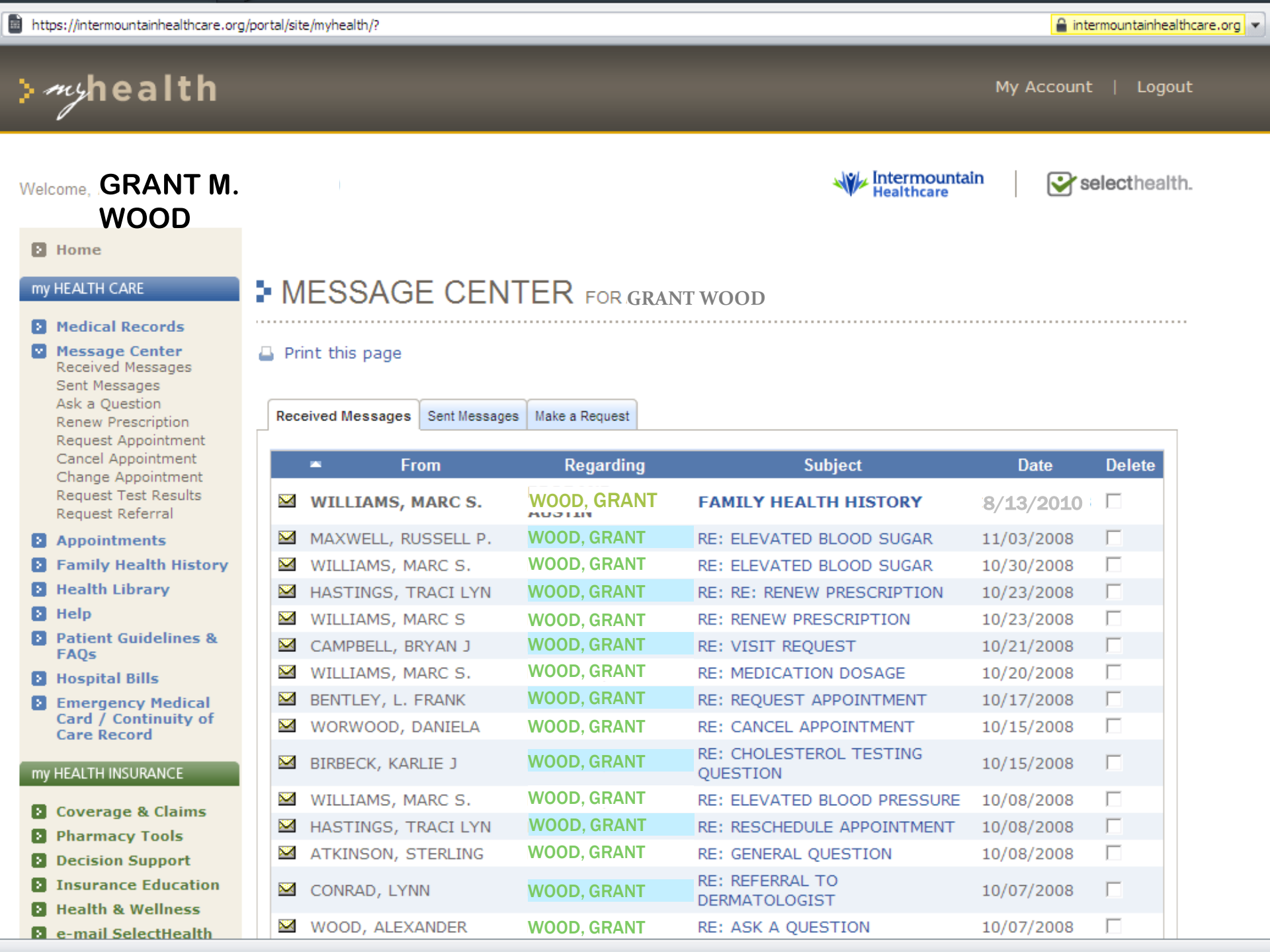
IM TEST

Order test and notify patient

Not Elevated

Elevated

LDL – C Goal < 160 mg/dL	**LDL – C Goal < 130 mg/dL**	**LDL – C Goal < 100 mg/dL**	LDL – C Goal < 70 mg/dL


https://intermountainhealthcare.org/portal/site/myhealth/?
intermountainhealthcare.org
myhealth
My Account | Logout
Welcome, GRANT M. WOOD
Intermountain Healthcare
selecthealth.
Home
my HEALTH CARE
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Received Messages
Sent Messages
Ask a Question
Renew Prescription
Request Appointment
Cancel Appointment
Change Appointment
Request Test Results
Request Referral
Appointments
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Hospital Bills
Emergency Medical Card / Continuity of Care Record
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Coverage & Claims
Pharmacy Tools
Decision Support
Insurance Education
Health & Wellness
e-mail SelectHealth
MESSAGE CENTER FOR GRANT WOOD
Print this page
Received Messages
Sent Messages
Make a Request
From
Regarding
Subject
Date
Delete
WILLIAMS, MARC S.
WOOD, GRANT
FAMILY HEALTH HISTORY
8/13/2010
MAXWELL, RUSSELL P.
WOOD, GRANT
RE: ELEVATED BLOOD SUGAR
11/03/2008
WILLIAMS, MARC S.
WOOD, GRANT
RE: ELEVATED BLOOD SUGAR
10/30/2008
HASTINGS, TRACI LYN
WOOD, GRANT
RE: RE: RENEW PRESCRIPTION
10/23/2008
WILLIAMS, MARC S
WOOD, GRANT
RE: RENEW PRESCRIPTION
10/23/2008
CAMPBELL, BRYAN J
WOOD, GRANT
RE: VISIT REQUEST
10/21/2008
WILLIAMS, MARC S.
WOOD, GRANT
RE: MEDICATION DOSAGE
10/20/2008
BENTLEY, L. FRANK
WOOD, GRANT
RE: REQUEST APPOINTMENT
10/17/2008
WORWOOD, DANIELA
WOOD, GRANT
RE: CANCEL APPOINTMENT
10/15/2008
BIRBECK, KARLIE J
WOOD, GRANT
RE: CHOLESTEROL TESTING QUESTION
10/15/2008
WILLIAMS, MARC S.
WOOD, GRANT
RE: ELEVATED BLOOD PRESSURE
10/08/2008
HASTINGS, TRACI LYN
WOOD, GRANT
RE: RESCHEDULE APPOINTMENT
10/08/2008
ATKINSON, STERLING
WOOD, GRANT
RE: GENERAL QUESTION
10/08/2008
CONRAD, LYNN
WOOD, GRANT
RE: REFERRAL TO DERMATOLOGIST
10/07/2008
WOOD, ALEXANDER
WOOD, GRANT
RE: ASK A QUESTION
10/07/2008

https://intermountainhealthcare.org/portal/site/myhealth/? intermountainhealthcare.org

myhealth

My Account | Logout

Welcome, **GRANT M. WOOD**

Intermountain Healthcare | selecthealth.

- Home

my HEALTH CARE

- Medical Records
- Message Center
  - Received Messages
  - Sent Messages
  - Ask a Question
  - Renew Prescription
  - Request Appointment
  - Cancel Appointment
  - Change Appointment
  - Request Test Results
  - Request Referral
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- Health Library
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- Patient Guidelines & FAQs
- Hospital Bills
- Emergency Medical Card / Continuity of Care Record

my HEALTH INSURANCE

- Coverage & Claims
- Pharmacy Tools
- Decision Support
- Insurance Education
- Health & Wellness
- e-mail SelectHealth

MESSAGE CENTER FOR GRANT WOOD

Print this page

Back Print Delete Reply

Subject: FAMILY HEALTH HISTORY
Date: 8/13/2010
From: WILLIAMS, MARC S.
Message:

We have reviewed your updated family history. We have identified increased risk for cardiovascular disease in your family. This information, when combined with your personal risk factors, places you in the moderate risk category for cardiovascular disease. There is a blood test that can help us determine treatment goals that can reduce your chances of having a problem. I have placed an order for this test in the system. Please contact our scheduling clerk at 801-555-2121 to make arrangements to have the test performed. I will contact you with the results. Please call or e-mail if you have any questions.

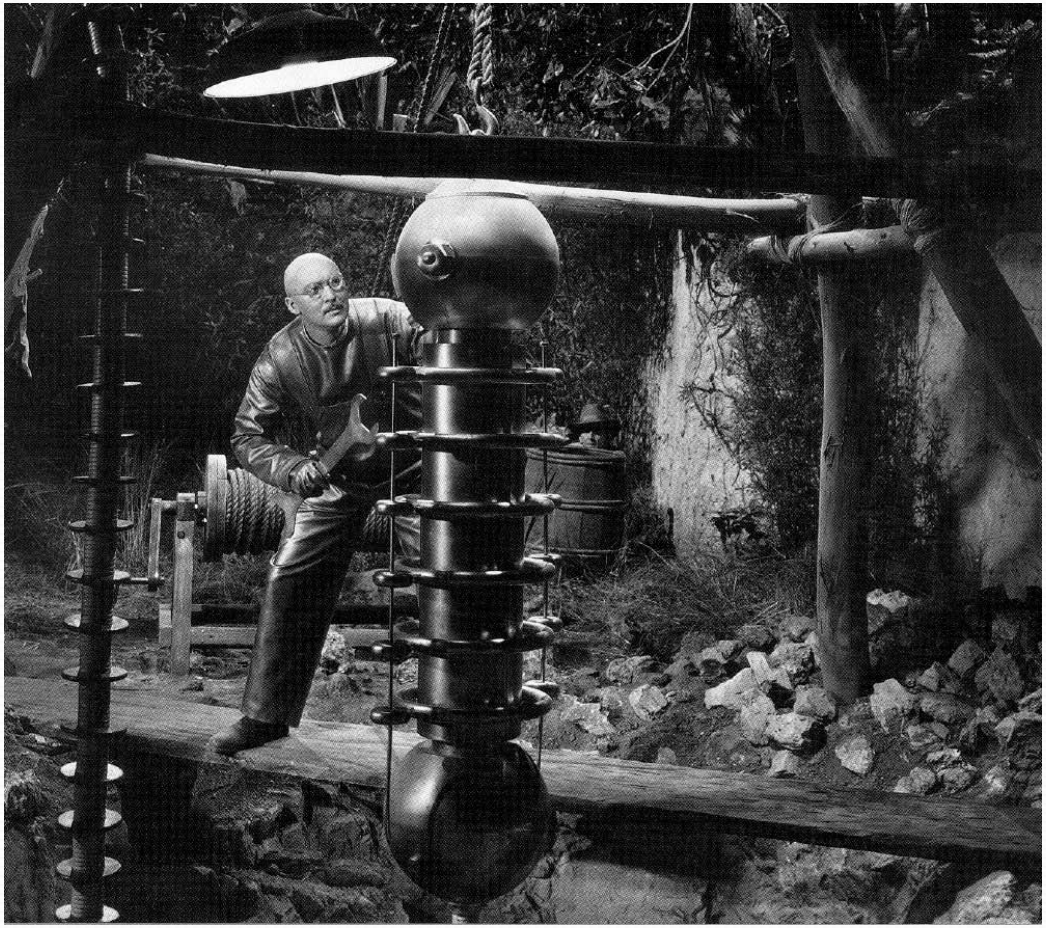
Thank you,

Dr. Marc S. Williams

Next Steps

- Fully deploy FH tool in patient portal
- Enhance patient education resources in tool (infobuttons)
- Build risk assessment modules to run against tool
- Pilot patient/provider communication in selected 'e-clinics' interested in FH
  - Negotiated patient visits
- Create and implement targeted tools
  - Breast/Ovarian Cancer tool in multi-disciplinary cancer clinics
  - Colorectal tool for patients in hospital for resection
- Test genealogic approaches to build and adjudicate FH

Clinical Genetics Institute



http://intermountainhealthcare.org/services/genetics/Pages/home.aspx