The PREDICT program: Implementing prospective pharmacogenetics for inpatient and outpatient clinical care

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Disclaimers

I receive funding from:

- NIH: NLM, NHGRI, NIGMS, NCI, NCATS
- Reynolds Foundation (Geriatrics Education)
- National Board of Medical Examiners
The vision

"Here's my sequence..."

New Yorker, 2000
How will this vision actually start to be tested and become reality?

"Here's my sequence..."

New Yorker, 2000

- Biomedical research
- Commitment to information technology
- Harnessing the healthcare system for discovery
- Ability to nimbly adapt a healthcare system to evolving evidence
EHR feeds both discovery and implementation

**Discovery**

Vanderbilt *BioVU*

De-identified DNA repository

>180k samples

**Implementation**

**PREDICT**

- CLIA genomics lab
- Integrated decision support for genomics
- Predictive algorithms on who to test
- Genomic databases
- Track outcomes
Vanderbilt BioVU: an Opt-Out DNA Biobank

Extracting DNA from leftover blood samples

DNA Research
Leftover blood from tests, treatment, or surgery may also be used for DNA research through the Vanderbilt BioVU Program. If I do not want my leftover blood to go to the Vanderbilt BioVU Program for DNA research, I must check the box below. If I have questions or want further information on BioVU, I may call 866-436-4710.

☐ I do NOT want blood left over from my tests, treatment, or surgery to be used for the Vanderbilt BioVU Program for DNA research

Please click "Next" and write your name on the next screen.

Cancel

Next

COPIES OF THE FORMS YOU SIGN ARE AVAILABLE UPON REQUEST
One way hash

John Doe

~2 million records

The Synthetic Derivative: updated regularly
John Doe

eligible?

One way hash

Extract DNA

BioVU

~180,000 DNAs

~2 million records

The Synthetic Derivative:
updated regularly
BioVU as a resource for discovery

<table>
<thead>
<tr>
<th>Disease</th>
<th>Marker</th>
<th>Gene / Region</th>
</tr>
</thead>
<tbody>
<tr>
<td>Atrial fibrillation</td>
<td>rs2200733</td>
<td>Chr. 4q25</td>
</tr>
<tr>
<td></td>
<td>rs10033464</td>
<td>Chr. 4q25</td>
</tr>
<tr>
<td>Crohn's disease</td>
<td>rs11805303</td>
<td>IL23R</td>
</tr>
<tr>
<td></td>
<td>rs17234657</td>
<td>Chr. 5</td>
</tr>
<tr>
<td></td>
<td>rs1000113</td>
<td>Chr. 5</td>
</tr>
<tr>
<td></td>
<td>rs17221417</td>
<td>NOD2</td>
</tr>
<tr>
<td></td>
<td>rs2542151</td>
<td>PTPN22</td>
</tr>
<tr>
<td>Multiple sclerosis</td>
<td>rs3135388</td>
<td>DRB1*1501</td>
</tr>
<tr>
<td></td>
<td>rs2104286</td>
<td>IL2RA</td>
</tr>
<tr>
<td></td>
<td>rs6897932</td>
<td>IL7RA</td>
</tr>
<tr>
<td>Rheumatoid arthritis</td>
<td>rs6457617</td>
<td>Chr. 6</td>
</tr>
<tr>
<td></td>
<td>rs6679677</td>
<td>RSBN1</td>
</tr>
<tr>
<td></td>
<td>rs2476601</td>
<td>PTPN22</td>
</tr>
<tr>
<td>Type 2 diabetes</td>
<td>rs4506565</td>
<td>TCF7L2</td>
</tr>
<tr>
<td></td>
<td>rs12255372</td>
<td>TCF7L2</td>
</tr>
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<td></td>
<td>rs12243326</td>
<td>TCF7L2</td>
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<td></td>
<td>rs10811661</td>
<td>CDKN2B</td>
</tr>
<tr>
<td></td>
<td>rs8050136</td>
<td>FTO</td>
</tr>
<tr>
<td></td>
<td>rs5219</td>
<td>KCNJ11</td>
</tr>
<tr>
<td></td>
<td>rs5215</td>
<td>KCNJ11</td>
</tr>
<tr>
<td></td>
<td>rs4402960</td>
<td>IGF2BP2</td>
</tr>
</tbody>
</table>

Ritchie et al., AJHG 2010
The eMERGE Network

electronic Medical Records & Genomics

A consortium of biorepositories linked to electronic medical records data for conducting genomic studies

eMERGE goals

- To perform GWAS using EMR-derived phenotypes
- To initiate implementation of actionable variants into the EMR

Coordinating Center

☆: pediatric sites
An eMERGE-wide phenotype analyzed with no extra genotyping: hypothyroidism

European Americans (1,306 cases and 5,013 controls)
Another perception of genomic medicine - pharmacogenomics
Variable drug response is common; Genetics may predict this too
<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Cases</th>
<th>Controls</th>
<th>% Reuse</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clopidogrel in CV disease</td>
<td>225</td>
<td>468</td>
<td>27%</td>
</tr>
<tr>
<td>Warfarin stable dose</td>
<td>1,167</td>
<td>N/A</td>
<td>47%</td>
</tr>
<tr>
<td>Early Repolarization</td>
<td>544</td>
<td>2,609</td>
<td>28%</td>
</tr>
<tr>
<td>Vancomycin stable dose</td>
<td>1,067</td>
<td>N/A</td>
<td>42%</td>
</tr>
<tr>
<td>C. difficile colitis</td>
<td>941</td>
<td>1,710</td>
<td>28%</td>
</tr>
<tr>
<td>Anthracycline cardiomyopathy</td>
<td>528</td>
<td>N/A</td>
<td>39%</td>
</tr>
<tr>
<td>Guillain-Barre Syndrome</td>
<td>97</td>
<td>6,536</td>
<td>99%</td>
</tr>
<tr>
<td>Heart Transplant</td>
<td>181</td>
<td>N/A</td>
<td>42%</td>
</tr>
<tr>
<td>Kidney transplant</td>
<td>1,078</td>
<td>N/A</td>
<td>42%</td>
</tr>
<tr>
<td>Clopidogrel in strokes/TIAs</td>
<td>6</td>
<td>123</td>
<td>22%</td>
</tr>
<tr>
<td>Statin-related myopathy</td>
<td>11</td>
<td>4,342</td>
<td>100%</td>
</tr>
<tr>
<td>Heparin-induced thrombocytopenia</td>
<td>73</td>
<td>2,300</td>
<td>99%</td>
</tr>
<tr>
<td>CV events with COX2 therapy</td>
<td>85</td>
<td>395</td>
<td>34%</td>
</tr>
<tr>
<td>Serious bleeding during warfarin</td>
<td>259</td>
<td>276</td>
<td>43%</td>
</tr>
<tr>
<td>Amiodarone toxicity (lung, thyroid)</td>
<td>97</td>
<td>343</td>
<td>34%</td>
</tr>
<tr>
<td>Chronic inflammatory polyneuropathy</td>
<td>12</td>
<td>14,000*</td>
<td>100%</td>
</tr>
<tr>
<td>Rheumatic Heart Disease</td>
<td>108</td>
<td>3,464</td>
<td>98%</td>
</tr>
<tr>
<td>ACEi cough</td>
<td>1,174</td>
<td>978</td>
<td>52%</td>
</tr>
<tr>
<td>Fluoroquinolones and tenopathy</td>
<td>87</td>
<td>537</td>
<td>90%</td>
</tr>
<tr>
<td>Warfarin stable dose in children</td>
<td>92</td>
<td>N/A</td>
<td>28%</td>
</tr>
<tr>
<td>Metformin efficacy</td>
<td>80</td>
<td>N/A</td>
<td>35%</td>
</tr>
<tr>
<td>Metformin and cancer</td>
<td>619</td>
<td>421</td>
<td>83%</td>
</tr>
<tr>
<td>Bisphosphonates and Atypical Fracture/Jaw Osteonecrosis</td>
<td>16</td>
<td>1,454</td>
<td>99%</td>
</tr>
<tr>
<td>Wolff-Parkinson-White</td>
<td>197</td>
<td>5,551</td>
<td>97%</td>
</tr>
<tr>
<td>Steroid-induced Osteonecrosis</td>
<td>83</td>
<td>352</td>
<td>57%</td>
</tr>
<tr>
<td>Shellfish Anaphylaxis</td>
<td>157</td>
<td>14,000*</td>
<td>99%</td>
</tr>
<tr>
<td>Aspirin Anaphylaxis</td>
<td>101</td>
<td>4,334</td>
<td>98%</td>
</tr>
<tr>
<td>Bell's Palsy#</td>
<td>577</td>
<td>14,000*</td>
<td>97%</td>
</tr>
</tbody>
</table>

Large scale GWAS for drug response discovery: the VESPA project

Vanderbilt Electronic Systems for Pharmacogenomic Assessment

clopidogrel → CYP2C19 → 2-oxoclopidogrel

clopidogrel failure = MI, stroke, revascularization, death following MI or PCI

n=225 cases / 468 controls

Warfarin Pharmacogenetics
Using genetics to predict effective dose

<table>
<thead>
<tr>
<th>SNP (Gene)</th>
<th>Beta</th>
<th>P</th>
</tr>
</thead>
<tbody>
<tr>
<td>rs1057910 (CYP2C9*3)</td>
<td>0.83</td>
<td>2.70x10^{-26}</td>
</tr>
<tr>
<td>rs9934438 (VKORC1)</td>
<td>0.87</td>
<td>4.48x10^{-61}</td>
</tr>
</tbody>
</table>

Ramirez et al. *Pharmacogenomics*. 2012
Two in-progress GWAS of Drug-ADEs from the EHR

ACEI-cough
(NLP of allergy sections, automated)

Heparin-induced thrombocytopenia
(automated+manual review)
Another pathway of adverse effects: Off target effects

Drug

Therapeutic Target

Off-target ADE (SJS, rashes, liver injury, long QT)

• carbamazepine
• clozapine
• haloperidol
• abacavir
• antibiotics
Ancestry matters… Carbamazepine-induced cutaneous adverse drug reactions

HLA-A*3101
OR ~ 11
(in Japanese)

HLA-B*1502
OR ~ 1400
(in Chinese, but not in Caucasians or Japanese)
The challenge of implementation

"Here's my sequence..."
New Yorker, 2000

...the right drug, the first time.
FDA’s role

- FDA began including pharmacogenomic (PGx) effects in labels in 2007
- Now lists >100 medications
  - Cancer (somatic mutations) becoming more common now

<table>
<thead>
<tr>
<th>Gene</th>
<th>Drug Metabolism Pathways</th>
<th>Drug</th>
</tr>
</thead>
<tbody>
<tr>
<td>TPMT</td>
<td></td>
<td>azathioprine</td>
</tr>
<tr>
<td>UGT1A1</td>
<td></td>
<td>irinotecan, nilotinib</td>
</tr>
<tr>
<td>CYP2D6</td>
<td></td>
<td>atomoxetine, fluoxetine</td>
</tr>
<tr>
<td>CYP2C19</td>
<td></td>
<td>clopidogrel, proton pump inhibitors</td>
</tr>
<tr>
<td>CYP2C9</td>
<td></td>
<td>celecoxib, warfarin</td>
</tr>
<tr>
<td>N-acetyl transferase</td>
<td></td>
<td>rifampin, isoniazid, pyrazinamide</td>
</tr>
<tr>
<td>DPD</td>
<td></td>
<td>capecitabine</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Gene</th>
<th>Other Germline Variants</th>
<th>Drug</th>
</tr>
</thead>
<tbody>
<tr>
<td>VKORC1</td>
<td></td>
<td>warfarin</td>
</tr>
<tr>
<td>HLA-B*1501</td>
<td></td>
<td>carbamazepine</td>
</tr>
<tr>
<td>HLA-B*5701</td>
<td></td>
<td>abacavir</td>
</tr>
<tr>
<td>CCR5</td>
<td></td>
<td>maraviroc</td>
</tr>
<tr>
<td>Familial hypercholesterolemia</td>
<td></td>
<td>atorvastatin</td>
</tr>
<tr>
<td>G6PD deficiency</td>
<td></td>
<td>rasburicase, primaquine</td>
</tr>
<tr>
<td>Protein C deficiency</td>
<td></td>
<td>warfarin</td>
</tr>
<tr>
<td>urea cycle disorder</td>
<td></td>
<td>valproate</td>
</tr>
</tbody>
</table>

http://www.fda.gov/Drugs/ScienceResearch/ResearchAreas/Pharmacogenetics/ucm083378.htm
A Case for Prospective Genotyping: identifying another high risk group

52,942 Vanderbilt “Medical Home” patients followed for up to 5 years....

**How many patients received drug(s) that have a recognized pharmacogenetic story?**

- 65% received ≥1 med within 5 years

Estimated number of severe adverse events mitigated: 383 (~12-18 events for the average PCP over 5 years)

Schildcrout et al, CPT 2012
A first step to pharmacogenomic implementation

The Vanderbilt PREDICT project: Pharmacogenomic Resource for Enhanced Decisions In Care and Treatment

1. Select populations of patients who are at “high risk” for receiving a drug with an actionable “pharmacogenetic” story.

2. Genotype subjects on a platform that assays genotypes important for variable actions of many drugs preemptively.

3. Store the genotypes. Embed “actionable” drug-gene pairs in the EMR coupled to informatics tools to provide point-of-care advice. Track outcomes.

“Here's my sequence...”

*New Yorker, 2000*
Vanderbilt Population
410,000

Target Clinics
90,000

Prognostic Flag for Testing
24,000

Prognostic Testing
~6,000

Reactive/Indication Testing
~8,000

Genotyped for PREDICT
~14,000

CLOPIDOGREL 22%
Clopidogrel Advisor

SIMVASTATIN 25%
Simvastatin Advisor

WARFARIN 100%
Warfarin Advisor

THIOPURINES 3%
Thiopurine Advisor
PREDICT: Multiplexing genotyping

CYP2C19
clopidogrel
poor metabolizer

CYP2C9
warfarin
dose/bleeds

CYP2C9
warfarin
dose/bleeds

VKORC1
warfarin
dose/bleeds

CYP2D6
tamoxifen,
antidepressants,
codeine
poor metabolizer

SLCO1B1
simvastatin
myopathy

PREDICT platform tests 184 variants in 34 drug-related genes
Patient comes in, selected for genotyping (cardiac cath, predictive algorithm, etc)

Genotype DB

184 variants

Drop variants that don’t work well

Select variants put into EMR
- Validated
- Clinical Decision Support
- Pharmacy & Therapeutics review

~130 other variants validated of unknown significance

New research for drug-genome interaction discovery
PREDICT research team
**CYP2C19 - Clopidogrel**

*17 = CYP219 Gain of Function Variant

---

**CYP19 Genotype**  
**Drug Metabolism Phenotype**

- ***/X/*X**  
  → Poor metabolizer

- ***/1/*X**  
  → Intermediate metabolizer

- ***/X/*17**  
  → Indeterminate

- ***/1/*17**  
  → Normal (Extensive) metabolizer

- ***/1/*1**  
  → Rapid metabolizer

- ***/17/*17**  
  → Uncharacterized genotype

- **Other**  
  → Uncharacterized genotype
Clinical Decision Support within E-Prescribing

### Drug-Genome Advisor

**Intermediate Metabolizer - clopidogrel (Plavix) - Rare Risk Allele**

Substitution recommended due to increased cardiovascular risks

<table>
<thead>
<tr>
<th>If not otherwise contraindicated:</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Prescribe prasugrel (Effient) 10 mg daily</td>
</tr>
<tr>
<td></td>
<td><strong>Prasugrel should not be given to patients:</strong></td>
</tr>
<tr>
<td></td>
<td>• history of stroke or transient ischemic attack</td>
</tr>
<tr>
<td></td>
<td>• &gt;= 75 years of age [Current patient age: 51]</td>
</tr>
<tr>
<td></td>
<td>• with body weight &lt; 60 kg [Current patient weight: 59.0 kg as of 10/12/2012]</td>
</tr>
<tr>
<td></td>
<td>Prescribe ticagrelor (Brilinta) 90 mg twice daily</td>
</tr>
<tr>
<td></td>
<td><strong>Ticagrelor should not be given to patients:</strong></td>
</tr>
<tr>
<td></td>
<td>• history of severe hepatic impairment</td>
</tr>
<tr>
<td></td>
<td>• intracranial bleed</td>
</tr>
<tr>
<td></td>
<td>Continue with clopidogrel (Plavix) prescription</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Primary override reason:</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Contraindicated for prasugrel or ticagrelor</td>
</tr>
<tr>
<td></td>
<td>Potential side effects</td>
</tr>
<tr>
<td></td>
<td>Provider/Patient opts for clopidogrel</td>
</tr>
<tr>
<td></td>
<td>Cost</td>
</tr>
</tbody>
</table>

This patient has been tested for CYP2C19 variants which has identified the presence of one copy of a rare risk allele which is associated with intermediate metabolism of clopidogrel. Intermediate metabolizers treated with clopidogrel at normal doses are associated with higher rates of stent thrombosis and other cardiovascular events. The Vanderbilt P&T Committee recommends that prasugrel or ticagrelor replace clopidogrel for poor metabolizers unless contraindicated. If not feasible, maintain standard dose of clopidogrel. The guidelines above were developed based on the outcome studies of patients who received a drug-eluting stent into a coronary artery. However, there is not a national consensus on drug/dose guidance particularly associated with the population possessing extremely rare genetic variants.
Decision Support for Warfarin Initial Dose

Warfarin Recommended Initial Dosing
This patient has been tested for CYP2C9 and VKORC1 genetic variants that can affect a patient's warfarin dosing requirements. The following dosing algorithm uses genetic and other patient information to estimate a weekly warfarin dose. This dosing recommendation ONLY applies to NEW starts of warfarin. If the patient has previously taken a stable dose of warfarin, please disregard this dosing recommendation.

- Age: 25
- Weight (kg): 86.2
- Height (cm): 188.0
- Genetic Variants: vkorc1 a/g; cyp2c9 *3/*3
- Is the patient currently taking amiodarone? No
- Is the patient currently taking an inducer (phenytoin, rifampin, carbamazepine)? Yes

Recommended WEEKLY starting dose of warfarin: 20.9 mg/week
The DAILY equivalent of this recommended starting dose is 3.0 mg/day.
Help me decide the tablet size and number of tablets per day

The advisor appears in the black box and shows the Recommended initial WEEKLY & DAILY dose

Evidence Link/View Algorithm

Links to clinical evidence and dosing table.
<table>
<thead>
<tr>
<th>Medication</th>
<th>Does your genetic test result affect your response to medicines?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clopidogrel/Plavix®</td>
<td>No</td>
</tr>
<tr>
<td>Simvastatin/Zocor®</td>
<td>No</td>
</tr>
<tr>
<td>Tacrolimus/Prograf®</td>
<td>No</td>
</tr>
<tr>
<td>Thiopurine Therapy</td>
<td>Yes</td>
</tr>
<tr>
<td>Warfarin/Coumadin®</td>
<td>Yes</td>
</tr>
</tbody>
</table>

**The Clopidogrel Test**

Clopidogrel (sounds like "kloh-PID-oh-grel") is a blood thinner used to prevent clots that can cause a heart attack or stroke. Your genes can affect how well the drug works. This genetic test identifies how well you may respond to clopidogrel.

**Your Risk**

Sometimes clopidogrel does not prevent harmful strokes or clots as well as it should because of your genes. Your provider, often with the results of a lab test, can determine if clopidogrel is the right medicine for you.

*Based on the results of your test, your genes do not put you at increased risk for this negative outcome*

**More About Clopidogrel**

**More About Your Risk**
Testing for clopidogrel efficacy

clopidogrel $\xrightarrow{\text{CYP2C19}}$ 2-oxoclopidogrel

- 78% no common variant
- 18.9% heterozygous
- 2.7% homozygous

Risk of MI:
- 3% (inactive)
- 19%
Multiplexed testing for pharmacogenetic variants
(after 5 drug-gene pairs, n=13,451)

- 0 variants (11.7%)
- 1 variant (29.5%)
- 2 variants (31.7%)
- 3 variants (18.4%)
- 4 variants (6.8%)
- ≥5 variants (1.9%)

88% with ≥1 risk variant

...but 99.8% of African Americans

Van Driest et al, Clin Pharmacol Therap. 2013
Multiplexed Genetics Testing can save money too

Van Driest et al, Clin Pharmacol Therap. 2013
Warfarin CDS Surveillance Example

Recommended Daily Dose = 9 mg/d
Initial Dose Prescribed = 1 mg/d

Gene Results = warfarin normal responder
Recommended Weekly Dose = 63.0
Amiodarone = 0
Inducer = 0
Age = 39
Height = 180
Weight = 78.5
Initial analysis of Rx rates by CYP2C19

- 7405 PREDICT genotyped patients from 10/1/2010 to 6/30/2012:
  - 1620 with stent placed
  - “final” antiplatelet therapy identified at 90 days

![Graph showing proportion prescribed drug within genotype group](image-url)
# PREDICT: Cost to Patient

**Clopidogrel vs. Alternatives**

<table>
<thead>
<tr>
<th>Antiplatelet Drug</th>
<th>Dose</th>
<th>Avg. Annual Retail Price</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clopidogrel</td>
<td>75 mg qd</td>
<td>$480.53</td>
</tr>
<tr>
<td>Prasugrel</td>
<td>10 mg qd</td>
<td>$3365.52</td>
</tr>
<tr>
<td>Ticagrelor</td>
<td>90 mg qd</td>
<td>$1,736.79</td>
</tr>
</tbody>
</table>

**Maximum out-of-pocket cost of PREDICT test:** $420
PREDICT helps match patient with proper drug

BY: KATHY WHITNEY

10/28/2010 - Had Scyble Van Cleve, a spry 83-year-old from Brentwood, had her heart procedure done a month ago instead of one week ago, she would have been prescribed the standard dose of clopidogrel, a blood thinner used to prevent blood clots from forming around her coronary stents.

Scyble Van Cleve, right, is the first patient at Vanderbilt to benefit from a new program that puts genetic information in the patient's medical records to help physicians like John McPherson, M.D., choose the drug and dose that will benefit them the most. (photo by Susan Urmy)
Our case: What personalizing medicine really means

57yo with admitted for angina, receives stent

Recath, stent
“Plavix x 1 year minimum. ASA life long.”

Cath, more stents
In-stent thrombosis, restent
In-stent thrombosis, restent

9th admission, 5th intervention, 9th stent
PREDICT: CYP2C19*2/*2

January
April
December

clopidogrel started

Switched to prasugrel
Cost per Genome

Moore’s Law

National Human Genome Research Institute

genome.gov/sequencingcosts

eMERGE-PGx – Overall Goal

A multi-site test of targeted sequencing of 84 genes, validation, and EMR decision support to guide care in ~9,000 eMERGE patients
Preliminary PGRN-Seq Results

*SCN5A* and *KCNH2* in 2,200 Patients

- 83 rare (MAF < 1%) in *SCN5A*, 45 in *KCNH2*
- 121/128 MAF < 0.5%, 92 singletons
- Three labs assessed known/likely pathogenicity

![Venn Diagram]

<table>
<thead>
<tr>
<th>Lab</th>
<th>Count</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lab 1</td>
<td>16/121</td>
</tr>
<tr>
<td>Lab 2</td>
<td>24/121</td>
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<td>Lab 3</td>
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Personalized medicine – not a new idea

The good physician treats the disease; the great physician treats the patient who has the disease.

Sir William Osler
Summary

- EMRs and EMR-linked DNA biobanks play an important role in both discovery and implementation of genomic medicine. EMRs biobanks can accelerate discovery and save money.

- Prospective testing has an opportunity to change prescribing patterns, but requires decision support to help the physicians navigate the complex prescribing process.

- Multiplexed testing of genetic variants prospectively reduces total numbers of tests and may reduce cost.
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- Mayo Clinic
- Group Health/UW
- Mount Sinai
- Geisinger

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