IGNITE & Beyond Workshop

• Identify and prioritize scientific opportunities to fill evidence gaps in genomic medicine implementation

• Make recommendations for future research in genomic medicine implementation

• Evaluate the key contributions of IGNITE
IGNITE Goals

• Develop new collaborations in diverse settings and populations
• Expand and link existing genomic medicine efforts
• Contribute to evidence base of outcomes to support using genomic information in clinical settings
• Incorporate genomic information into EHR systems; new CDS strategies
IGNITE Goals (cont.)

• Develop novel patient and provider educational models to promote genomic literacy

• Engage patients, clinicians, researchers, and payers to demonstrate the value of using genomic information through genotyping, sequencing, and family history

• Define, share, and disseminate best practices of implementation, diffusion, and sustainability
IGNITE Network

• 6 demonstration projects, hub and spoke design
• Focus on real-world health care delivery
• Coordinating Center
  – Logistics planning
  – Support and coordinate network activities
  – Cultivate communication and collaboration with external groups implementing genomic medicine
  – Increase visibility of IGNITE
<table>
<thead>
<tr>
<th>IGNITE Project Sites</th>
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<tbody>
<tr>
<td><strong>Duke University</strong></td>
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<tr>
<td>Geoff Ginsburg, Lori Orlando</td>
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<tr>
<td>• Family health history risk data collection via web-based platform</td>
</tr>
<tr>
<td>• Variety of practice environments and populations</td>
</tr>
<tr>
<td>• Generate high quality risk information for patients</td>
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<p>| <strong>Indiana University</strong> |
| Todd Skaar, Paul Dexter |
| • Pharmacogenomic panel for 24 drugs |
| • 26% of participants have a clinically actionable PGx variants |
| • Rapid implementation across state system |</p>
<table>
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<th>IGNITE Project Sites</th>
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<tr>
<td>Mount Sinai</td>
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<tr>
<td>Carol Horowitz</td>
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<tr>
<td>University of Florida</td>
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<tr>
<td>Julie Johnson</td>
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- **Mount Sinai**
  - APOL1 testing for adults with hypertension and self-reported African ancestry
  - High levels of interest in genomics research
  - Limited concern regarding testing in patients

- **Carol Horowitz**
  - CYP2C19 PGx testing for clopidogrel post-PCI
  - Improved patient outcome via guided approach
  - Creation of professional education and training programs
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<tr>
<td><strong>University of Maryland</strong></td>
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<tr>
<td>Toni Pollin</td>
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<tr>
<td>• Genotyping for 40 genes associated with highly penetrant form of diabetes</td>
</tr>
<tr>
<td>• Improved accuracy of diagnosis, reduces unnecessary testing</td>
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<tr>
<td>• Improves overall patient care, limits diagnostic odyssey</td>
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<tr>
<td><strong>Vanderbilt University</strong></td>
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<tr>
<td>Josh Denny, Mia Levy</td>
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<tr>
<td>• Routine tumor gene mutation testing</td>
</tr>
<tr>
<td>• Prospective germline PGx testing for antiplatelet therapy</td>
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<tr>
<td>• Reduce adverse drug side effects</td>
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## Diversity in ancestry

<table>
<thead>
<tr>
<th>Race/ethnicity</th>
<th>N</th>
<th>%</th>
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</thead>
<tbody>
<tr>
<td>White, non-Hispanic</td>
<td>54,698</td>
<td>75</td>
</tr>
<tr>
<td>Black or African American</td>
<td>13,004</td>
<td>18</td>
</tr>
<tr>
<td>Hispanic/Latino</td>
<td>3,268</td>
<td>4</td>
</tr>
<tr>
<td>Asian</td>
<td>1,149</td>
<td>2</td>
</tr>
<tr>
<td>American Indian/Alaskan Native</td>
<td>15</td>
<td>0.02</td>
</tr>
<tr>
<td>Native Hawaiian/Pacific Islander</td>
<td>13</td>
<td>0.02</td>
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IGNITE contributions to genomic medicine

• Informatics:
  – Variant to phenotype translation; using CDS to provide reliable information and interpretations
  – Developing CDS rules for different EMR systems; CDS-KB

• Reimbursement: Inform coverage decisions
  – Pharmacogenomics:
    • CYP2C19 testing and clopidogrel

• Education:
  – Developed guidelines for clinicians to communicate information about lower-risk testing to patients (e.g. PGx, family history)
IGNITE SPARK Toolbox

Get Started

Implementing Genomics in Practice?
Search **Clinical** Tools

Researching Genomics in Practice?
Search **Research** Tools

Search **All** SPARK Tools

or

Explore the SPARK Toolbox

https://ignite-genomics.org/
IGNITE & Beyond Workshop Objectives

• Evaluate the key contributions of IGNITE

• Identify and prioritize scientific opportunities to fill evidence gaps in genomic medicine implementation

• Make recommendations for future research in genomic medicine implementation

*How do we make genomic medicine part of routine clinical care?*

*Outstanding barriers and challenges to uptake?*
Workshop Format

Four sessions
• Implementation
• Clinical informatics
• Clinical evidence
• Economic considerations

Focus topics
• State of the science and gaps
• IGNITE highlights and opportunities
• Recommendations

Attendees
• IGNITE Network investigators
• IGNITE External Scientific Panel members
• IGNITE Affiliate members
• Scientists engaged in genomic medicine implementation research

Total 86 attendees including 6 Council members
Key Recommendations: Implementation

• Foster robust **collaborations**: academic + community centers

• Prioritize inclusion of **underrepresented populations** and diverse researchers and clinicians to reduce disparities
  – Focus on linguistic diversity, rural populations, smaller clinics, economic diversity

• Create **genomic medicine resource center**; educational materials
Key Recommendations: Clinical informatics

- Promote **harmonization** and consolidation of information standards across various EHR/CDS network
- Focus on **CDS interfaces** at different levels (patient, provider, etc.)
- Ensure that **new advances** in genomic medicine or CDS are made available to the community
- Collate CDS rules into a **public repository** using existing resources
Key Recommendations: Clinical evidence

• Conduct **larger, network-wide studies**
  – Address important clinical issues; **specific outcomes**
  – Generate **evidence**

• Involve representatives from **health insurance companies**

• Utilize **cost effectiveness** information of different treatments

• Communicate **utility of genomic medicine** to different types of clinicians (nurses, residents, etc.)

• Foster systematic collection, creation, evaluation and evidence synthesis of genomic studies made **publicly available**
Key Recommendations: Economics

• Develop measures of *societal and personal utility* for genomic testing
  – Need to validate these measures

• Communicate with *stakeholders* during study development

• Establish *economic data source standards*

• Explore *economic downstream value* of genomic medicine
Prioritization and Implementation of Recommendations
Acknowledgements

IGNITE NHGRI Program Staff
- Colette Fletcher-Hoppe
- Heather Junkins
- Nicole Lockhart
- Ebony Madden
- Teri Manolio
- Jeff Struewing
- Simona Volpi

IGNITE and Beyond Planning Committee
- Lon Cardon
- Chris Chute
- Katrina Goddard
- Howard McLeod
- Casey Overby

*IGNITE and Beyond* workshop speakers, discussants, moderators, and other participants