

## Translational Genomics and Health Outcomes: Some Ongoing and Future Research

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\*Note: Some information included contains  
unpublished data.\*



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### Translational Genomics and Health Outcomes

*Component studies in translational genomics will allow the development of key categories in the integration of genomics into clinical medicine.*

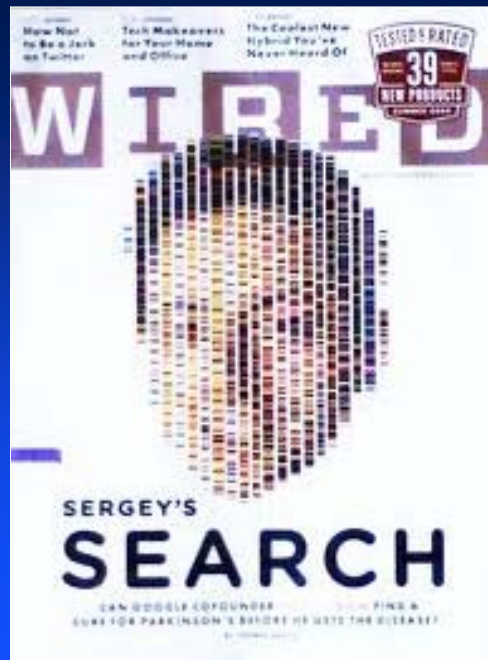


## The ACCE Model for Genetic Testing Do “Benefits” = Clinical Utility?



- Alytic Validity
- Clinical Validity
- Clinical Utility
- ELSI

What about ‘personal utility’?



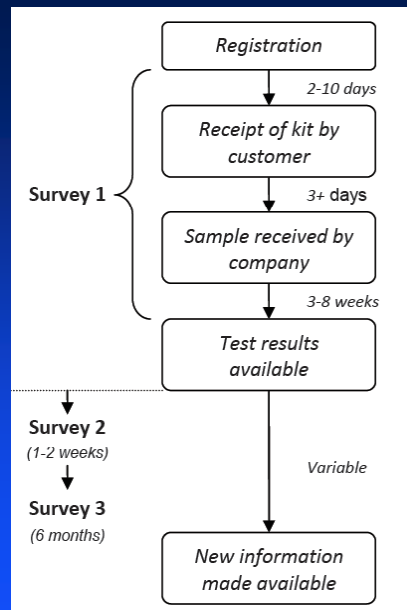
## DTC Customer (IPeG) Study

Natural experiments in the impact of genetic risk disclosure!

### Impact of Personal Genomics Testing Study

(Green-Roberts, PIs)

NHGRI funding has begun  
October, 2010



## Impact of Personal Genomics Services Study

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## Communicating Genetic Risk for Obesity

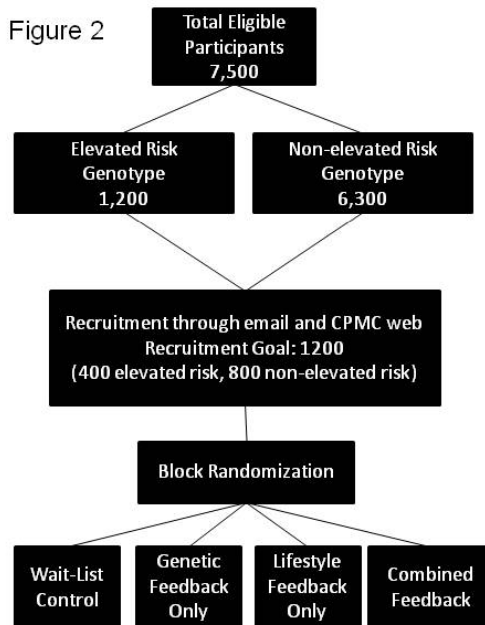
(Wang, PI)

R21 HG00603

### Assessments:

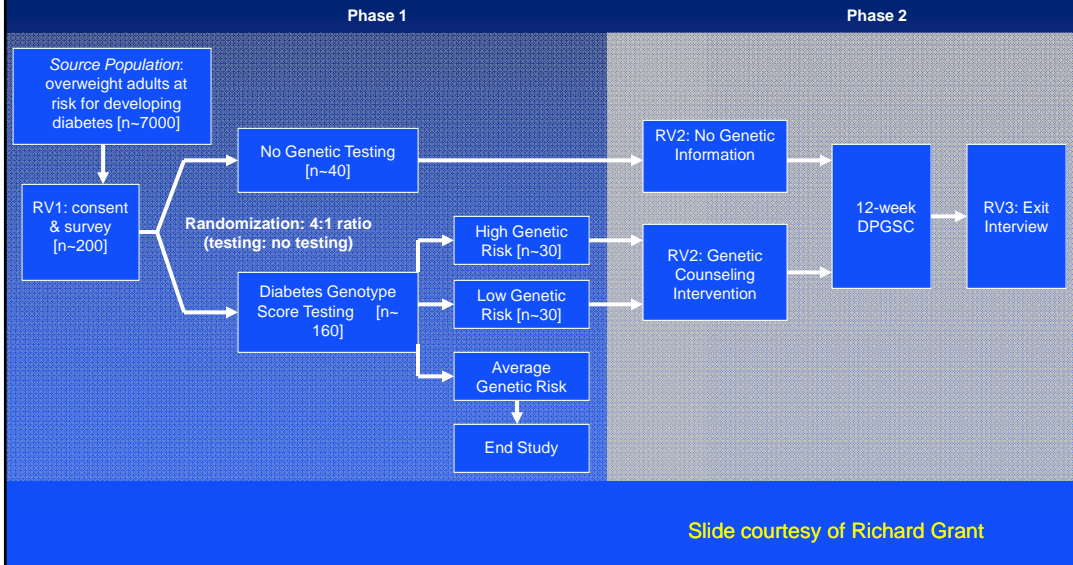
- Illness Representation
- Causal Attributions
  - Perceived Risk
    - Worry
  - Perceived Control
- Genetic Determinism
  - Behavior
- Change in weight/BMI


Figure 2



Slide courtesy of Catharine Wang

# Communicating Genetic Risk for Diabetes (Grant, PI) R21 DK084527






## Genomics in Medicine

*“...even though evidence is insufficient, the clinician must still provide advice, patients must make choices, and policymakers must establish policies”*

*US Preventive Services Task Force, 2009*



## WGS in the Practice of Medicine

General Genomic Medicine	Disease-Specific Genomic Medicine
<p style="text-align: center;"><b>General Genome Report</b> <i>All Patients</i></p> <ul style="list-style-type: none"> <li>Penetrant disease variants</li> <li>Recessive carrier status</li> <li>PGx Variants</li> <li>Blood and HLA Type</li> </ul>	<div style="background-color: #d9e1f2; padding: 10px; margin-bottom: 10px;"> <p style="text-align: center;"><b>Symptoms/Disease/FamHx Specific Genome Report</b> <i>Selected Patients</i></p> <p style="text-align: center;">Annotation of variants in relevant disease genes</p> </div> <p style="text-align: center; font-size: 2em; color: white;">+</p> <div style="background-color: #ffffcc; padding: 10px;"> <p style="text-align: center;"><b>General Genome Report</b> <i>All Patients</i></p> <ul style="list-style-type: none"> <li>Penetrant disease variants</li> <li>Recessive carrier status</li> <li>PGx Variants</li> <li>Blood and HLA Type</li> </ul> </div>

## Study Arms

	Standard of Care	Standard of Care plus Whole Genome Sequencing
Primary Care	Healthy patients managed according to family history and presenting symptoms	Healthy patients managed according to family history, presenting symptoms and WGS
Cardiology	Patients with HCM managed according to family history, presenting symptoms and conventional genetic testing	Patients with HCM managed according to family history, presenting symptoms and WGS

## Support for Genomics in Medicine

