GENERATIONS PROJECT
and DNA-based Health Screening

CT Forum on DNA Testing and Genomic Medicine
Thursday, January 23, 2020
Legislative Office Building (LOB) Room 2C
300 Capitol Ave, Hartford, CT 06106

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New Haven, CT
Genomic Screening Has the Power To Make Important Disease Risks Visible
1 in 75 have risk
4 out of 5
and risk
A project that will enroll a cohort of \( \geq 100,000 \) volunteers and be an engine for 21st century preventive health care
Generations

• Led by Yale Center for Genomic Health.
• Formal launched September 2019.
• Plan is to enroll > 100K volunteers who agree to have their genome sequenced (Exome & Genotype Array)
• Link their electronic health record (EHR) to their DNA sequence data in a EHR-DNA dataset that enables both clinical care and research.
• Collaboration between: Yale New Haven Health System & Yale School of Medicine
What kind of Health Screens have you undergone?

- Blood Pressure
- Cholesterol
- Mammogram
- Colonoscopy

Health Screens make risk visible

When is a screening method ready for the Population?
In December (1710) I caused a mare to be tied down alive on her back; she was 14 hands high, and about 14 years of age, had a fistula on her withers, was neither very lean nor very lusty: having laid open the left crural artery about 3 inches from her belly, I inserted into it a brass pipe whose bore was 1/6 of an inch in diameter; and to that, by means of another brass pipe which was fitly adapted to it, I fixed a glass tube, of nearly the same diameter, which was 9 feet in length; then untying the ligature on the artery, the blood rose in the tube 8 feet 3 inches perpendicular above the level of the left ventricle of the heart: but it did not attain to its full height at once; . . . when it was at its full height, it would rise and fall at and after each pulse 2, 3, or 4 inches. . . .” (Reverend Stephen Hales, 1738; 1).
Public Health Campaign for Blood Pressure Screening

Did it work?

4 out of 5 adults with high blood pressure know that they have this risk for heart attack and stroke.

[https://www.cdc.gov/bloodpressure/facts.htm]
What kind of Health Screens have you undergone?

- Blood Pressure
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- Mammogram
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Health Screens make risk visible

When is a screening method ready for the Population?

- Is it acceptable?
- Does it focus on important issues?
- Is it better than what we’re currently doing?
- Is it worth the investment?
10 Leading Causes of Death
(CDC statistics)

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<th>LEADING CAUSES OF DEATH</th>
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<td>2. Cancer</td>
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<td>3. Accidents</td>
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<td>4. Chronic Lower Respiratory Disease</td>
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<td>5. Stroke</td>
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<td>6. Alzheimer’s Disease</td>
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2013 - The Angelina Effect

BRCA testing recommendations based on personal history or strong family history of Breast and/or Ovarian Cancer.

Angelina Jolie’s Family History

Mother  Ovarian CA  
Aunt   Breast CA  
Aunt   Breast CA  
Cousin  Breast CA  
G-mother Ovarian CA
How are we doing with identifying individuals with BRCA 1/2 Associated Risk for Cancer?

4 out of 5 people with this cancer risk didn’t know it but for DNA-based screening
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1 in 75

• Screening 10 Genes can identify people with significant elevated risk for
  ▪ Breast Cancer
  ▪ Ovarian Cancer
  ▪ Colon Cancer
  ▪ Heart Attack
  ▪ Stroke

• In CT - estimated 45 thousand people
• In USA - estimated 4.6 million people
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Thank you!