Genetic Knowledge and Return of Results Preferences in the Mayo Clinic Biobank

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The Mayo Clinic Biobank

- A Mayo Clinic initiative to enroll 50,000 Mayo Clinic patients regardless of their health history (no specific disease) by end of 2016
- Launched 4/1/2009
- Eligible:
  - Mayo patients
  - 18 years+
  - US resident
Mayo Clinic Biobank

- Invited via mail prior to medical appointment
- Over 40,000 consented to date
  - Blood
  - QN data
- Over 110 projects approved to date
Excerpt from Biobank Consent form:

“9. What if researchers discover something about my health?

► During individual studies, researchers could find out important information about your health. They might discover something about your health right now, or about your risk of getting sick in the future. Researchers will not discover something about every donor, so you are not guaranteed to receive results.
Excerpt from Biobank Consent form:

- Since decisions about health and disease are very personal, no one can predict which results donors will want in the future. One of the important jobs that BTOG has is to decide which research results, if any, will be returned to Biobank donors. They will make this decision for each individual study after consulting with the appropriate researchers, doctors, and the Community Advisory Board. Names will not be mentioned during this process."

Outcome of Biobank Access Committee on February 16, 2011:
- Genetic test results would be returned if warranted
Genetic Knowledge Survey
Questions

► What is the level of genetic knowledge in our population?

► As we think about returning results in the future what are the interests of our population in receiving results?

► What do they think about the use of new Whole Genome Sequencing technology?
Survey Study Design

- Stratified random sample of 1200 Biobank participants stratified by
  - Age Group (18-30, 30’s, 40’s, 50’s, 60’s, 70+)
  - Education (∆ High School, > High School)
  - Sex

- Mailed packet (letter, QN) to home address
  - Two mailings, 1 month apart
## Responders vs. Non-Responders

<table>
<thead>
<tr>
<th>Category</th>
<th>Responders N=685 (57%)</th>
<th>Non-Responders N=515</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age (mean, years)</td>
<td>55</td>
<td>46</td>
</tr>
<tr>
<td>Sex, Female (%)</td>
<td>52</td>
<td>47</td>
</tr>
<tr>
<td>Education (% &gt; High school)</td>
<td>56</td>
<td>45</td>
</tr>
<tr>
<td>Race (% white)</td>
<td>96</td>
<td>88</td>
</tr>
</tbody>
</table>
Table 1 – Description of Participants

<table>
<thead>
<tr>
<th>Category</th>
<th>Completers</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age Categories</strong></td>
<td></td>
</tr>
<tr>
<td>18-30</td>
<td>68 (10%)</td>
</tr>
<tr>
<td>31-40</td>
<td>94 (14%)</td>
</tr>
<tr>
<td>41-50</td>
<td>101 (15%)</td>
</tr>
<tr>
<td>51-60</td>
<td>136 (20%)</td>
</tr>
<tr>
<td>61-70</td>
<td>149 (22%)</td>
</tr>
<tr>
<td>70+</td>
<td>137 (20%)</td>
</tr>
<tr>
<td><strong>Education</strong></td>
<td></td>
</tr>
<tr>
<td>HS or Less</td>
<td>304 (44%)</td>
</tr>
<tr>
<td>Some College</td>
<td>146 (21%)</td>
</tr>
<tr>
<td>College degree +</td>
<td>235 (34%)</td>
</tr>
</tbody>
</table>
Sections of the Survey

- General Knowledge of Genetics
- Interest in Return of Research Results
- Case Scenarios – Return of Research Results
  - Cystic Fibrosis
  - HBOC
- Whole Genome Sequencing
Overall Genetic Knowledge

If I were told I had an increased genetic risk for a disease, it would mean that I have a greater possibility of getting that disease because I have one or more genetic variants that are linked to that disease.

When people talk about “Individualized” medicine or “personalized” medicine they are usually referring to using information from a person’s genome (their DNA) to help guide a person’s health care.
Testing my genes can…

- Show if I have a genetic risk for one or more diseases or conditions
  - 87% correctly identified this as true

- Show if my genetic makeup plays a role in a disease or condition that I already have
  - 82% correctly identified this as true

- Give me a clean bill of health
  - 81% correctly identified this as false

- Give me information about me and my relatives
  - 75% correctly identified this as true
Return of Results Preferences
It is important to me to find out if I have other genetic variants that might be important to **my** health.

It is important to me to find out if I have other genetic variants that might be important to my **children’s** health.

Answers to Q1 and Q2 are strongly associated. (Concordance of 92%)
I would be concerned about any of my genetic information going into my medical record.

I would be concerned if any of my genetic information was available to health and life insurance companies.

People who were concerned tended to be in poor health.

Q4 had no association with health status.
I would want to know about my genetic information even if I or my doctor could not do anything to diagnose, treat, or prevent a disease or disorder.

When I die, I would want my family members to have access to my genetic information.
Case Scenarios

- **Recessive - Cystic Fibrosis**
  - Pulmonary disease with early onset
  - Expected younger subjects to be most interested

- **Dominant – Hereditary Breast & Ovarian Cancer (HBOC)**
  - Increased risk for cancers of the breast, ovaries, prostate, pancreas with adult onset
  - Expected most interest among older women
Scenarios: Cystic Fibrosis

% CF-Yes

Age

0 10 20 30 40 50 60 70 80 90 100
18-30 31-40 41-50 51-60 61-70 70+
Scenarios: Cystic Fibrosis – by sex

% vs. Age

- CF-Yes
- Females
- Males
Scenarios: HBOC

%  

HBOC-Yes  Females  Males

Age

18-30  19-40  41-50  51-60  61-70  70+

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Scenarios: HBOC – by age

- HBOC-Yes
- %Yes Females
- % Yes Males

Age

18-30 31-40 41-50 51-60 61-70 70+

%
Preferred Method of Receiving Results

1. In person – Genetic Counselor
   ▶ Ranked #1 by 61% (CF); 64% (HBOC)

2. On the phone – genetic counselor
   ▶ Ranked #1 by 20% (CF); 18% (HBOC)

3. E-visit
   ▶ Ranked #1 by 15% (CF); 12% (HBOC)
Whole Genome Sequencing

- Obtain genetic information about all sequences in their genomic materials
  - Potential for obtaining risk information on hundreds of different diseases

- Potential for large-scale WGS within the Mayo Clinic Biobank
  - Large scale return of results

- Discussed at time of survey development
I approve of the Mayo Clinic Biobank applying this new technology on stored participant DNA samples.
I would want the Mayo Clinic Biobank to re-contact me so I can give my permission for this particular project before they apply this new technology on my stored DNA sample.
If the Mayo Clinic Biobank asked for my permission to allow my stored DNA sample to be used in a whole genome sequencing project, I would approve the request.

- Strongly disagree: 1
- Somewhat disagree: 2
- Somewhat agree: 23
- Strongly agree: 71
- Missing: 4

Percentages:
- Strongly disagree: 1%
- Somewhat disagree: 2%
- Somewhat agree: 23%
- Strongly agree: 71%
- Missing: 4%
Conclusions

- Generally high level of genetic knowledge in our population
- Interest in receiving results is high for
  - Recessive traits, regardless of age
  - Dominant traits
- Approve of WGS technology and its use on samples
  - Some sort of re-contact desired
  - Making plans for re-contact via bi-annual newsletter
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