

DTC Genetic Testing Companies as Research Entities

Elissa Levin, MS, CGC
Director, Navigenics Genetic
Counseling Program



What is “DTC” research?

Types:

- Internal vs external (collaborative) research
- Randomized control trials
- Lab-based investigations
- Consumer-based surveys/questionnaires
- Non-for-profit and corporate
- “Research Revolution”

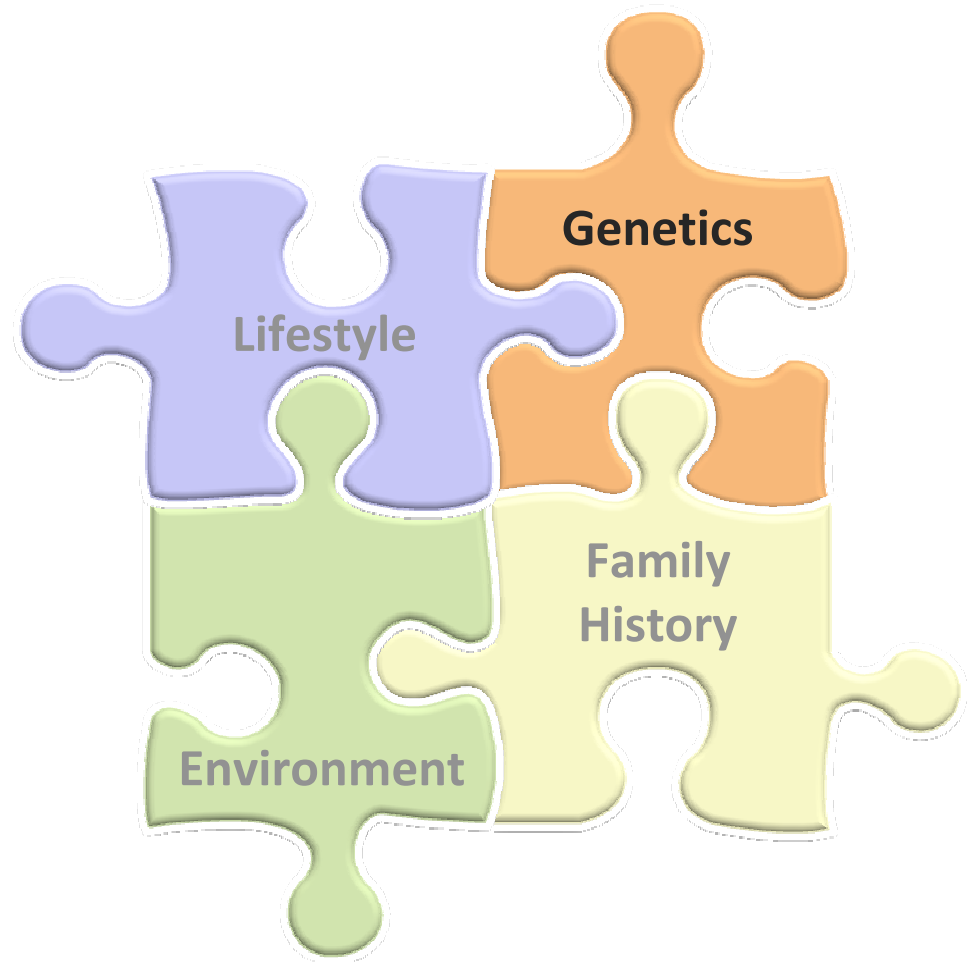
Topics:

- Discovery (new associations)
- Translational/clinical outcomes
- Exploring clinical delivery models and infrastructure
- Psychosocial
- “Citizen scientist”

NAVIGENICS SERVICE

Navigenics Mission

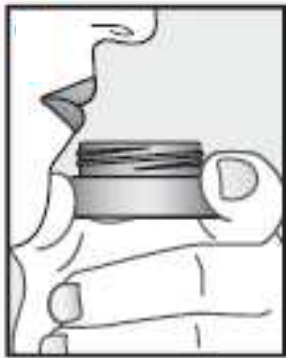
- Improving health outcomes by providing insight into genetic risk factors – enabling prevention and earlier diagnosis of disease.



How it works

1

Sample Acquisition



2

CLIA Laboratory



3

Bioinformatics

```
SNP_A-1917680 rs1783662  
SNP_A-1917683 rs6669915  
SNP_A-1917685 rs2909678  
SNP_A-1917686 rs632683  
SNP_A-1917688 rs7598926  
SNP_A-1917689 rs2086753  
SNP_A-1917691 rs1717276  
SNP_A-1917692 rs9548221  
SNP_A-1917693 rs1044422  
SNP_A-1917694 rs1828288  
SNP_A-1917695 rs1262045  
SNP_A-1917697 rs2019535  
SNP_A-1917698 rs1756405  
SNP_A-1917701 rs1115531  
SNP_A-1917702 rs2371333  
SNP_A-1917705 rs1379947  
SNP_A-1917707 rs2850232  
SNP_A-1917708 rs9933070  
SNP_A-1917709 rs6419643
```

4

Confidential Results



5

Ongoing Updates



Updates

Genetic Counseling Services

Navigenics Health Compass

Cancers

Breast
Colon
Lung
Melanoma
Prostate
Gastric

Autoimmune

Graves' Disease
Psoriasis
Rheumatoid Arthritis
Sarcoidosis

Vascular

Abdominal &
Aortic Aneurysm
Cerebral Aneurysm
Deep Vein Thrombosis

Cardiac

Atrial Fibrillation
Myocardial Infarction

Ocular

Macular Degeneration
Glaucoma

Gastrointestinal

Crohn's disease
Celiac disease
Hemochromatosis
Lactose Intolerance

Endocrine

Diabetes, type 2
Obesity

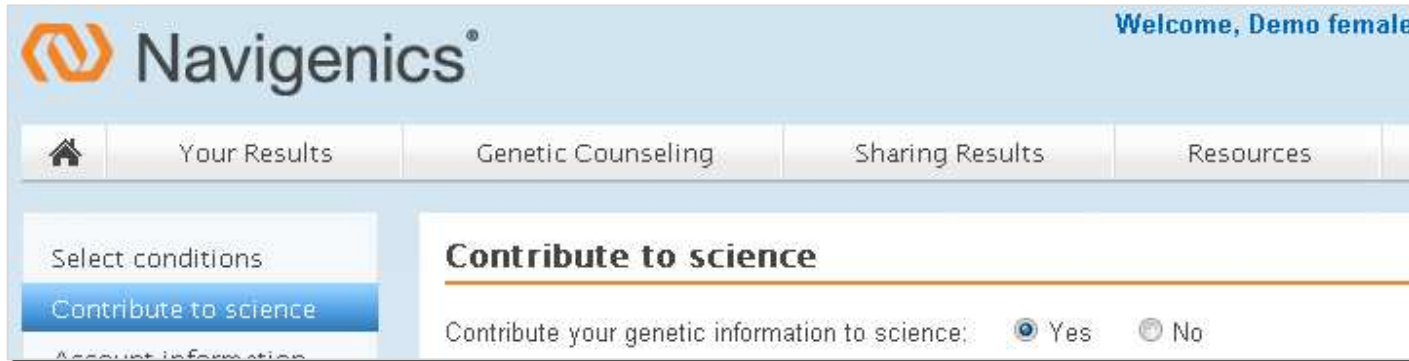
Neurologic

Alzheimer's
Multiple Sclerosis
Restless Leg

Bone

Osteoarthritis

Contribute to science



The screenshot shows the Navigenics website interface. At the top, the Navigenics logo is on the left, and 'Welcome, Demo female' is on the right. Below the logo is a navigation bar with links: Home, Your Results, Genetic Counseling, Sharing Results, and Resources. On the left side, there is a sidebar with links: Select conditions, Contribute to science (highlighted in blue), and Account information. The main content area is titled 'Contribute to science' and contains the text 'Contribute your genetic information to science:' followed by two radio buttons: 'Yes' (selected) and 'No'.

“If you choose to contribute your genetic information to science, you allow:

- Navigenics to share Your Genetic Data and Your Phenotype Information (de-linked from Your Account Information) with organizations that perform genetic or medical research,
- The organizations to separately or jointly publish study results that include Your Genetic Data and Your Phenotype Information (but not Your Account Information), in peer-reviewed scientific and medical journals, and to deposit such data and information into public data repositories or otherwise make them publicly available to the extent required by such journals.”

Contribute to science

- No financial compensation or incentives provided to participants
- “...you may at any time in the future withdraw your consent... effective within 90 days... Your Genetic Data and Your Phenotype Information will not be withdrawn from any ongoing or completed studies or publications in which Your Genetic Data or Your Phenotype Information were utilized because your genetic information will be coded and not able to be identified.”

If you elect not to contribute your genetic information to science:

- “Navigenics will not share Your Genetic Data or Your Phenotype Information with such organizations. Moreover, declining to contribute does not in any way affect the nature of the service Navigenics will provide you.”

COLLABORATIONS

Collaborators



Duke



Randomized control trial



- **Mayo Clinic**
- **PURPOSE:** Multi-arm trial to assess delivery models to understand how best to communicate genetic relative risk to Mayo Clinic patients and to understand how to most effectively integrate this relative risk analysis into the Mayo Clinic's Executive Health Program.
- *Currently in final pilot phase*

OBJECTIVES:

- Understand how to most effectively meet the communications needs of Mayo Clinic patients, physicians and genetic counselors
- Understand the response behavior of Mayo Clinic Executive Health patients regarding their follow-on testing, behavior modification, and counseling
- Learn how to establish a positive and successful working relationship between Mayo Clinic and Navigenics

- **Scripps Genomic Health Initiative**
- Collaborators:
 - Scripps Genomic Medicine Program and Translational Science Institute (PI: Eric Topol, MD)
 - Navigenics
 - Affymetrix
 - Microsoft HealthVault

- Prospectively research the predictive value of genetic risk markers, outcomes and behavior modification efficacy
- Methodology:
 - Enroll up to 10,000 patients/participants
 - Test using the Navigenics Health Compass test
 - Prospectively follow for 20 years
 - Pre-test (baseline)
 - Post-test: 3 months, 1 year, multi-year increments
- *Initial findings will be presented at ASHG (2009)*

MDVIP Pilot Study

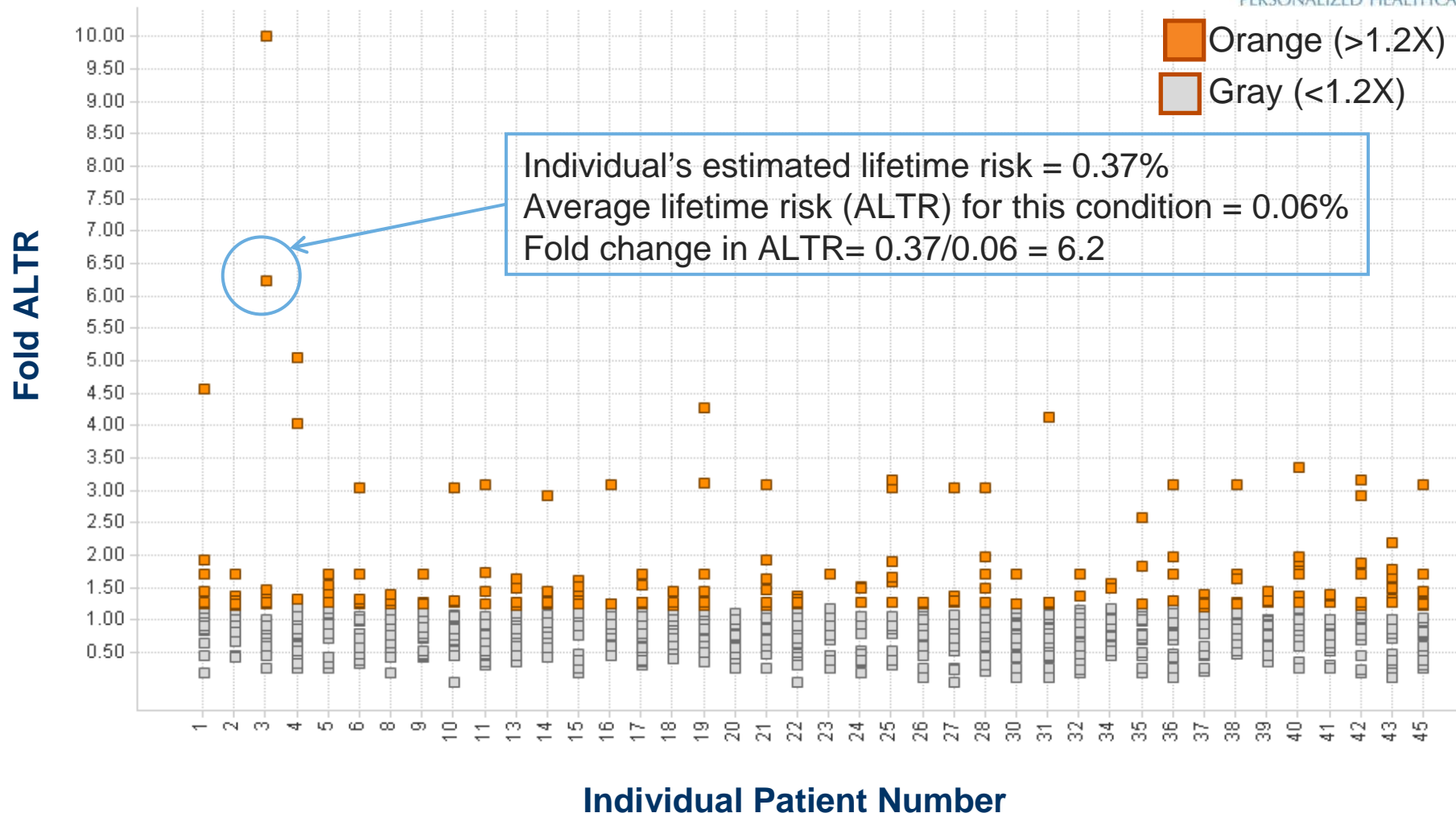


- **MDVIP** is a national network of physicians who practice proactive, preventive and personalized healthcare... These outcomes include lower hospitalization rates.
- Methodology
 - 3 physician investigators
 - Enrolled 40 patients (10-15 per practice)
 - Patient surveyed 1 week and 6 months post-results
 - Data available from 1 week survey

Questions asked

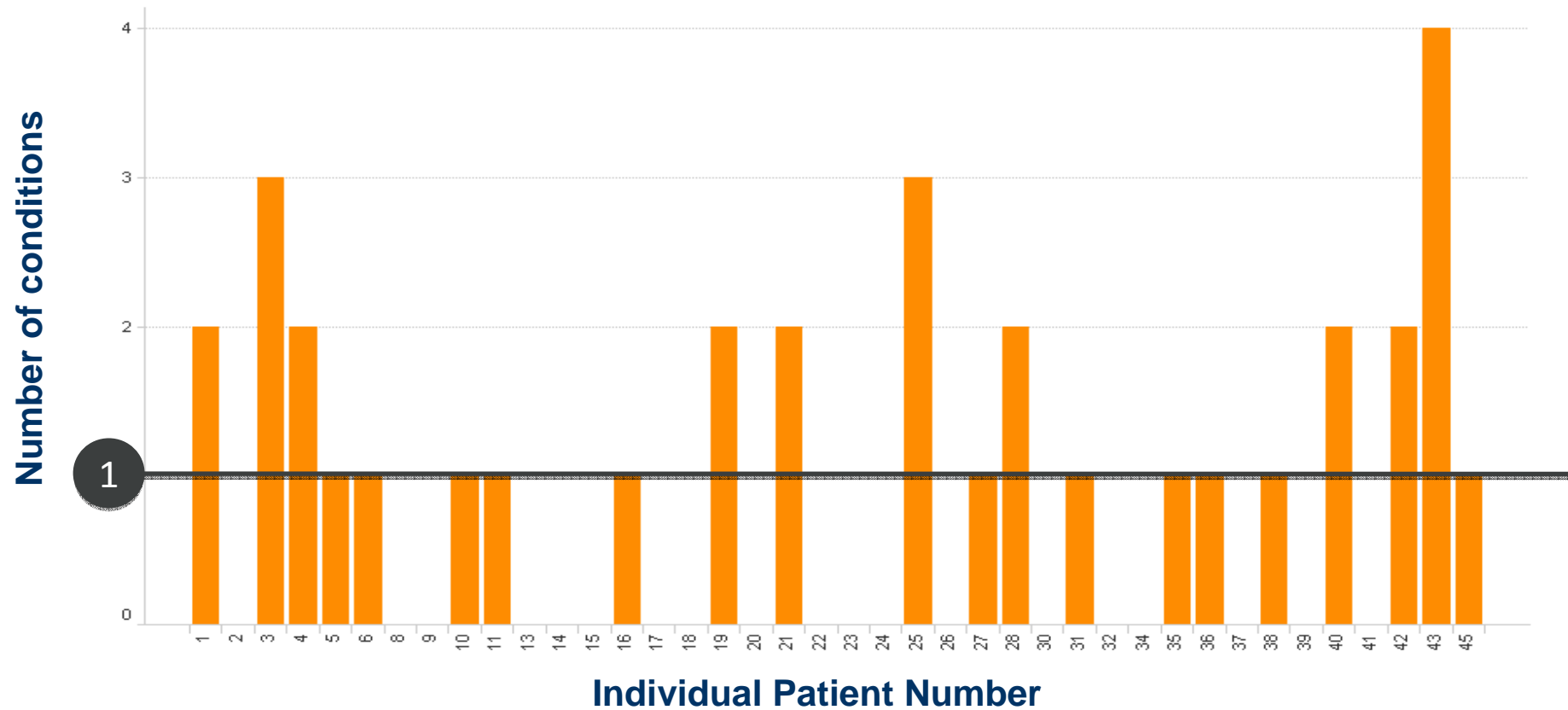
- Is the information relevant?
- Will I be overwhelmed with patient concerns?
- How do I use this information?
- How do patients feel about their results?

Overview of results by patient



- Across the entire population:
 - 98% of patients showed at least one condition with >1.2X increase in average lifetime risk
 - 45% of patients showed at least one condition with >3X increase in average lifetime risk

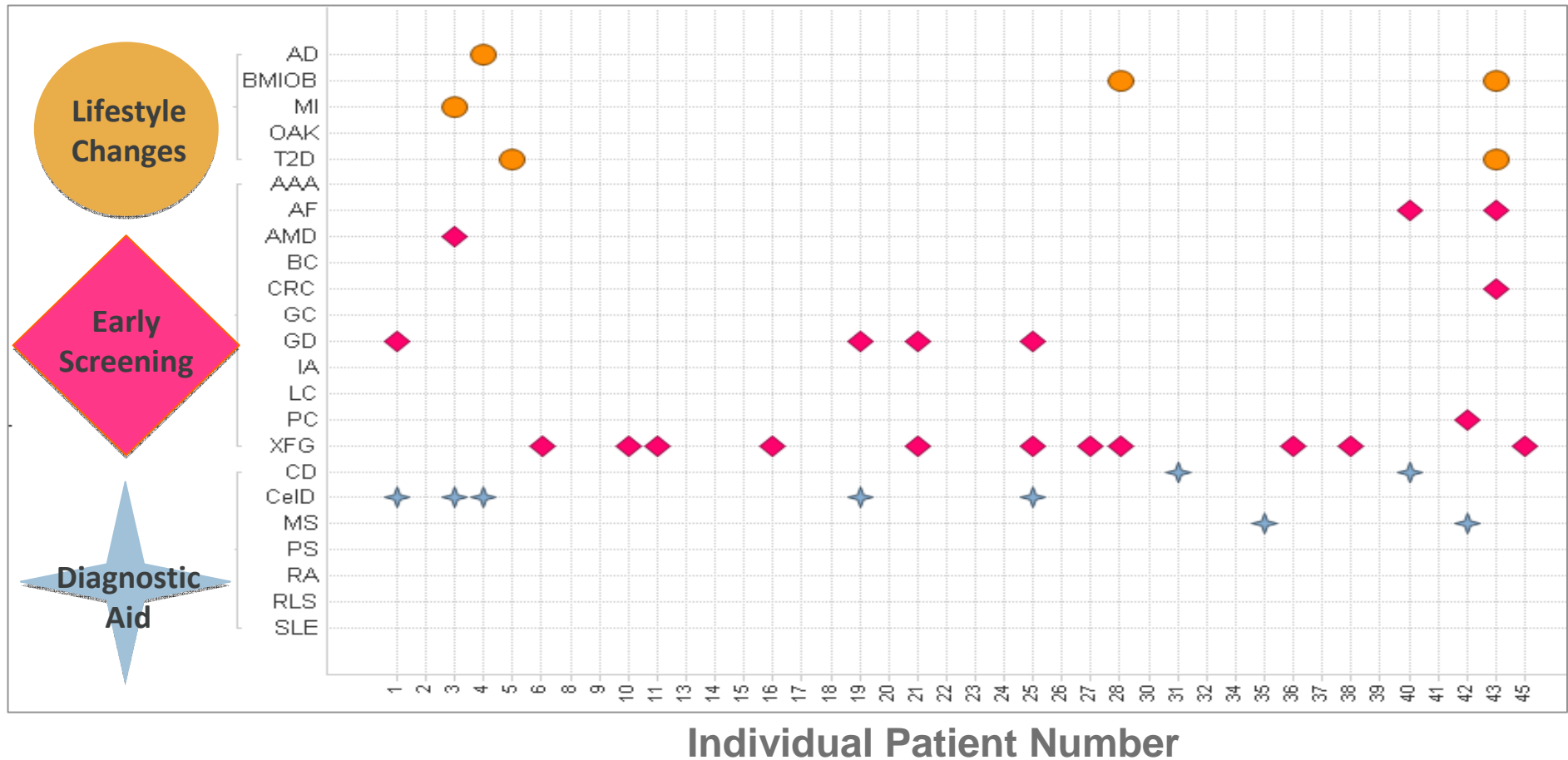
Number of high-risk conditions by patient



- **Only 25%** of patients had more than one condition with either 3x fold increase risk over the average or were in >95% risk percentile

High-risk conditions grouped by intervention

($>3X$ risk or $>95\%$ percentile)



- What percentage of patients where benefit could be derived from:
 - Lifestyle changes 13%
 - Personalized screening 43%
 - Aiding differential diagnosis 23%

Patient satisfaction and motivation



- **97%** said they were “***glad they did this***” (the single outlier wasn’t sure if he was glad or not)
- **83%** said they “***gained new knowledge*** that should be helpful for my long-term health”
- **93%** indicated that they will make some sort of change or ***take an action*** as a result of test
- **77%** say they will “***have more productive dialogue with my physician*** to improve my long-term health”

Internal research

Genetic counseling outreach initiative

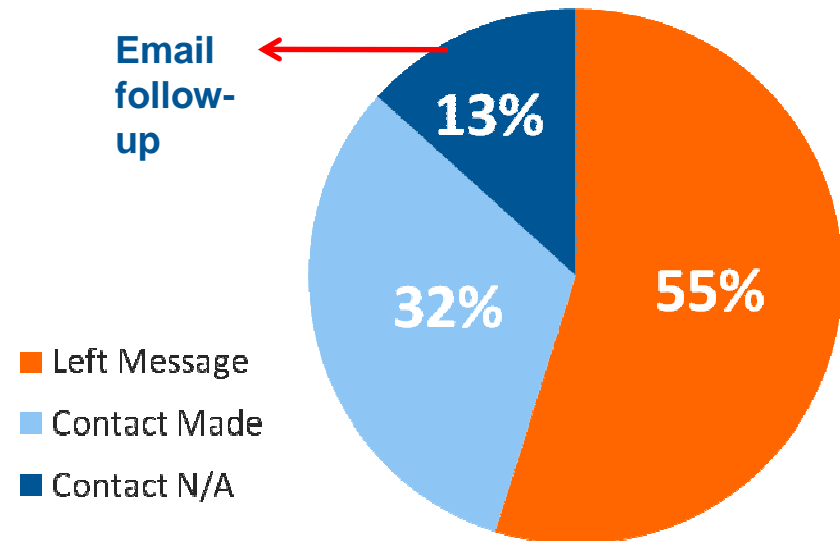
- Navigenics' staff of certified genetic counselors performed proactive phone outreach to individuals who received test results

PURPOSE:

- Are people aware that GC services are available?
- Do they have a sense of the value of GC sessions?
- What impact does proactive follow-up have on people's understanding of results and next steps?

Outreach outcomes (June)

- Outreach caused session uptake to increase > 3x
- GC session characteristics
 - Focus on 1-4 conditions
 - Avg: 35 min
 - Range: 15-65 min
- GC sessions scheduled
 - Proactive = 30%
 - Reactive = 70%



GC session outcomes

- ✓ Most don't know GCs are available (especially at no charge) – value is underestimated
- ✓ Majority say speaking with the GC was helpful, reassuring, helped them understand their results, and they'd recommend it to others
- ✓ People are enthusiastic about testing, satisfied, and not anxious from results
- ✓ People are enthusiastic about participating in Scripps

Conclusions

- Utilize data to improve our services
 - Consistent with HIPPA
- External research (with any potential PHI)
 - Partner with reputable institutions
 - Require unique IRB approval
- Goal is to make data accessible to clients and beyond
- Many issues raised in relation to DTC services fundamentally reflect larger issues that need to be more broadly addressed

Thank you!

