

A Systematic Approach to Genetics for Early Disease Detection and Intervention

June 17, 2010

James O'Leary
Genetic Alliance

Posted in the Resource Repository at:
<http://www.resourcerepository.org/documents/1965/asystematicapproachtogeneticsforearlydiseasedetectionandintervention/>



**Genetics for Early Disease Detection and
Intervention to Improve Health Outcomes
(GEDDI)**

**James C. O'Leary
Genetic Alliance**

GEDDI





Goal

Clinical, genetic and family history information for early diagnosis of disease



Improved health outcomes

Timeline

September	Invite Working Group		
November	Working Group Call		
December	Identify ?'s and Interviewees	Schedule Salons	
January	Interviews	Salons	
February	In-person Meeting	Schedule Webinar	Survey
April-May	Working Group Call	Draft White Paper	Workshop Agenda
June-July	Webinar	Final White Paper	Workshop

Early Disease Detection Salons

- No agenda
- Three responsibilities
 - ▶ Think beyond turf
 - ▶ Bring whole self
 - ▶ Be open to different perspectives
- 4 salons
 - ▶ Washington, DC
 - ▶ Ann Arbor, Michigan
 - ▶ Salt Lake City, Utah
 - ▶ Atlanta, GA

Stakeholder Interviews

- 21 interviews conducted
- 30 minutes each
- Mix of public health, clinical, and consumer focus



Interview Questions

- **Question 1: Which genetic conditions are ready for widespread screening and intervention in a public health context, if any?**
 - ▶ Follow-up 1: Why are these ready? What criteria makes conditions “ready” in terms of evidence, successful examples, existing infrastructure, and/or resources?
 - ▶ Follow-up 2: What opportunities do you see being missed for these conditions (prompts: late diagnosis, improper follow-up, education, etc.)?
 - ▶ Follow-up 3: Have you or your organization attempted to implement any systems in public health or clinical care for these conditions? Were they successful? If they were not successful, why?

Interviews, cont.

- **Question 2: Are there major gaps in evidence/information that need more examination (prompts: evidence base, methods, infrastructure issues, workforce, information and education)?**
 - ▶ Follow-up 1: Can you give specific examples of conditions that would be close to meeting the evidence threshold with this additional research?
 - ▶ Follow-up 2: What research/review would help us to identify these types of conditions that are “ready” or close to ready for organized screening and intervention systems?
 - ▶ Follow-up 3: What other criteria would make a condition “ready” (If not answered earlier)?
 - ▶ Follow-up 4: How do we maximize the benefits of unexpected findings that result from screening or testing? (prompts: results of uncertain significance, unexplained phenotype)

Interviews, cont.

- Question 3: If you were to develop an early disease detection and intervention campaign for these conditions, who would it be directed toward and how would you conduct it (If not answered earlier)?
- Question 4: Are there any other things the working group should focus on? Anything else they should know?

Survey of Working Group

1. Which genetic conditions are ready for widespread screening and intervention in a public health context, if any? Please include prenatal, newborn, childhood, and/or adult conditions.
2. What criteria makes these conditions ready, in terms of evidence, successful examples, existing infrastructure, and/or resources?
3. What opportunities do you currently see being missed for these conditions (such as late diagnosis, improper follow-up, education, etc)?
4. For conditions being considered for screening generally, what are the major gaps in evidence/information that need more examination (for example, evidence base, methods, infrastructure, workforce, information and education, etc)? Please explain.
5. Please describe any examples of successful systems (in your opinion) with the group.

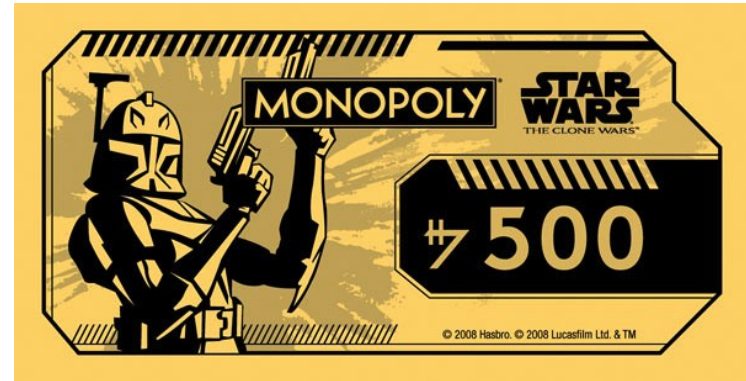
Interview, Survey, and Salon Findings

- Conditions and Criteria
- Gaps and Solutions
- Unexpected findings



Put your money where your mouth is!

A Game of Funding Priorities



- Rules:
 - ▶ You must choose one and only one funding priority
 - ▶ You are the sole funding source. Don't assume others will fund anything.
 - ▶ These are meant to be mid-range priorities.

Exemplar conditions

- Introduction
- Defining Early Disease Detection
- Core Criteria
- Exemplar Conditions
- Hearing Screening
- Duchenne Muscular Dystrophy
- Familial Hypercholesterolemia
- Lynch Syndrome
- Discussion

Thoughts, questions, comments?