



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

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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
March-April	Working Group Call	Draft White Paper	Workshop Agenda
Future	Webinar	Final White Paper	Workshop

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

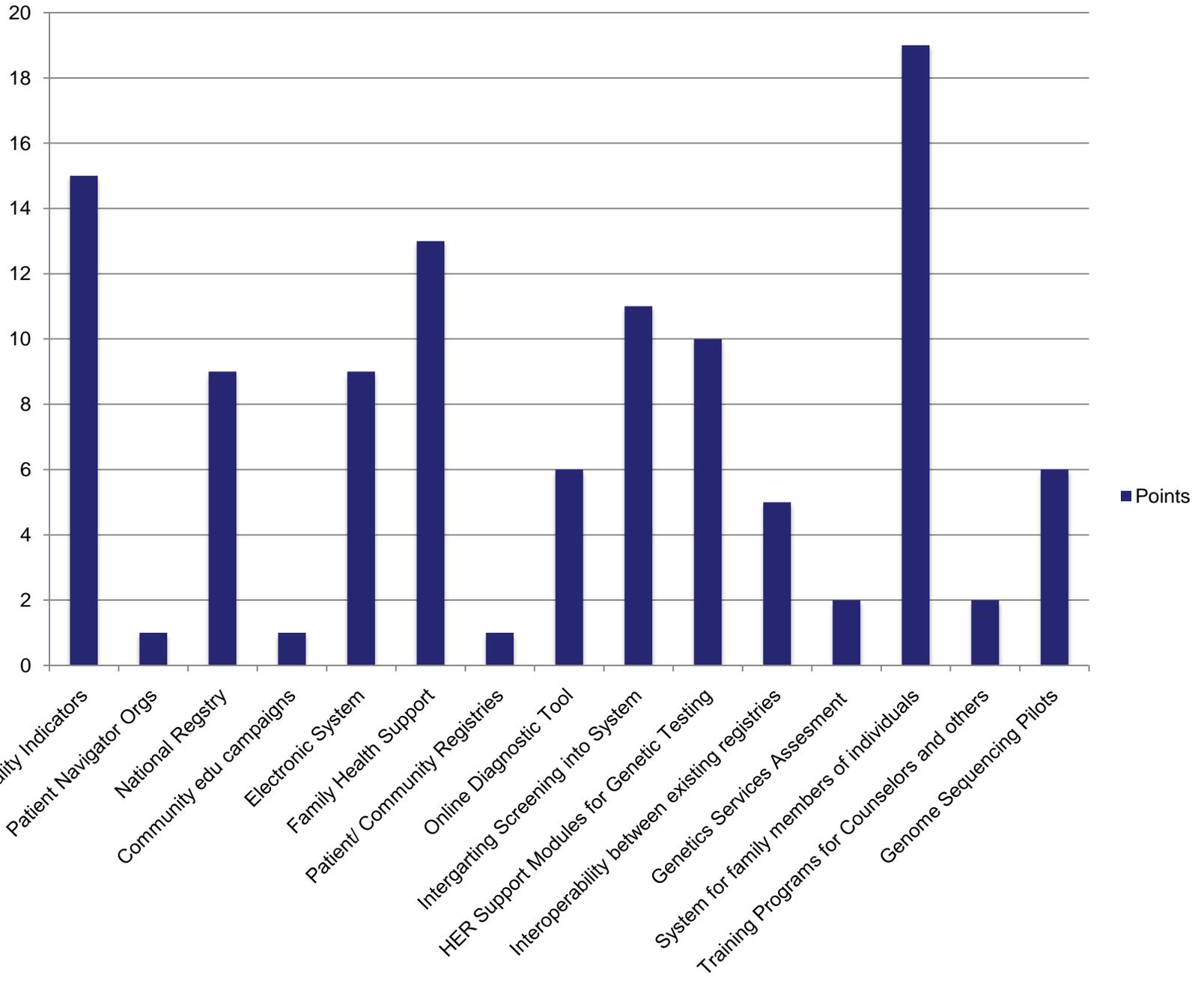
1. Which genetic conditions are ready for widespread screening and intervention in a public health context, if any? Please include prenatal, newborn, childhood, and/or adult conditions.
2. What criteria makes these conditions ready, in terms of evidence, successful examples, existing infrastructure, 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



Funding Priorities



Core criteria

Question 1: “Is there direct evidence that screening for the condition at birth leads to improved health outcomes for the infant or child to be screened? Are there potential benefits for the child’s family?”

Question 2: “Is there a case definition that can be uniformly and reliably applied. What is the incidence and prevalence of the condition? What are the natural history and spectrum of disease of the condition, including the impact of early recognition and treatment versus later recognition and delayed or no treatment?”

Question 3 : “Is there a screening test or screening test algorithm for the condition with sufficient analytic validity?”

Question 4: “Has the clinical validity of the screening test or screening algorithm, in combination with the diagnostic test or test algorithm, been determined and is that validity adequate?”

Question 5: “What is the clinical utility of the screening test or screening algorithm? What are the benefits associated with the use of the screening and diagnostic tests and the treatment? What are the harms associated with screening, diagnosis and treatment?”

Question 6: “How cost effective is the screening, diagnosis, and treatment for this disorder compared with usual clinical case detection and treatment?”

Exemplar conditions

Hearing Screening
DMD

Lynch Syndrome
Familial Hypercholesterolemia

Thoughts, questions, comments?