Genomic Medicine XV: Genomics and Population Screening
November 8-9, 2023
Hyatt Regency Bethesda

Goal: To identify needs, opportunities, and challenges for using genomics in population screening. Persistent barriers and evidence gaps will be examined as opportunities for additional research.

Objectives:

- Review the current state of population genomic screening in the US
- Examine obstacles and opportunities for expanded screening and available evidence of the impact of screening on outcomes and cost
- Identify research directions to inform expanded screening as appropriate
  - Variants and conditions to be screened for
  - Populations to screen, including appropriate ages and other characteristics
  - Role of community engagement
  - Providers to order screening and manage results (primary care, specialty care, health department, etc.)

Wednesday, November 8

8:30	**Breakfast (30min)**	
9:00	**Welcome and Introductions**	
10’	Goals of Genomic Medicine XV	Gail Jarvik
5’	Structure, Goals, and Products of Prior NHGRI Genomic Medicine meetings	Teri Manolio
9:15	**Session 1: Laying the Groundwork (1hr30min)**	Moderators: Eric Green, Marc Williams
20’	Keynote 1: Genomic Screening and the Reverend Bayes - Principles of screening studies - Differences between genetic/genomic screening and “non-genetic” population screening - Differences between molecular findings and clinical diagnosis	Les Biesecker
20’	Keynote 2: Genomic Screening: Who is Ready? - Current state of genomic screening: newborn screening, NIPT, pre-conceptual carrier screening, adult screening programs, polygenic risk scores - ACMG working group charge/report	Mike Murray
50’	Discussion	
10:45	**Break (20min)**	
11:05	**Session 2: Genomic screening technologies (1hr30min)**	Moderators: Jeff Brosco, Erin Ramos

15’	Technical and Logistical Approaches to Genomic Screening	Christine Eng
15’	Genomic Sequencing in Public Health Newborn Screening—Hopes and Challenges	Robert Currier
15’	Considering the factors involved in calculating the “number needed” in genomic screening	Jonathan Berg
45’	Discussion	
12:35	**Lunch (45min)**	
1:20	**Session 3: Logistics of population screening (1hr45min)**	Moderators: Carol Bult
15’	Opening the Flood Gate of Results, are we ready, and how will we handle this in Health Care?	Mylynda Massart
15’	Whom to screen when and how	Peter Kraft
15’	Addressing the challenges of genomic screening in populations underrepresented in genomic databases	April Adams
15’	Provider training and patient education: needs and opportunities	Kelly East
45’	Discussion	
3:05	**Break (20min)**	
3:25	**Session 4: Community engagement and population genomic screening (1hr45min)**	Moderators: Rex Chisholm, George Mensah
15’	American Indian/Alaska Native Community Engagement Preferences and Tribal Code Requirements	Vanessa Hiratsuka
15’	Opportunities for Meaningful Indigenous Community Engagement for Population Genomic Screening	Krystal Tsosie
15’	Advancing Genomic Research through Community Engagement: The All of Us Research Program	Minnkyong Lee
30’	Panel: Selecting conditions for screening	Caitlin Allen, Ned Calonge, Jessica Hunter, Bob McNellis
30’	Discussion	
5:10	**Adjourn Day 1**	

Thursday, November 9

8:00	**Breakfast (30min)**	
8:30	**Welcome**	
10’	Day 1 Recap	Gail Jarvik Teri Manolio
8:40	**Session 5: Evidence needed to support screening (1hr45min)**	Moderators: Pat Deverka, Dan Roden
15’	The Value Proposition for Population Genomic Screening	Marc Williams
15’	Cost effectiveness of population genomic screening	David Veenstra
15’	Uptake and Follow-up on Population Genomic Screening: Experience in the Alabama Genomic Health Initiative	Bruce Korf
15’	APOL1 Screening: Opportunities, Lessons Learned and Evidence to Support Screening	Carol Horowitz
45’	Discussion	
10:25	**Break (20min)**	
10:45	**Session 6: Obstacles to screening (1hr45min)**	Moderators: Gillian Hooker, Dan Rader
15’	Why are payers reluctant to cover genetic screening tests?	Michael Hultner
15’	Why is sharing genomic data so challenging?	Robert Freimuth
15’	How do we integrate genomic medicine into the EHR?	Kate Nathanson
15’	Why is it so hard to see health benefits from genetic screening?	Alanna Rahm
45’	Discussion	
12:30	**Break to grab lunches (15min)**	
12:45	**Research Directions (1hr)**	Moderators: Gail Jarvik, Teri Manolio
15’	Research on pre-testing phase: getting providers and patients/public to take up testing	Marc Williams

15’	Research on testing phase: what tests to recommend, how are they offered and what types of results are provided	Heidi Rehm
15’	Research in follow-up to testing: uptake of interventions, cascade testing, transportability throughout lifespan	Bruce Korf
15’	Discussion	
1:45	**Summary and Next Steps (30min)**	Gail Jarvik Teri Manolio
2:15	**Adjourn Day 2**	