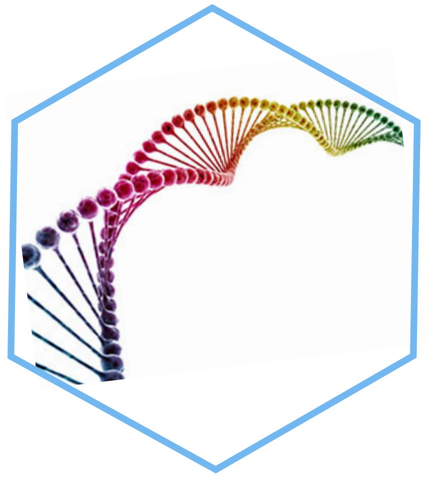
IMPLEMENTING NEWBORN SEQUENCING AT SCALE

Health Systems Challenges and Opportunities



Sylvia Mann, M.S., C.G.C
Supervisor,
Hawaii Department of Health Genomics Section
Project Director,
Western States Regional Genetics Network
sylvia@hawaiigenetics.org

WSRGN
Western States Regional Genetics Network

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DISCLOSURE

I have no financial interests or relationships to disclose.

My Journey to Here

- 35 years as a genetic counselor
- 30 years overseeing a state newborn screening program
- Over 20 years as the project director of the Western States Regional Genetics Network (AK, CA, Guam, HI, ID, OR and WA). It is one of seven Health Resources and Services Administration funded Regional Genetics Networks which work to increase access to genetic services.
- One of the few clinical genetic providers employed by a state health department outside of a laboratory.
- Oversee the Minority Genetic Professionals Network to increase ethnic and racial minorities to enter genetics professions and support practicing minority genetics providers.

Public Health System Challenges

- Continuous pressure to add new disorders
- Disorder screening and follow-up is becoming more complex for NBS programs, primary care providers, specialists, and families
- Loss/ Shortage of Workforce

  Lack of clinical genetics trained staff
- Weekday, Weekend, Holiday Work Schedule
- Increasing Concerns about Data Use, Privacy, and Security
- Increasing Refusals

Healthcare System Challenge

1.9-2.2 Clinical Geneticists per 1 million individuals*

(51 cardiologists per 1 million
57 adult orthopedic surgeons per 1 million
34 neurologists per 1 million)**

2019 Survey*
- 36% have practiced 21 yrs or more
- 25% plan to retire in next 10 yrs
- Less than 20% geneticists are non-White

Approximately, 6,000 ABGC certified genetic counselors***

45% response on 2023 NSGC survey
- 60% <10 yrs experience
- 68% <40 yrs old
- 11% non-White****

Non-genetics healthcare workforce not well trained to understand, provide accurate interpretation, and act on genetic test results

Increasing concerns about data use, privacy, and security

*2019 ACMGG Workforce Survey
**Bureau of Labor Statistics
***American Board of Genetic Counselors
2023 NSGC Professional Status Survey

ADDITIONAL CHALLENGES TO USE IN PUBLIC OR PRIVATE HEALTH SETTINGS

Consent

Genomic sequencing does not seem to be part of mandated NBS. How will informed consent be obtained especially for families that don’t speak English well or have low literacy or understanding?

Health Literacy and Access

Individuals have difficulty understanding health information especially if English is not their primary language.

Individuals, especially underserved individuals, may have difficulty using the internet to find information and often have difficulty even accessing a device with a cellular or internet connection.

Value of Results

Currently, non-white families have a higher chance of receiving results that can not be interpreted (variants of unknown significance) at the time of testing. This puts an additional burden on these parents.

Duty to Re-contact

What type of work force or system will be needed to re-contact families when new findings about the genomic results occur over time?

ACTIVITIES TO SUPPORT INITIATIVE

- Develop a healthcare provider work force to support genomic sequencing of healthy individuals in all communities in the U.S.
  - Incentivize training about genomic sequencing and its use in healthcare
  - Provide continuing education
  - Provide just in time genetics expertise support to healthcare providers

- Develop genetics work force to support genomic sequencing of healthy individuals in all communities in the U.S.
  - Fund training programs for this activity
  - Support more diversity in genetics professions
  - Support state health departments to have staff trained in clinical genetics and public health

- Develop supports for families
  - Increase genetics providers that represent the diverse families in the U.S.
  - Provide resources so that families understand what is being tested and the various outcomes
  - Provide navigators to families
  - Provide a centralized support system for families to ask questions and update information for re-contact

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