

THE HIDDEN COST OF NEWBORN SCREENING?

A DECADE OF DECLINING PUBLIC HEALTH GENETICS IN THE UNITED STATES



Sylvia M. Au, MS, CGC

State Genetics Coordinator

sylvia@hawaiiogenetics.org

Arthur Yu, MS, CGC

Genetic Counselor/Project Specialist

Hawai'i Genetics Program

Presenter Disclosure

The following personal financial relationships with commercial interests relevant to this presentation existed during the past 12 months:

No relationships to disclose

Public Health Genetics (PHG)

Public Health Genetics is an multidisciplinary field that applies advances in human genetics, genomics, and molecular biotechnology to improve public health and prevent disease

Institute for Public Health Genetics – University of Washington

Introduction

- ◎ 2002 – Secretary’s Advisory Committee of Genetics, Health and Society (SACGHS)
 - Assess integration of genetics into health care
 - Study the implication of these applications
 - Identify opportunities and gaps
 - Examine current practices
 - Serve as public forum on genetic / genomic issues

Introduction

- 2003 – Secretary’s Advisory Committee on Heritable Disease in Newborns and Children (SACHDNC)
 - Provide recommendations on projects under Heritable Disorders Program
 - Provide technical information to develop policies and priorities for newborn and child screening
 - Provide recommendations to aid the Secretary of Health reduce mortality / morbidity in newborns and children from heritable disorders

Introduction

- ◎ 2004 – HRSA grants awarded to form regional genetics collaboratives (RCs)
 - Part of effort is to improve health through translation of genetics into public health
- ◎ RCs goal is to strengthen and support genetics and NBS
 - Address maldistribution of services/resources

Regional Genetics and Newborn Screening Service Collaboratives



Relevance

“Translating the knowledge we are gaining from gene discoveries into practical clinical and public health applications will be critical for realizing the potential of personalized health care and improving the health of the nation.”

Muin J. Khoury, MD, PhD, Director, CDC Office of Public Health Genomics

Healthy People 2020

Improve health outcomes and prevent harm through valid and useful genomic tools in clinical and public health practices

Future of Genomics in Public Health

- ⦿ Evaluating evidence of valid / useful tests
- ⦿ Develop evidence based recommendations
- ⦿ Conduct translational research
- ⦿ Facilitate use of family health history to identify at-risk individuals
- ⦿ Monitor use of genetics and any disparities
- ⦿ Incorporate genomics into educational system
- ⦿ Assure the privacy of genomic information

Background

- Survey first done in 1999/2000 by Coalition of State Genetics Coordinators (CSGC)
- Aimed to collect and disseminate information to promote research, education and policy development in public health genetics (PHG)

Purpose

- Use results to provide nationwide snapshot of the status of state-level PHG activities
- Use results to strengthen or augment the policy decisions of state PHG programs and federal funding agencies

Methods

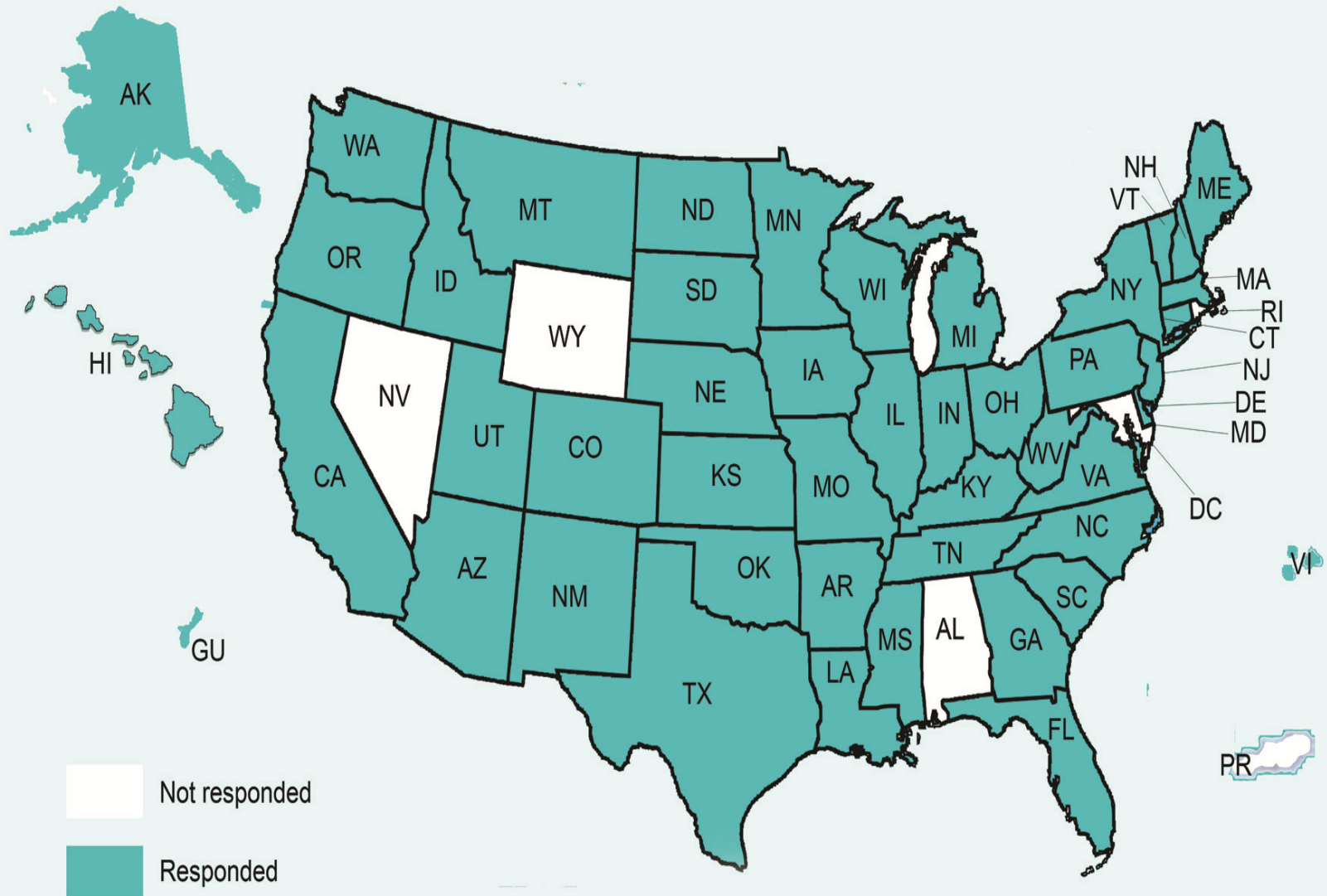
- Consisted of 11 sections & 62 questions
- Topics ranged from demographics/staffing/legislation and clinical services.
- SGCs for each state/US territory were confirmed by phone or email (N=54)
- All 54 SGCs invited to participate - those who agreed received a survey and were then contacted by phone for an interview

Methods

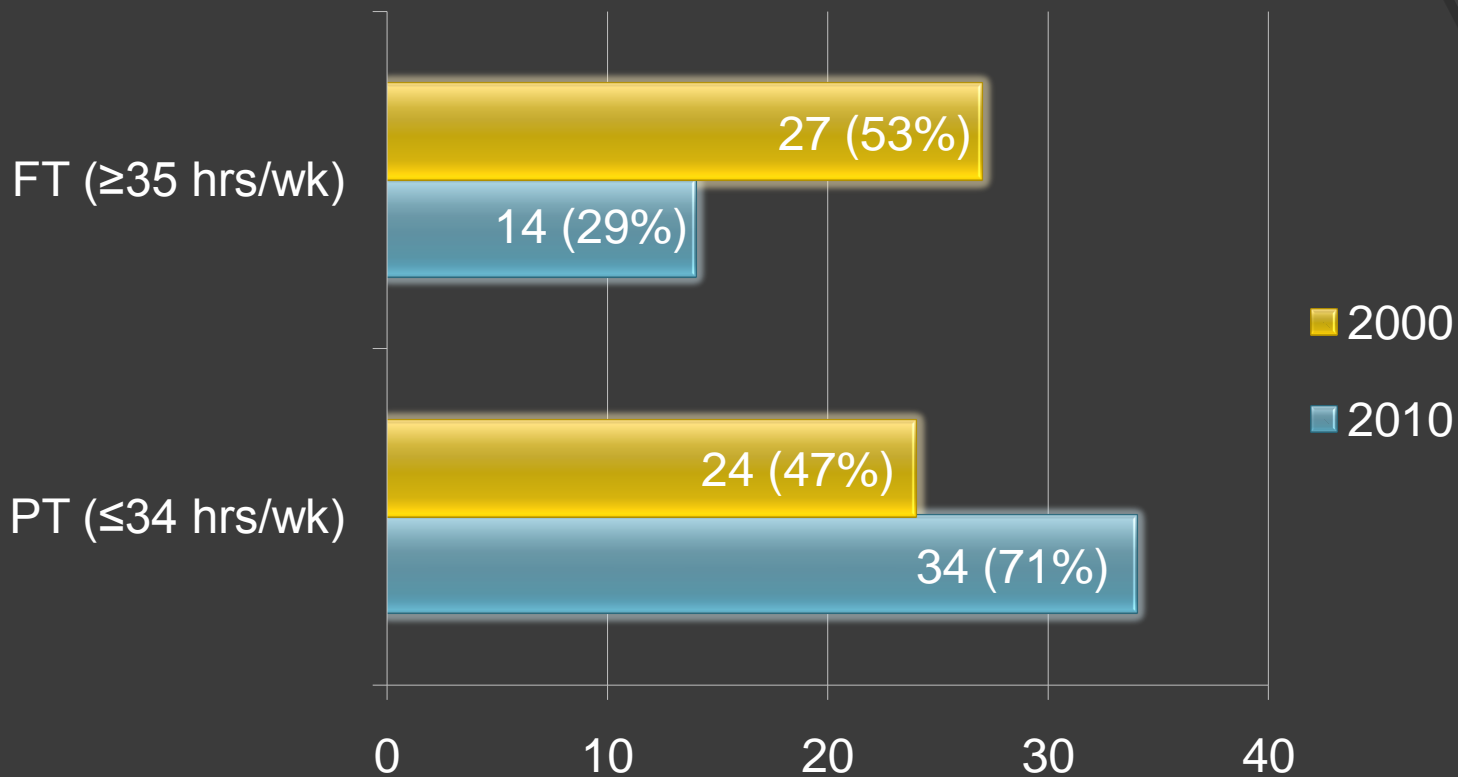
- A genetic counselor from the Hawai'i DOH interviewed the SGC to complete the survey (30-45mins)
- Before analysis, each survey was sent back to each SGC to review for inaccuracies
- All responses were compiled online (Survey Monkey) and analyzed utilizing SPSS and qualitative techniques

Results

YEAR	2000	2010
RESPONDENTS	51 / 54	48 / 54
PERCENTAGE	94.4%	88.9%



What did we observe?

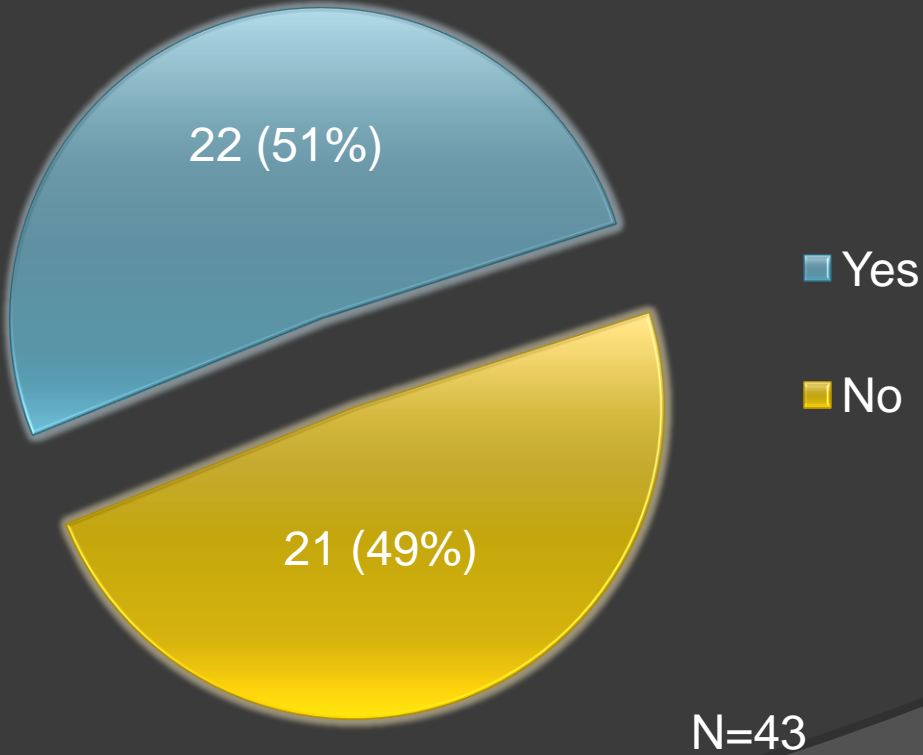


Number of Full-time / Part-time SGCs

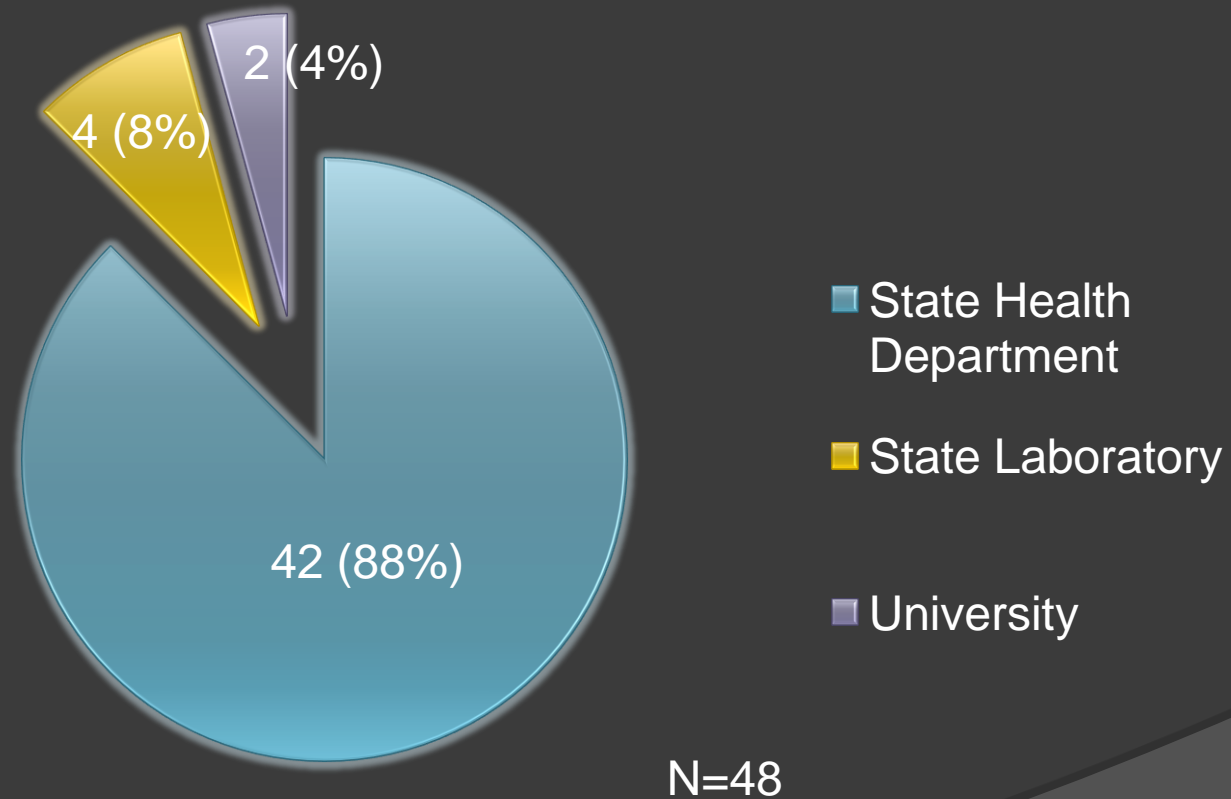
Other responsibilities

- ◎ Many of the SGCs were also:
 - **NBS coordinator**
 - **NBHS coordinator**
 - Birth Defects Program Coordinator
 - Laboratory Manager / Director
 - Clinician
 - Other

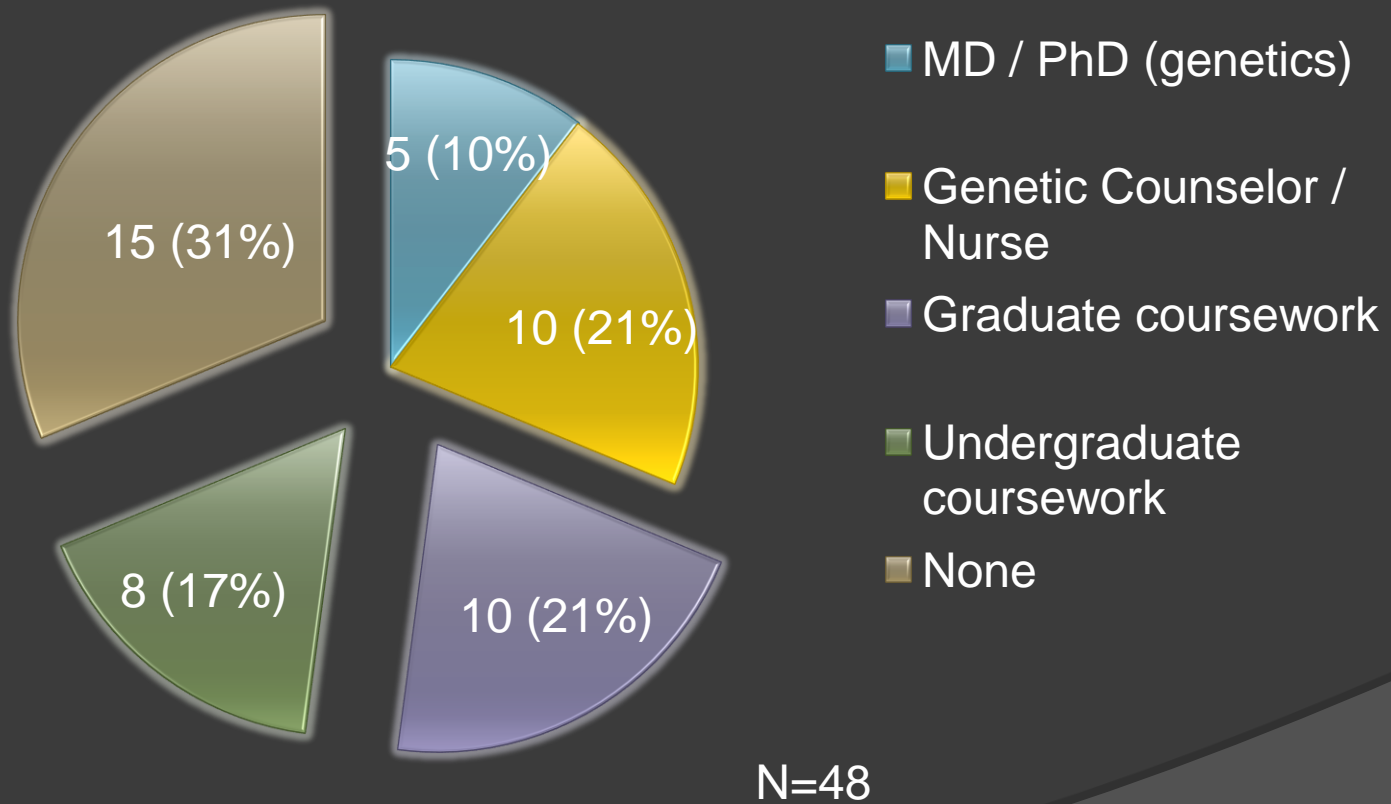
Do your responsibilities involve NBS related activities?



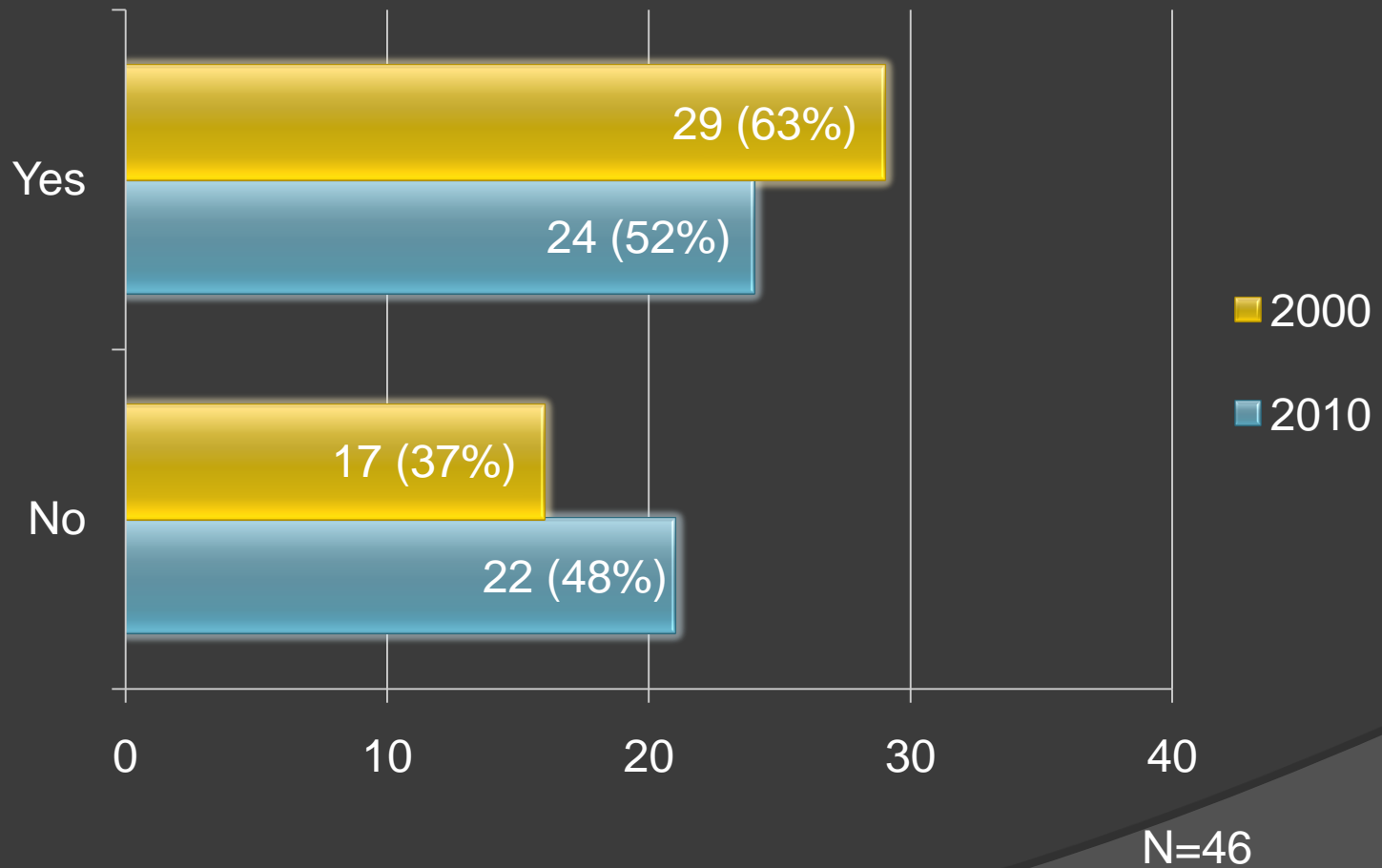
Where is your primary work setting?



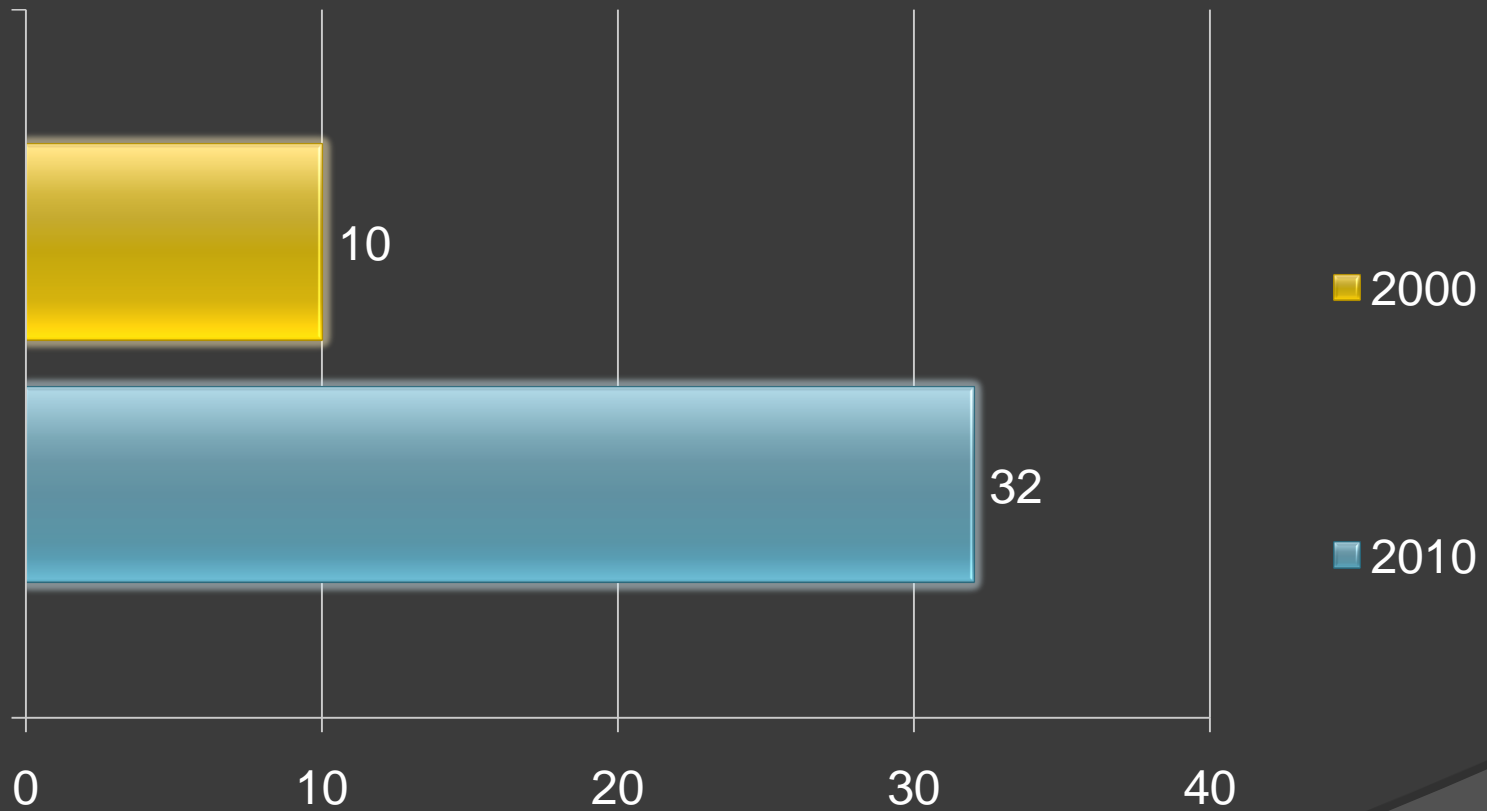
What is the highest level of formal genetics education that you have completed?



Genetics Advisory Committee: 2000 vs 2010



Has your state developed a state genetics plan?



What challenges do we face?

Challenges (State)

- The number of personnel and funding dedicated to genetics activities other than NBS has decreased substantially in the past decade.
- Lack of resources to sustain state level public health genomics activities outside of NBS.

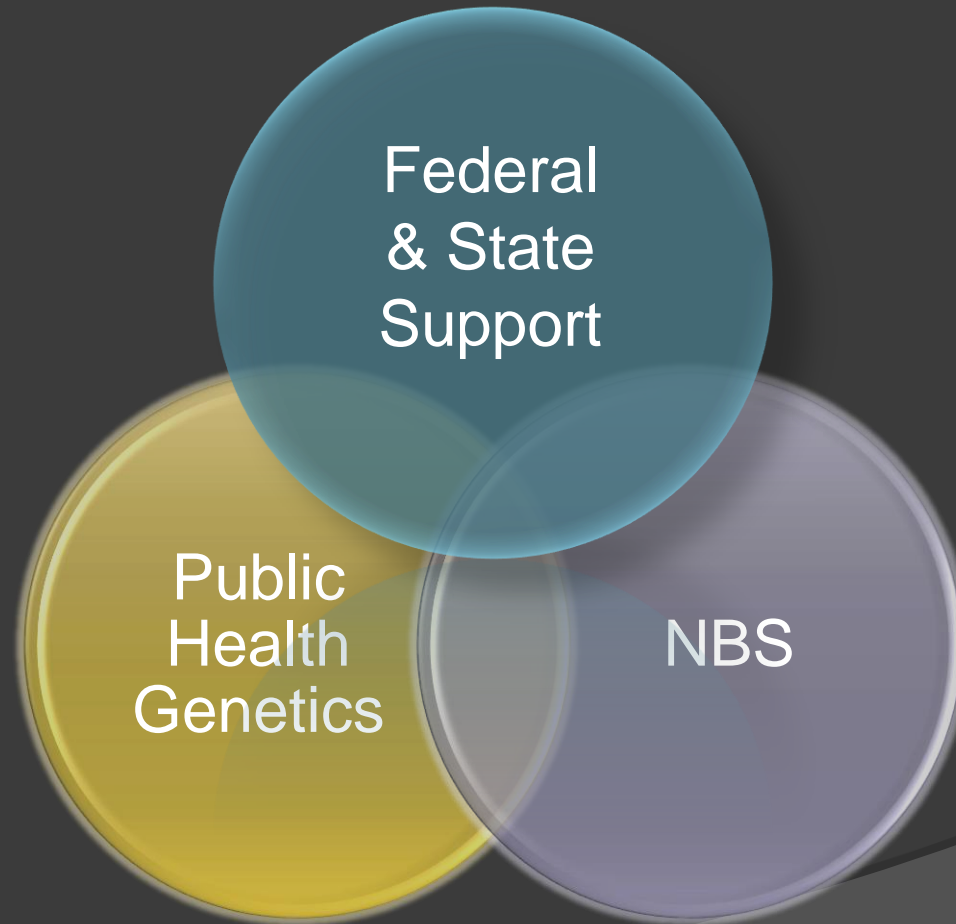
Challenges (State)

- Few states have strong and consistent leadership and programs for public health genetics.
- Lack of trained work force to lead translation of genomics into public health practice at the state level.

Challenges (Federal)

- Dissolution of the Secretary's Advisory Committee on Genetics, Health and Society
 - SACGHS addressed all major topics related to genetic / genomics
- CDC funding reduced for Office of Public Health Genomics (FY 2012)
 - Budget cutback of >\$11 million (~94%)

Imbalance



Take Home Message

The potential for genomics to improve public health can be significant. However, the imbalance between public health genomics and newborn screening activities has likely hindered the translation of genomics into public health practice.

Acknowledgements

Hawai 'i Department of Health

- ❖ Lianne Hasegawa, MS, CGC
- ❖ Kirsty McWalter, MS, CGC
- ❖ Sarah Scollon, MS, CGC
- ❖ Pauline Mui (Mother Hen)

State Genetics Coordinators across the country



...Mahalo

APHL 2014 ???

V.K. Taha