

# Public Health Genomics: Research and Education

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How can genomic knowledge  
improve the health and well-  
being of the population?



# Genetic screening

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Genetic evaluation of an asymptomatic person in a defined population, to detect an unsuspected disease or risk



***Take action***



Improve outcome



# Or...

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How can the benefits of newborn screening be extended?

- Detection of early cancer
- Cardiac prevention
- Potentially, any condition for which genetic susceptibility can be identified and preventive care is available



# Benefits beyond screening

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## Tests, e.g.

- Diagnostic
- Pharmacogenetic

## Biological products, e.g.

- Therapeutics
- Vaccines



# Harms of screening

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Harms of actions that occur with screening or medical follow-up

False positive results

Overdiagnosis

“Bluntly put, every adverse outcome of screening is iatrogenic and entirely preventable”

Grimes DA, Schulz KF. Uses and abuses of screening tests.  
Lancet 2002; 359: 881-4



# Blurry line between population health and medical care

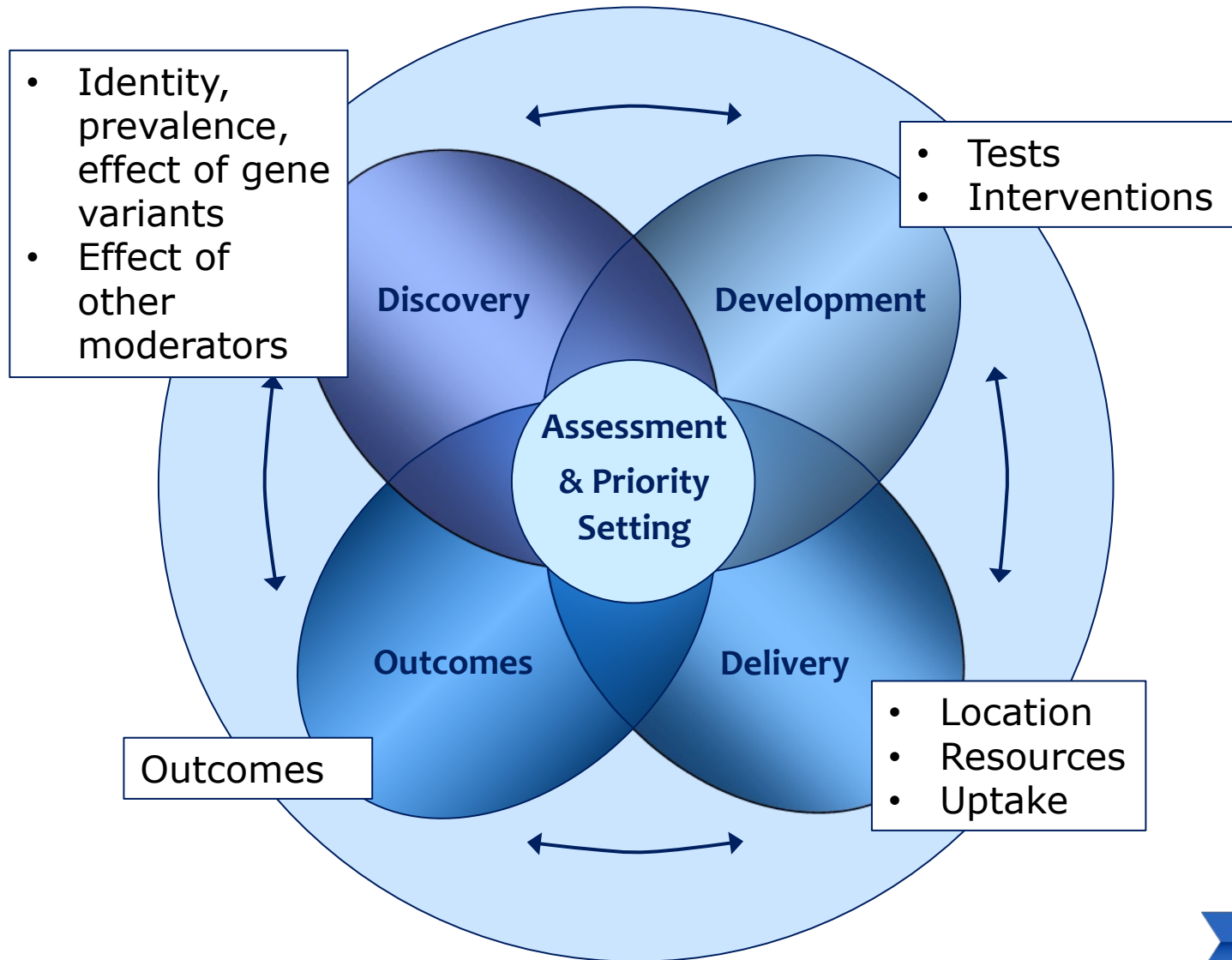
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Most screening occurs as part of medical care

Key research questions may have both medical and population health implications



# Pathway to benefit





# Multidisciplinary research agenda (a partial list)

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- Basic knowledge underlying tests and interventions
- Accuracy of tests
- Safety and effectiveness of interventions
- Knowledge synthesis to support policy development
- Implementation strategies



# University of Washington Institute for Public Health Genetics

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- Founded 1998
- Housed in Public Health with 7 participating UW Schools and Colleges, 3 partner institutions
- Offers MPH, PhD, MS in Genetic Epidemiology and Graduate Certificate for other grad students
- 50 faculty (4 PHG graduates); graduates also at CDC, NIH, State Public Health Depts, other universities – and in a range of health profession roles

<https://iphg.biostat.washington.edu>



# Institute for Public Health Genetics Curriculum

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- Requirements include genome sciences, genetic epidemiology, pharmacogenetics and ELSI (ethical, legal and social implications), with additional elective course offerings
- Multidisciplinary thesis research – eg, combining epidemiological and policy analysis
- Weekly interactive seminar

<https://iphg.biostat.washington.edu>

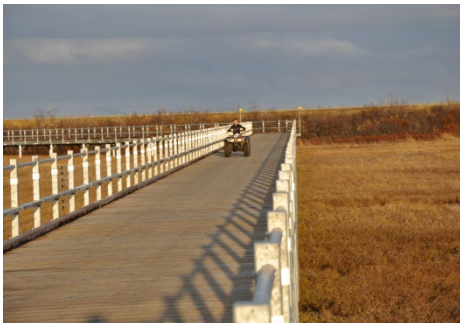


# Northwest-Alaska Pharmacogenomics Research Network (NWA-PGRN)

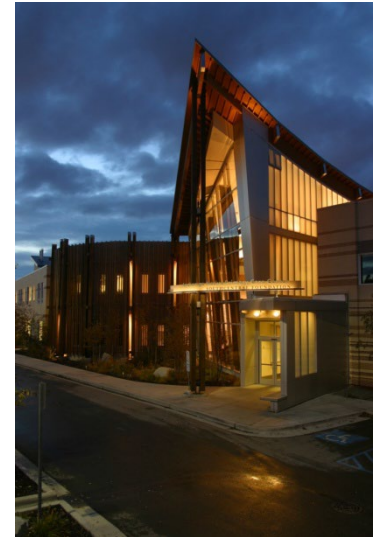
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Partnership-based network (2009-present)

PIs: Ken Thummel, Wylie Burke



- Southcentral Foundation  
Site PI: Denise Dillard
- Confederated Salish and Kootenai Tribes
- Yukon-Kuskokwim Health Corporation
- University of Washington
- University of Montana  
Site PI: Erica Woodahl
- University of Alaska Fairbanks
- Oregon Health and Science University  
Site PI: Bert Boyer



# NWA-PGRN: Focus on pharmacogenomics

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- Strong Tribal interest in research with potential to improve health care and in cost effectiveness – thus on applications that may have broad impact

In parallel:

- Commitment to incorporating community views about uses of genetics
- Independent research at each site, expanding scope



# Goals of Tribal research oversight

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- Prevent research abuses and group harm
- Assure **both** community benefit and good science
  - Research questions relevant to the community
  - Potential for community benefit
  - Opportunity for community to review/ contribute to research methodology



# Periodic partnership meetings

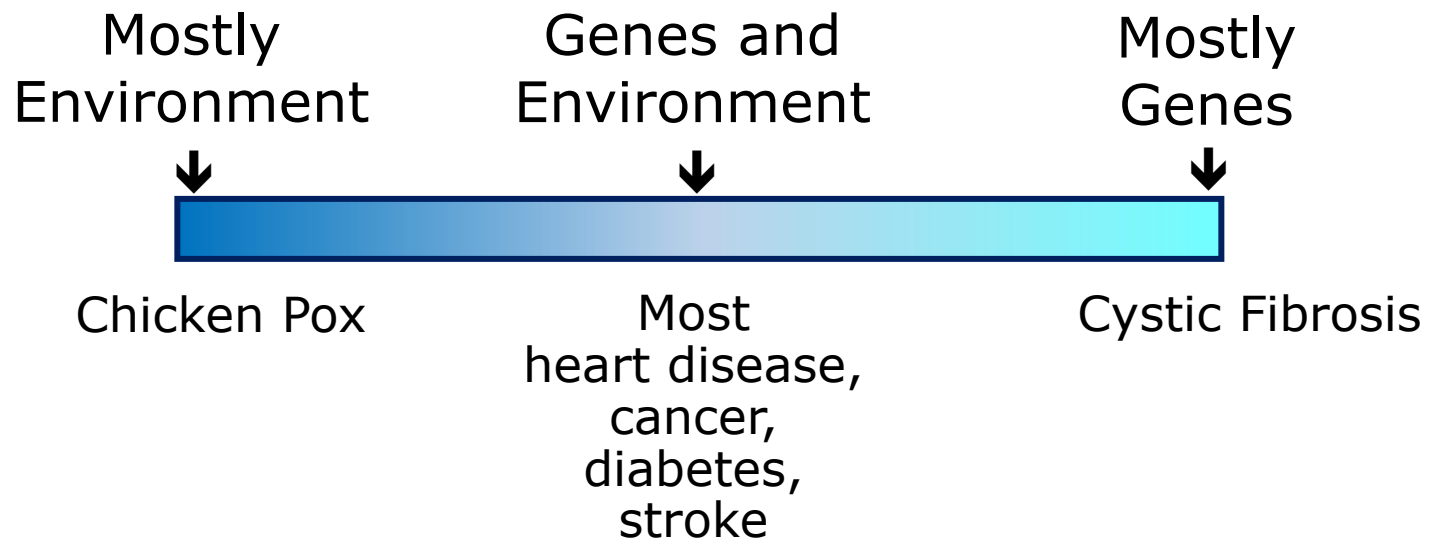
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- Discussion among researchers and community members
  - Share progress
  - Discuss research goals
  - Consider strategies to strengthen partnership



# Contribution of genes and social environment to health outcome

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Burke W. Ann Rev Genom Hum Genet 2021





# Including social determinants in genomic research

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Comment from 2019 partnership meeting:  
*"Omics data untethered to context is poor science at best and harmful at worst. In terms of cost-benefit analysis, Tribes need to know where to invest their time and money. This data is essential not only to contextualize research outcomes but also in determining whether a Tribe feels it's worth it to go down the path of genomic inquiry."*

Burke et al Prog Comm Health Partnerships 2022; 16(4):583-592.

