

Public Health Genomics: National and International Perspectives

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Using genome-based research to improve population health

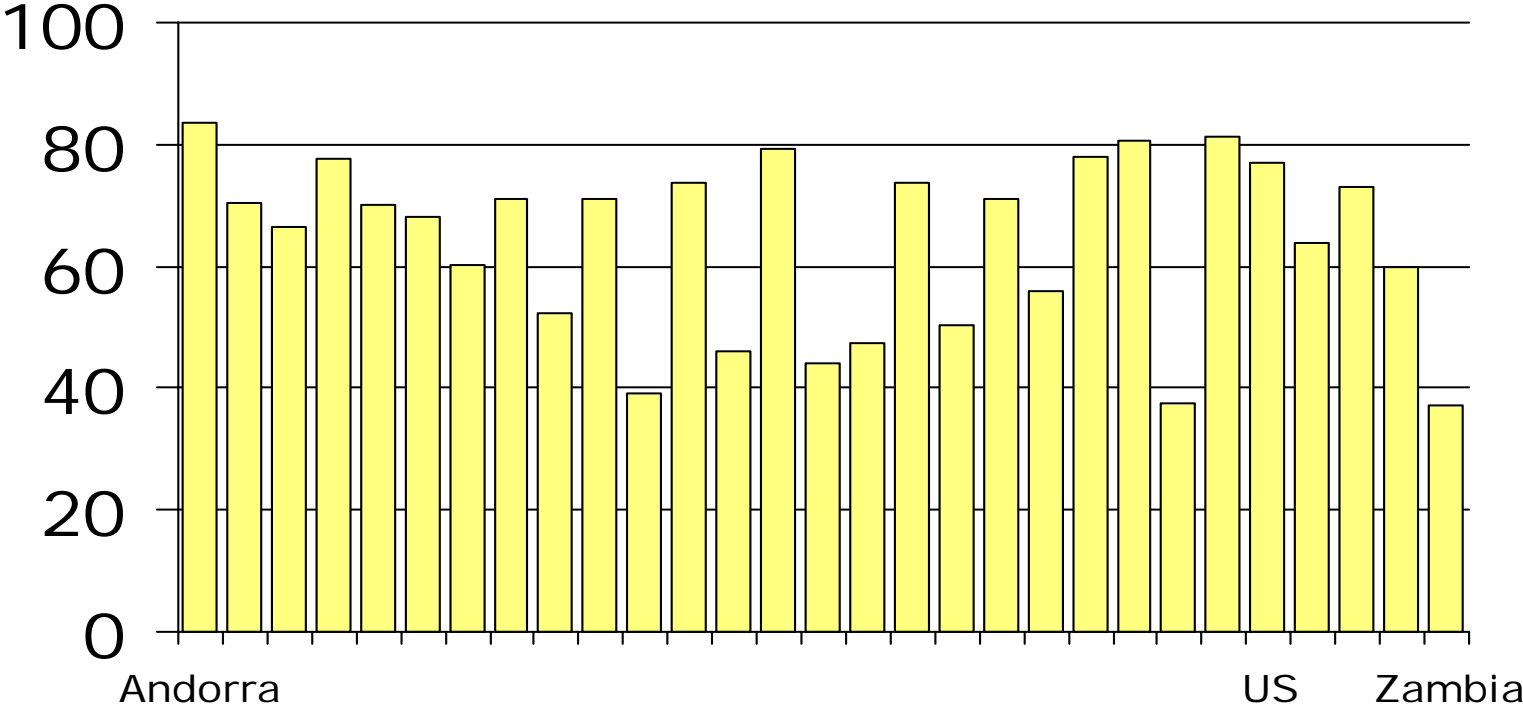
- Create and integrate knowledge
 - ↓
- Developing & evaluating health services
- Communication/stakeholder engagement
- Informing public policy
- Education and training

Burke et al for the Bellagio Group. Genet Med 2006;8:451-8



Life expectancy by country, 2000

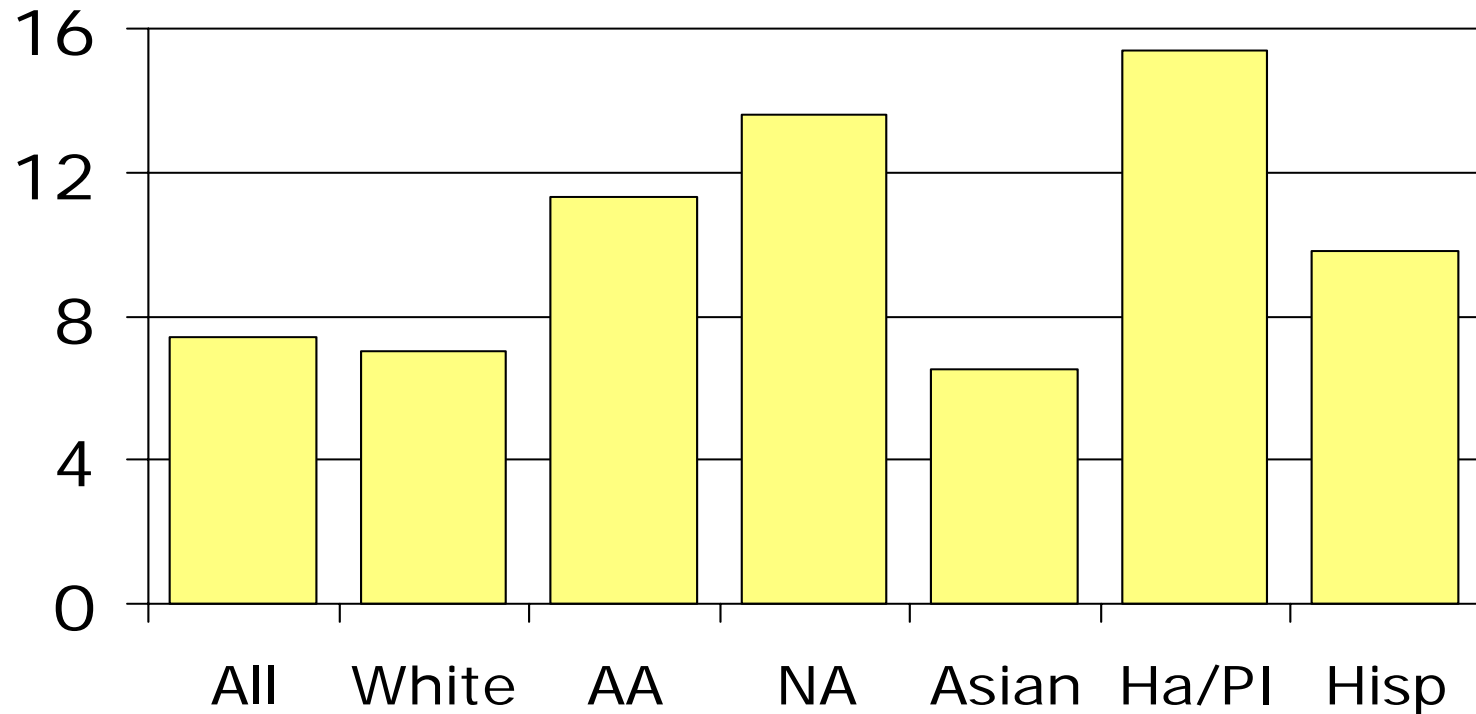
(Partial data)



US Census Bureau



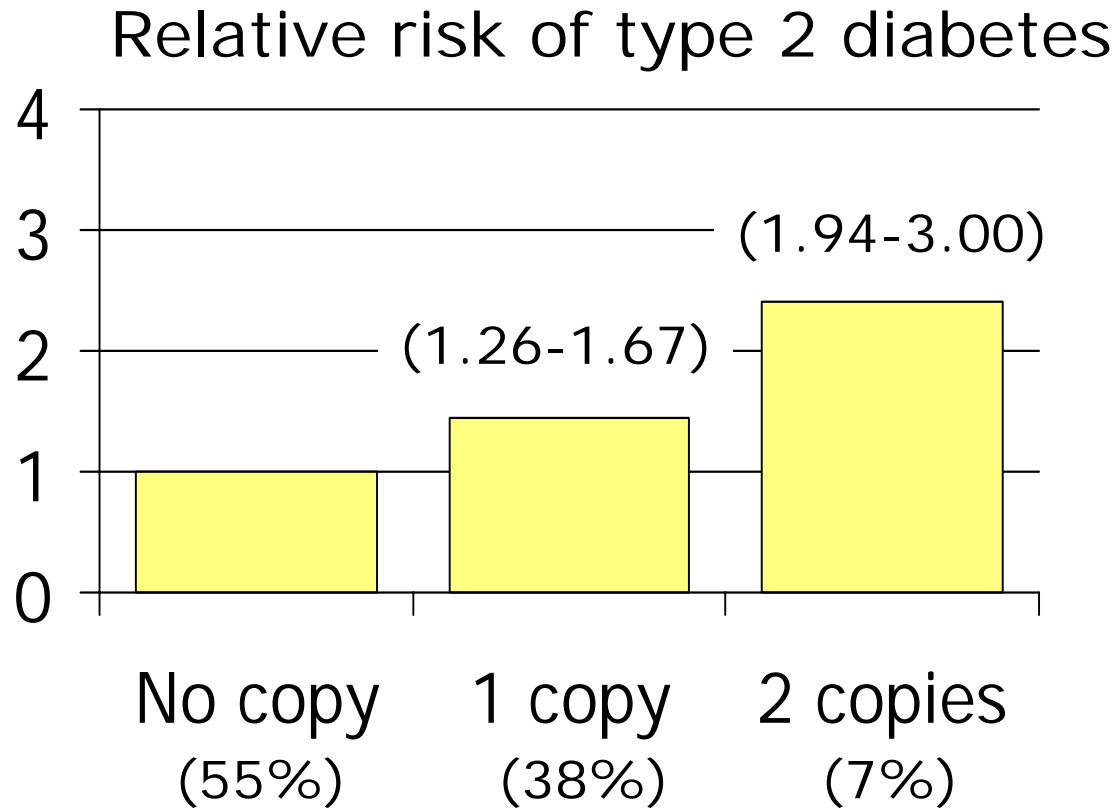
Age corrected % of US adults with diabetes, by race/ethnicity



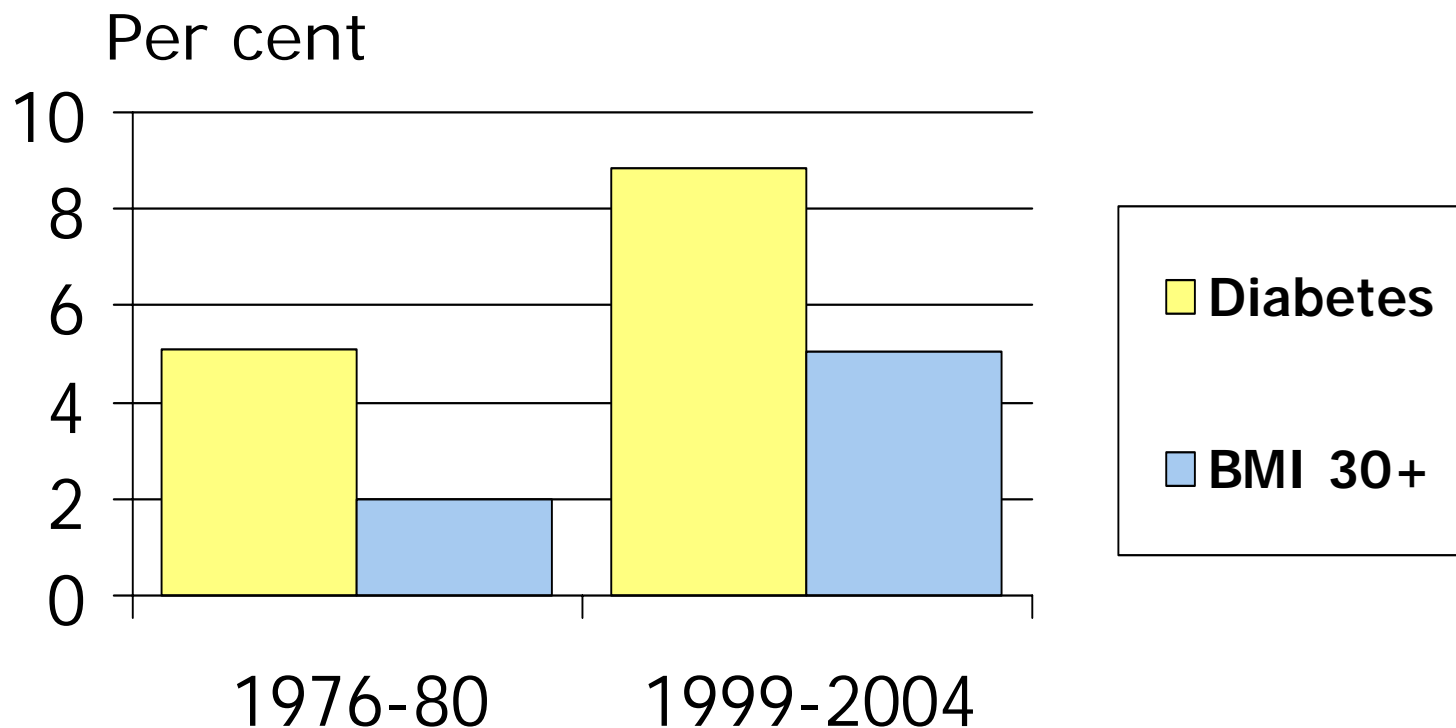
CDC National Health Interview Study 2005



TCF7L2 variant DG10S468 and type 2 diabetes



Increasing prevalence of diabetes and obesity in US



NHGRI Planning Process 2003

Grand Challenge II-6: Develop genome based tools that improve the health of all

- Social and other environmental factors are major contributors to health disparities
BUT
- Population differences in allele frequencies could be a contributing factor

“Research is needed to understand the relationship between genomics and health disparities by rigorously evaluating the diverse contributions of socioeconomic status, culture, discrimination, health behaviors, diet, environmental exposures and genetics”



Racial health disparities

- To what extent can genetic differences between racial groups explain group differences in health outcomes?
- What constitutes an adequate evaluation of genetic and alternative hypotheses?



Implications of “thrifty gene” hypothesis

“The focus on genetic causes of diabetes related to ‘race’ since the 1960s may well have deflected attention away from finding effective ways to manage what has, in the same period, become a global public health crisis.

“Those who study disease incidence using ‘race’ as a ‘crude proxy’ therefore have a duty to make clear just how ‘crude’ that proxy is and whether their language unnecessarily focuses on ‘racial’ distinction or even connects to racist discourses.”

Fee, Soc Sci Med 2006; 62: 2988-2997.



Group harm

- Not addressed in the Belmont Report or necessarily included in beneficence, respect for persons, or justice
- Should we consider 4th principle for research?
 - “Respect for communities”
 - obligation to respect values and interests of the community
 - wherever possible, protect community from harms

Emanuel & Weijer, Protecting communities in research, in *Belmont Revisited*, Eds. Childress et al, 2005



Call for tribal research regulation

- American Indian/ Alaska Native communities need to protect themselves from harm in research, to maximize benefit
- As sovereign nations, they have legal authority and imperative to regulate research occurring on their lands

Sahota, Research Regulation in American Indian/Alaska Native Communities: Policy and Practice Considerations – NCAI Policy Research Center



Brief history of a scientific finding

- Using samples from data repositories, Lahn and colleagues report a gene variant presumed associated with brain development, selected for in European and Asian populations, not in African

Science 2005; 309: 1720-2

- Subsequent studies

- Show no association with brain size or cognition

Science 2007; 317: 1036

- Dispute evidence of selection

Science 2006; 313: 172



Questionable history in the search for genetic explanations

“Minority groups, particularly blacks in the United States, are assumed to be genetically predisposed to virtually all common chronic diseases. Genes are regularly proposed as the cause when no genetic data have been obtained, and the social and biologic factors remain hopelessly confounded...”

Cooper et al, New Engl J Med 2003; 348:1166



How can genomic research help to diminish health disparities?

- Identify alternative disease mechanisms or modifying factors?
- Identify innovative, cost-effective therapies for the major public health burdens?



The central role of deliberation

- *Transdisciplinary* creation and integration of new knowledge
- Partnerships between communities and researchers
- Meaningful involvement of the public in policy-making



Methodological challenge: Effective deliberation

- Strong support from principle for deliberative processes, but little evidence on effectiveness
- Design not neutral for outcome
- Desirable characteristics
 - consultation with all parties affected by outcome
 - fair representation of scientists and stakeholders
 - high-quality syntheses of the scientific evidence
 - skillful chairing

Lomas et al. Conceptualizing and combining evidence for health system guidance, Canadian Health Services Research Foundation

www.chsrf.ca/other_documents/pdf/evidence_e.pdf



High expectations for the Human Genome Project

“Scientists wanted to map the human genetic terrain, knowing it would lead them to previously unimagined insights, and from there to the common good.”

Francis Collins

New Engl J Med 1999; 341:28



Achieving the common good

- Critical assessment of assumptions
- Genomics placed within the larger population health context
- Conversation across disciplines and perspectives

