## **Goals for the Future**

Eliminate reliance on the construct of race

Improve methods to interpret variation in relative and absolute risk across populations

Engage populations that adequately reflect human diversity

Choose populations on the basis of utility, not convenience (intensity of exposure, patterns of LD, etc)

Refine strategies to assess ancestry for regional populations – genome-wide and at define loci

### Health Status Measures in US Racial/Ethnic Groups, 2001

	White	Black	
Cause of Death	Age – Adjust	Ratio: Black:White	
All Causes	842.9	1101.2	1.3
Heart Disease	245.6	316.9	1.3
Coronary Heart Disease	177.5	211.6	1.2
Stroke	56.0	78.8	1.4
Cancer	197.4	243.1	1.2
COPD	47.0	30.9	0.7
Pneumonia / Influenza	21.7	24.1	1.1
Diabetes mellitus	22.1	49.2	2.2
HIV infection	2.1	22.8	10.9
Infant Mortality (/1000)	5.7	13.5	2.4
Life Expectancy	77.7	72.2	

Data Source: National Center for Health Statistics. Health, United States, 2003 With Chartbook on Trends in the Health of Americans. Hyattsville, Maryland: 2003.

#### Cancer Death Rates\* by Sex and Race, US, 1975-2003



\*Age-adjusted to the 2000 US standard population.

Source: Surveillance, Epidemiology, and End Results Program, 1975-2003, Division of Cancer Control and Population Sciences, National Cancer Institute, 2006.

### Cancer Death Rates\*, for Men, US, 1930-2003



\*Age-adjusted to the 2000 US standard population. Source: US Mortality Public Use Data Tapes 1960-2003, US Mortality Volumes 1930-1959, National Center for Health Statistics, Centers for Disease Control and Prevention, 2006.

# Female Breast Cancer Incidence Rates\* by Race and Ethnicity, US, (SEER), 1975-1976 to 1999-2000



\*Rates are age-adjusted to the 2000 US standard population. Source: Surveillance, Epidemiology, and End Results program, 1973-2000, Division of Cancer Control and Population Sciences, National Cancer Institute, 2003.

From Ghafoor, A. et al. CA Cancer J Clin 2003;53:342-355.

# Female Breast Cancer Death Rates\* by Race and Ethnicity, US, 1975-1976 to 1999-2000



\*Rates are age-adjusted to the 2000 US standard population. Source: US Mortality Public Use Data Tapes, 1969-2000, National Center for Health Statistics, Centers for Disease Control and Prevention, 2003.

From Ghafoor, A. et al. CA Cancer J Clin 2003;53:342-355.

### Geographic Clustering Based on Alu Polymorphisms



### A Neighbor-joining Tree Based on Variants in Angiotensinogen



### **Public Health and Molecular Genetics**

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Effect of Individual Loci	<u>Examples:</u>	<u>Drift</u>	<u>Selection</u>	<u>Few/Large</u> Many/	<u>/Small</u>
	Sickle cell		X	X	
	Lactase persistence		X	X	
$\downarrow$	Low skin melanin		?X	X	
	Prostate Cancer	<b>?X</b>		? moderate	
$\downarrow$					
	Hypertension	?X	?X	?X	
↓ ↓	Diabetes	?X	?X	few - ? mode	rate

#### Ongoing Adaptive Evolution of ASPM, a Brain Size Determinant in Homo sapiens

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**Supplementary Figure S2:** Frequency distribution of *ASPM* haplotypes in the 90-individual Coriell panel. Haplotypes belonging to haplogroup D are shown in open blue bars, whereas non-D haplotypes are shown in solid red bars.



### Recommendations

Caveat: Unfortunately the problems that need to be addressed are "systemic" (ie, widespread in theory and practice) not "programmatic" (ie, a result of inadequate technology, analysis methods, etc)

Moving beyond categories:

- 1. Analytic methods that allow pooling rather than stratification
- 2. Inclusion of more populations, particularly from Africa
- 3. Assessment of population structure and haplotype diversity of known susceptibility loci in cosmopolitan populations (eg, NYC)
- 4. Research on how genetic information can be used and communicated in minority populations
- More empirical research on reporting and using racial/ethnic labels

   assessing the trade-offs of inclusion vs. reinforcing stereotypes and bias.
- 6. Assess evidence of recent selection (use with caution . . ); migration