POPULATION-BASED CARRIER SCREENING FOR SINGLE GENE DISORDERS: LESSONS LEARNED AND NEW OPPORTUNITIES

February 6-7, 2008
Hilton Washington DC/Rockville Executive Meeting Center
Rockville, Maryland

Agenda

February 6

7:30 am	**Informal networking**
8:00 am	**Opening remarks: Why Now?** *Duane Alexander, M.D., Director, NICHD*
8:10 am	**Logistical details:** *Greg Feero, M.D., Ph.D.*
8:15 am	**The genome era: A public health perspective** *Ned Calonge, M.D., M.P.H.*
8:45 am	**An overview of the issues facing carrier screening in large populations** *Louis J. Elsas, M.D., FFACMG*
9:15 am	**Lessons learned from carrier screening: Tay-Sachs** *Robert J. Desnick, M.D., Ph.D./Elisa Ross*
9:45 am	**Discussion**
10:05 am	**Break**
10:25 am	**Lessons learned from carrier screening: Sickle Cell Anemia** *James R. Eckman, M.D./Janet Ohene-Frempong, M.S.*
10:55 am	**Discussion**
11:10 am	**Lessons learned from carrier screening: Cystic Fibrosis** *R. Rodney Howell, M.D./Martin Kharrazi, Ph.D.*
11:40 am	**Discussion**
11:55 am	**Remarks by Francis S. Collins, M.D., Ph.D., Director, NHGRI**
12:05 pm	**Working lunch: An update on technologies relevant to carrier screening** *Eric P. Hoffman, Ph.D.*
1:00 pm	**Current challenges: SMA** *Thomas W. Prior, Ph.D./Deborah Heine, J.D.*
1:30 pm	**Discussion**

1:45 pm **Current challenges: Fragile X**
Thomas J. Musci, M.D./Don Bailey, Ph.D.

2:15 pm **Discussion**

2:30 pm **Break**

2:55 pm **Carrier screening: Populations, stigmatization, and eugenics**
Keith A. Wailoo, Ph.D.

3:25 pm **Discussion**

3:45 pm **Small group sessions I**

A. What to screen for and when to screen? Developing criteria for disorder selection in the setting of economic and social constraints.	**Jackson Room Red**
B. How should we balance the screening interests of individuals, communities, and society?	**Lincoln Room Orange**
C. Should services be targeted to subpopulations? If so, on what basis can subpopulations be accurately identified? Balancing science, ethics, and clinical utility.	**Monroe Room Yellow**
D. How is informed consent defined and obtained? Models for multiple complex tests applied to the general population.	**Truman Room Green**
E. How can we measure the success of carrier screening programs? Developing an evidence base.	**Roosevelt Room Light Blue**
F. What will be the "next generation" screening methods? Technology development, screening, and the $1,000 genome.	**Washington Theater Black**

5:00 pm **Adjournment Day 1**

7:00 pm **Informal after-dinner coffee: Issues of interest**

February 7

7:30 am	**Informal networking**	
8:00 am	**Small group sessions II**	
	A. What to screen for and when to screen? Developing criteria for disorder selection in the setting of economic and social constraints.	**Jackson Room Red**
	B. How should we balance the screening interests of individuals, communities, and society?	**Lincoln Room Orange**
	C. Should services be targeted to subpopulations? If so, on what basis can subpopulations be accurately identified? Balancing science, ethics, and clinical utility.	**Monroe Room Yellow**
	D. How is informed consent defined and obtained? Models for multiple complex tests applied to the general population.	**Truman Room Green**
	E. How can we measure the success of carrier screening programs? Developing an evidence base.	**Randolph Room Light Blue**
	F. What will be the "next generation" screening methods? Technology development, screening, and the $1,000 genome.	**Washington Theater Black**
9:30 am	**Small Group Reports A, B, C**	
11:00 am	**Break**	
11:10 am	**Small Group Reports D, E, F**	
12:40 pm	**Concluding remarks: Defining the next steps** *Duane Alexander, M.D.* *Francis S. Collins, M.D., Ph.D.*	
1:10 pm	**Adjournment**	

Sponsored by:

National Human Genome Research Institute, National Institute of Child Health and Human Development, Office of Rare Diseases of the NIH, Health Resources and Services Administration, Centers for Disease Control and Prevention, Genetic Alliance, and American College of Medical Genetics