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