Newborn Screening Activities at the Centers for Disease Control and Prevention:

An Update for the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

June 5, 2006

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Emergency Response: Hurricane Katrina

- CDC assisted Louisiana's Department of Health and Hospitals/Office of Public Health in conducting a rapid assessment to determine the extent of newborn screening disruption in August and September, 2005.
  - Immediate impact:
    - Dissemination of new instructions to ensure optimal screening procedures
    - Prompting of hospital staffs to check log books for missing screening results
    - Identification of approximately 1200 infants in 53 hospitals with missing results, to be contacted for retesting

Descriptive Epidemiology of Missed or Delayed Diagnoses for Conditions Detected by Newborn Screening (Henderson et al.)

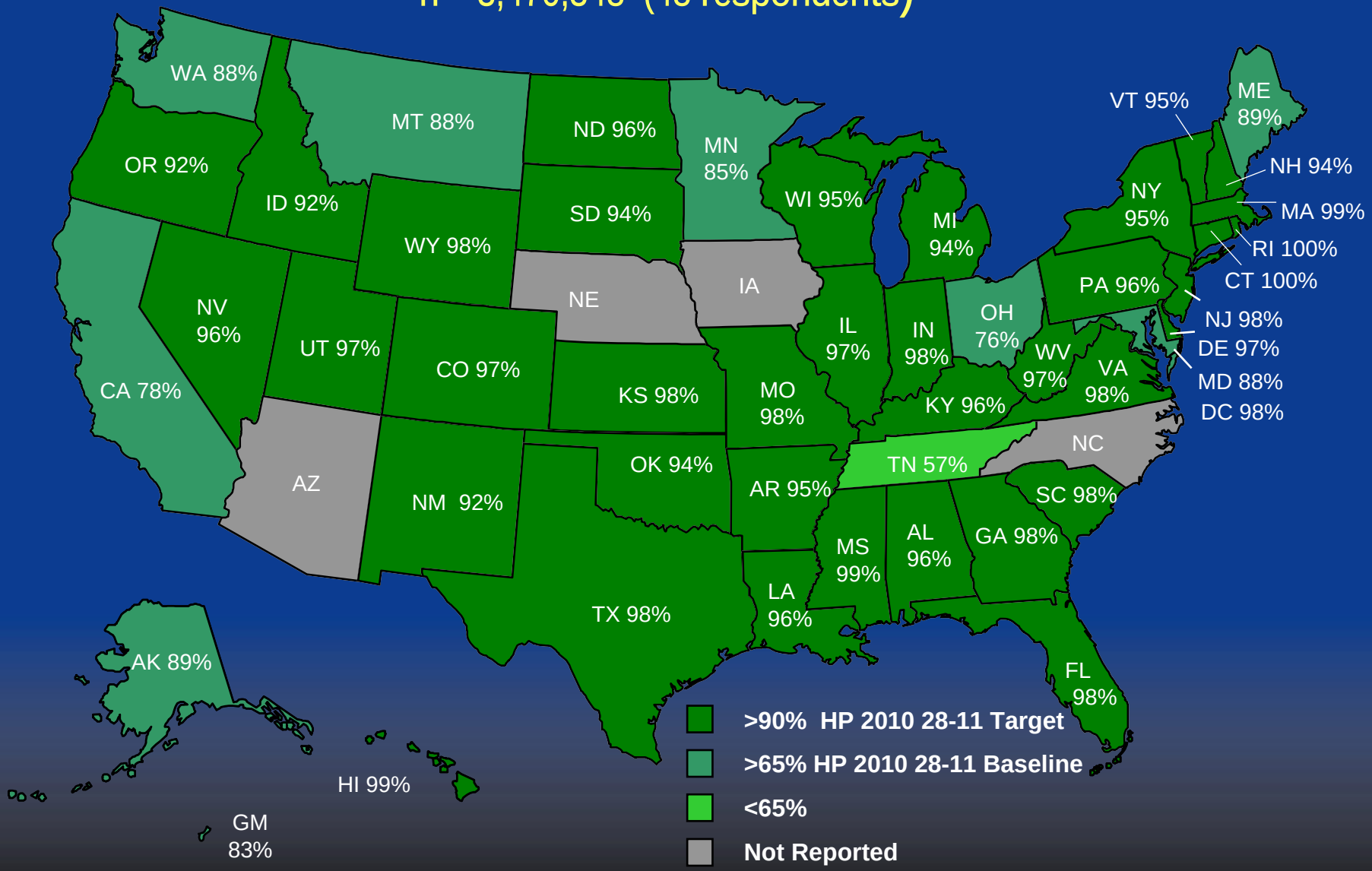
- Objective: To determine the number, reasons why and health outcomes for children with delayed or missed diagnoses
- Time Period: 1984-2004
- Methods: Survey of state NBS lab and follow-up personnel, metabolic clinics and affected families through parent advocacy groups
  - Identify reasons for miss/delay: specimen collection, specimen transportation, screening lab procedure, health provider practices, follow-up and biologic variants
  - Examine procedures and actions taken by state programs to identify such cases/ corrective actions to minimize recurrences
- Encourage routine sharing of such information; Identify procedures for routine surveillance of missed cases

Examining the Prevalence of SCID among Deaths in Children <18 mths in CA (Vogt, et al)

- CDC and the California State Department of Health Services will analyze 4000 newborn dried blood spots (DBS) using the TREC assay developed for newborn screening by Dr. Jennifer Puck at NIH.
  - 3500 children who died <18 months of age
  - 500 control children
- Laboratory analysis will repeated in Dr. Puck's lab for DBS that appear deficient in TREC.
- Information from death certificates will be used to determine whether the cause of death was consistent with SCID.
- The results from this mortality cohort could reveal the extent to which SCID is an underlying cause of death in infants and very young children.

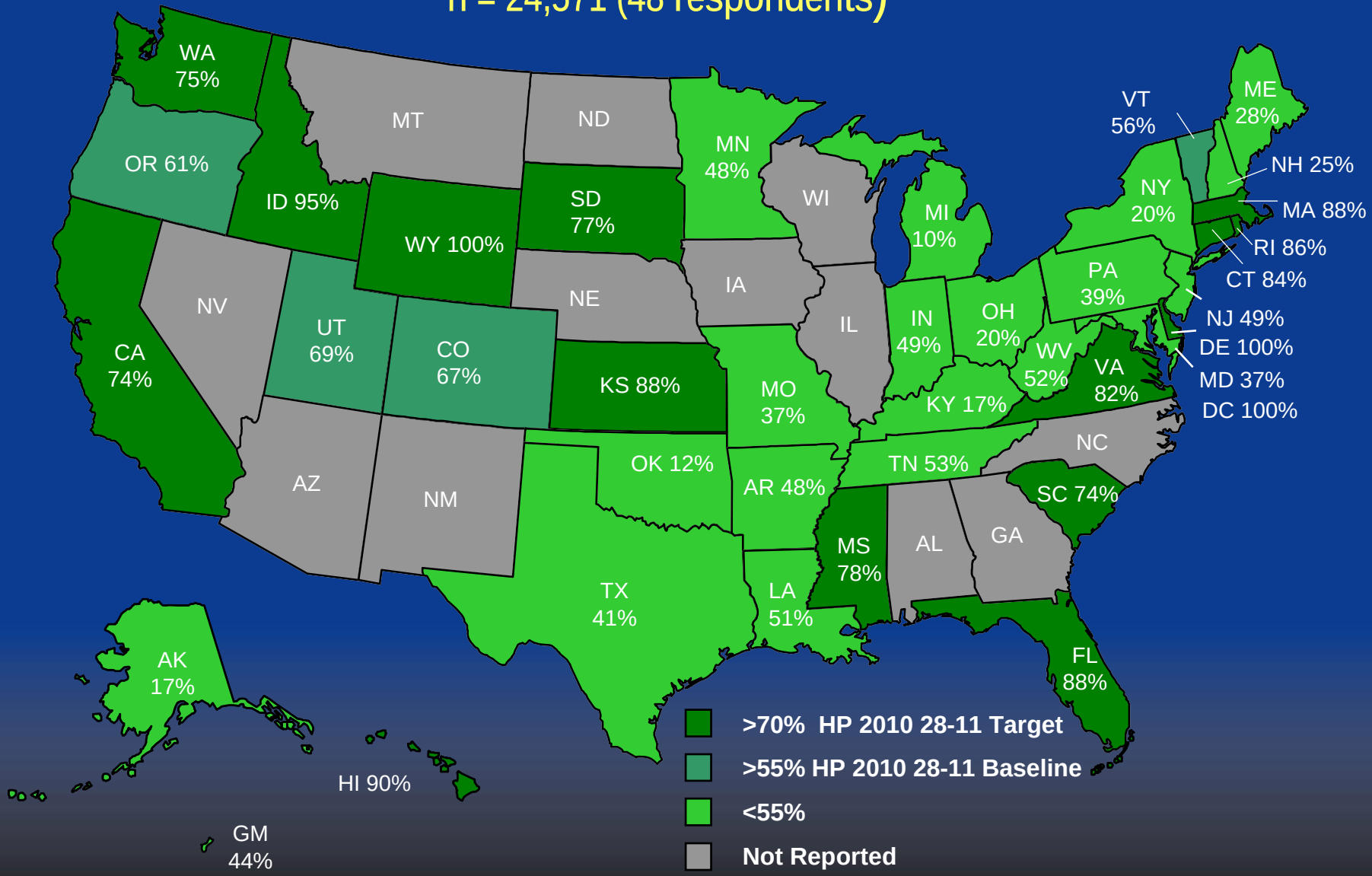
EHDI -- 91.7% Newborns Screened in 2004

n = 3,470,348 (48 respondents)



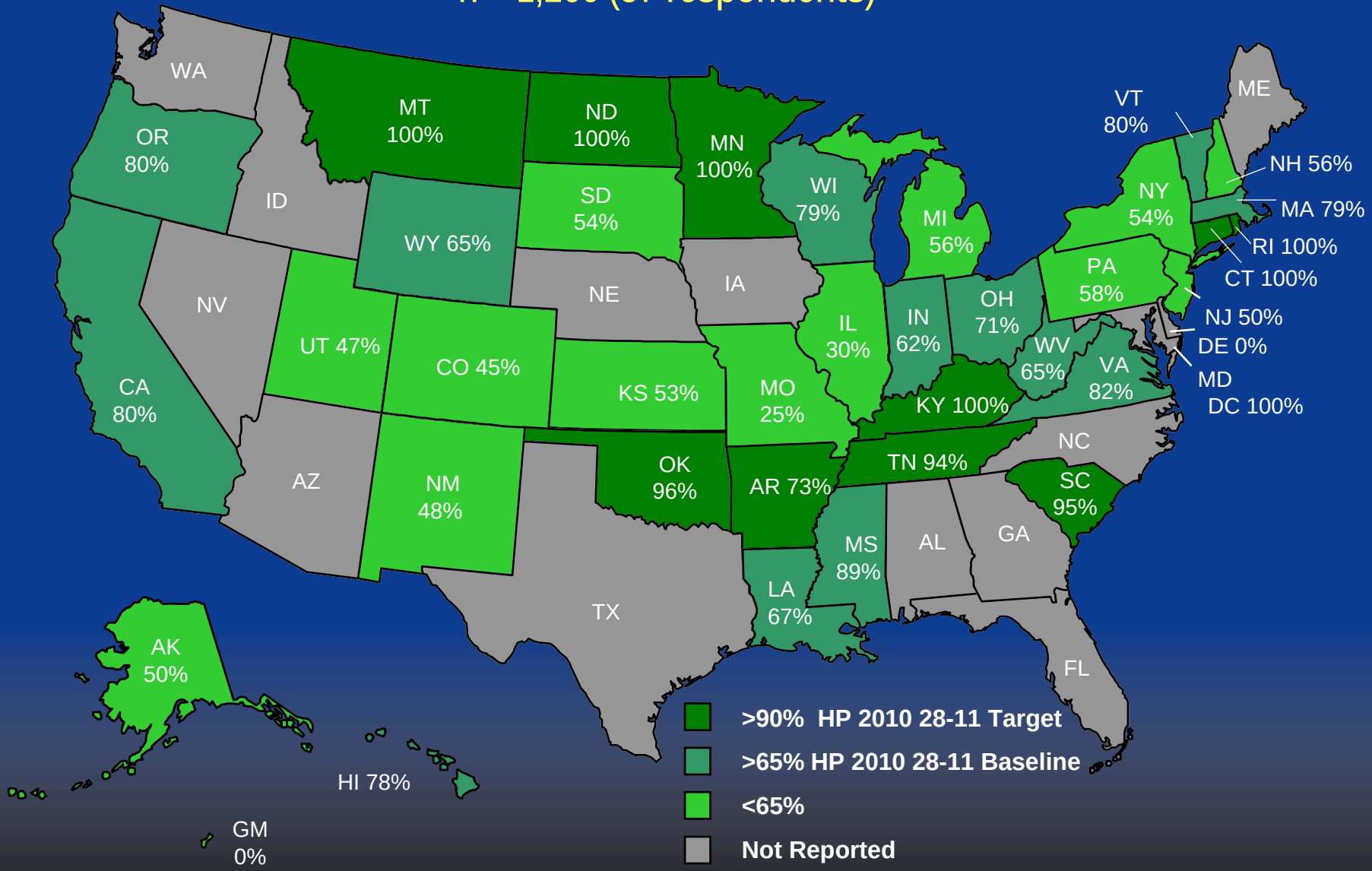
EHDI -- 48.0% Audiological Evaluation in 2004

n = 24,571 (48 respondents)



EHDI -- 69.9% Intervention by 6 months (2004)

n = 1,266 (37 respondents)



Public Health Practice in Laboratory Genetics

- **Current practices associated with DNA-based testing in Newborn Screening Programs within State Public Health Laboratories**
  - CDC in collaboration with the Wadsworth Center, New York Department of Health, the Association of Public Health Laboratories, and Mt. Sinai School of Medicine to develop a survey to assess the extent to which DNA testing is used in the reporting of results from newborn screening programs within State Public Health Laboratories. The results of this survey will be used to identify gaps in the implementation of DNA-based testing and useful for identifying efforts to help laboratories that are integrating DNA-based testing into their services.

- DNA-based [molecular] genetic laboratory test reporting to primary care physicians.
  - We are exploring a model successfully applied in other areas of laboratory medicine, termed synoptic reporting, to accomplish this. A key part of the process is to solicit clinician input to the development of the report. A clinician focus group will be held in association with the AAP meeting later this year. Model conditions, include CF and Fragile X

Recent publications by CDC staff on newborn screening, June 2006

Dried blood spot screening

1. Li L, Zhou Y, Bell CJ, Earley MC, Hannon WH. Development and characterization of dried blood spot materials for the measurement of immunoreactive trypsinogen (IRT). *Journal of Medical Screening*., 2006: In press.
2. Dantonio P, Meredith N, Earley M, Cordovado S, Callan WJ, Rollin D, Morris D, Vogt RF, Hannon H. Development of a screening system for detecting genetic risk markers of type 1 diabetes. *Diabetes Technologies and Therapie*s. 8, 2006: In press.
3. Grosse SD, Rosenfeld M, Devine OJ, Lai HJ, Farrell PM. Potential impact of newborn screening for cystic fibrosis on child survival: A systematic review and analysis. *Journal of Pediatrics*. 148, 2006: In press.
4. Olney RS, Moore CA, Ojodu JA, Lindegren ML, Hannon WH. Storage and use of residual dried blood spots from state newborn screening programs. *Journal of Pediatrics.* 148 (5), May 2006: 618-622.
5. Sweetman L, Millington DS, Therrell BL, Hannon WH, Popovich B, Watson MS, Mann MY, Lloyd-Puryear MA, Van Dyck PC. Naming and counting disorders (conditions) included in newborn screening panels. *Pediatrics*. 117 (5, supplement), May 2006: 308-314.
6. Dott M, Chase D, Fierrom M, Kalas TA, Hannon WH, Williams J, Rasmussen SA.Metabolic disorders detectable by tandem mass spectrometry and unexpected early childhood mortality: a population-based study. *American Journal of Medical Genetics A*.140 (8), April 15 2006: 837-842.
7. Grosse SD, Khoury MJ, Greene CL, Crider KS, Pollitt RJ. The epidemiology of medium chain acyl co-A dehydrogenase deficiency (MCADD): An update. *Genetics in Medicine*. 8 (4), April 2006: 205-212.
8. Grosse SD, Boyle CA, Kenneson A, Khoury MJ, Wilfond BS. From public health emergency to public health service: The implications of evolving criteria for newborn screening panels. *Pediatrics*. 117 (3), March 2006: 923-929.
9. Grosse SD, Olney R, Baily MA. Cost-effectiveness of universal versus selective screening for sickle cell disease in the United States and United Kingdom: A critique. *Applied Health Economics and Health Policy*. 4 (4), 2005: 239-247.
10. Baily MA, Becker Jr W, Hayes M, Clayton EW, Grosse S. Exploring options for newborn screening. *Journal of Law, Medicine, and Ethics.* 33 (4), special supplement, 2005: 46–48.
11. Skogstrand K, Thorsen P, Norgaard-Pedersen B, Schendel DE, Sorensen LC.Simultaneous measurement of 25 inflammatory markers and neurotrophins in neonatal dried blood spots by immunoassay with xMAP technology. *Clinical Chemistry*. 51 (10), October 2005: 1854-66.
13. Green DR, Grosse SD, Earley M, Mei J. Newborn screening for cystic fibrosis: a public health response. *Genomics and Population Health 2005*. Atlanta, GA: Centers for Disease Control, Office of Genomics and Disease Prevention, 2005, pp. 41–47.

Hearing screening

1. Eichwald J, Forsman I. Untangling early hearing detection and intervention. *Volta Voices*. 13 (3), May/June 2006: 16-19.
2. Brush CA, Kelly MM, Green D, Gaffney M, Kattwinkel J, French M. Meeting the challenge: using policy to improve children's health. *American Journal of Public Health*. 95 (11), November 2005: 1904-1909.
3. Hinman AR, Eichwald J, Linzer D, Saarlas KN. Integrating child health information systems. *American Journal of Public Health*. 95 (11), November 2005: 1923-1927.