Groups with Activities in Newborn Screening and Genetics:

- National Center for Birth Defects and Developmental Disabilities, CoCHP
- National Office of Public Health Genomics, CoCHP
- Division of Laboratory Systems, CCID
- Newborn Screening Quality Assurance Program, DLS, CCEHIP
- Newborn Screening Translational Research Initiative, DLS, CCEHIP
New Director, National Center on Birth Defects and Developmental Disabilities

• From Washington University School of Medicine, St. Louis

• Previous: Neurologist-in-Chief at St. Louis Children’s Hospital

• Recently worked with Missouri Department of Health as a Principal Investigator -- Autism and Developmental Disability Monitoring Network in Missouri

Edwin Trevathan, MD, MPH
Early Screening and Diagnosis of Duchenne Muscular Dystrophy (DMD)

• Pilot projects to test feasibility of newborn and infant screening for DMD
  – Laboratory:
    • Developed creatine kinase (CK) screening test on dried blood spots
    • Samples with elevated CK have dystrophin mutation analysis
    • Quality assurance program developed
  – Surveys developed to assess:
    • Informed consent process
    • Reasons why parents accept or decline screening
    • Families’ experiences with the screening programs
    • Healthcare providers’ attitudes and opinions
Feasibility and Prevalence Screen for Fragile X Syndrome (FXS)

- Automated, high-throughput screening test developed for dried blood spots
  - 100% sensitivity and specificity using 100 normal male samples and 20 full mutation male samples
  - Only identifies full mutations – no premutations
  - Will also identify sex chromosome abnormalities

- Determining the incidence of FXS using 70,000 to 100,000 blood spots leftover from NBS cards
  - De-identified cards from Georgia Newborn Screening Program
  - Prevalence of FXS and sex chromosome abnormalities
Assessing and Evaluating Historical Data Contained in the National Newborn Screening and Genetics Resource Center - Database

Vicki Stover Hertzberg, Ph.D.
Cherie James, MSPH
Emory University
Cynthia Hinton, Ph.D., MPH
Stuart Shapira, MD, Ph.D.
Centers for Disease Control and Prevention (CDC)
Lessons Learned from the Impact of Hurricane Katrina on Newborn Screening in Louisiana

Publication: Impact of hurricane Katrina on newborn screening in Louisiana, Pediatrics, October 2007

Emad A. Yanni, MD, MSc
EIS Officer, Pediatric Genetics Team
Newborn Screening and Molecular Biology Branch, DLS, CCEHIP

- New merged branch – 47 people
  - Acting Branch Chief, Harry Hannon, Ph.D.
  - Seeking person (announcement) for position -- early 2008
- NSQAP/Endocrine Disorders Laboratory and Metabolic and Hemoglobin Disorders Laboratory
- NSTRI/Immune Disorders Laboratory
- Diabetes and Molecular Risk Assessment Laboratory
- DNA-Banking and Genetic Studies Laboratory
Co-Sponsors

Newborn Screening and Molecular Biology Branch Programs

NSQAP

Newborn Screening Translational Research Initiative [ NSTRI ]

http://www.cdc.gov/labstandards/nsqap.htm
http://wwwwn.cdc.gov/nsqap
Newborn Screening Quality Assurance Program

“Gaining Confidence Through an External Quality Assurance Program for Dried-Blood Spot Testing”

• **Services provided:**
  - Filter paper evaluation
  - Reference materials
  - Quality control materials
  - Proficiency testing
  - Training, consultations, network resources

• **Partners**
  - Association of Public Health Laboratories
  - 70 domestic screening laboratories
  - Laboratories in 54 countries
  - 400 plus screening laboratories worldwide
478 Laboratories in 72 Countries - in at least one program of NSQAP
Analytes/Biomarkers included in NSQAP

- Biotinidase
- Thyroxine
- Thyroid-stimulating hormone
- 17 α-hydroxyprogesterone
- Total galactose
- Uridyltransferase (GALT)
- Citrulline
- Phenylalanine
- Leucine
- Valine
- Methionine
- Tyrosine
- Arginine
- Free carnitine (C0)
- Acetyl carnitine (C2)
- Propionyl carnitine (C3)
- Malonyl carnitine (C3DC)
- Isobutyryl carnitine (C4)
- Isovaleryl carnitine (C5)
- Glutaryl carnitine (C5DC)
- Hexanoyl carnitine (C6)
- Octanoyl carnitine (C8)
- Decanoyl carnitine (C10)
- Decenoyl carnitine (C10-1)
- Myristoyl carnitine (C14)
- Tetradecenoyl carnitine (C14-1)
- Palmitoyl carnitine (C16)
- Stearoyl carnitine (C18)
- Immunoreactive trypsinogen/Δ508 mutations
- CF DNA Mutation Panel
- Hemoglobinopathies and SS, SC, SD, SE mutations
- Diabetes Type 1 risk mutations
- Toxoplasmosis: IgG, IgM
- HIV type 1 antibodies
- Creatine kinase (DMD)
- Androstenedione/cortisol/11-deoxy

New analytes
CF Mutation Detection PT Program

- Blood is collected from adult CF patients with known mutations
- Allows testing of less common mutations and demonstrates limitations of assays.
- Collaboration between CDC, University of Wisconsin School of Medicine and Public Health, Johns Hopkins Medical Center, and Case Western University.
LSD Projects

- Objective: To facilitate newborn screening for lysosomal storage disorders (LSDs) in public health laboratories, in conjunction with Genzyme Corporation

- Current MS multiplex assay allows screening for *Pompe, Fabry, Krabbe, Niemann-Pick* and *Gaucher* diseases
CLSI Standard

“Blood Collection on Filter Paper for Neonatal Screening Programs, Approved Standard”

New Edition  LA4-A5 - 2007

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10 Million Served

Thanks