Current Practices and Expansion of Newborn Screening

R. Rodney Howell, M.D.

Professor of Pediatrics, Miller School of Medicine, University of Miami

Chair, the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children

U.S. Department of Health and Human Services
Centers for Disease Control and Prevention

Newborn Screening
Historical Perspective

- **Beginning**
  - Keen interest in inborn errors of metabolism
  - Term introduced by Garrod in 1908
- **Initial focus**
  - Conditions that adversely affect the central nervous system
- **Expansion**
  - Immune and cardiac systems
  - Influenced by
    - Available technology
    - Better understanding of conditions
    - New diagnostic technologies and treatments

Newborn Screening for Genetic Diseases in the United States

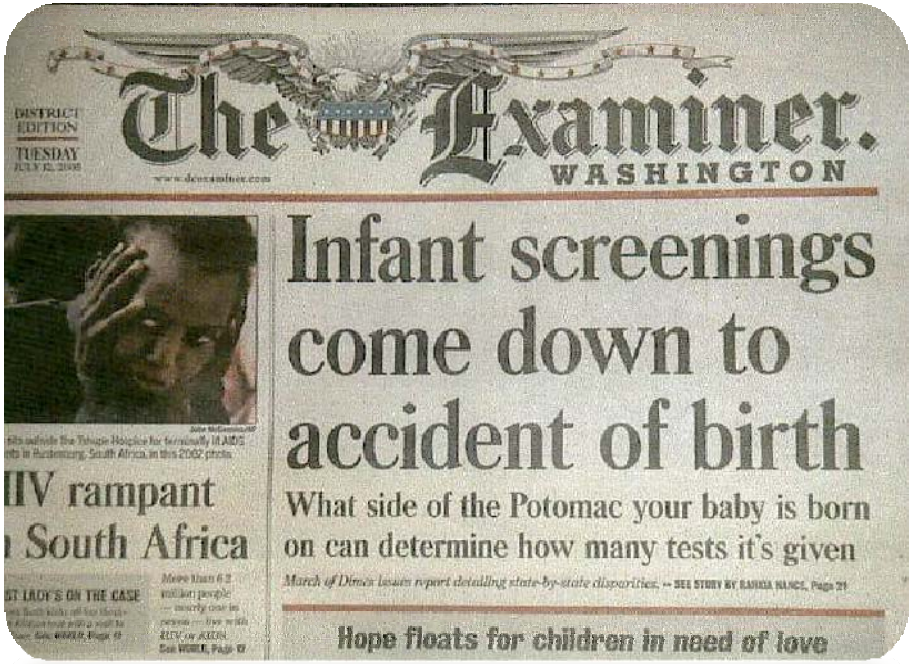
- **Routine newborn screening**
  - Began in 1960s; now carried out in all 50 states
  - State-sponsored public health programs; most successful
- **Initial testing targets**
  - Phenylketonuria and similar conditions
  - Simple, reliable screening tests and proven treatment efficacy
- **Expansion of targets**
  - State-by-state basis
- **Challenge**
  - Extraordinary variation from state to state
  - Little systematic evaluation of the rationale for and/or the outcomes of screening

Newborn Screening for Genetic Diseases in the United States

Over 4 million infants are screened each year

Newborn screening is by far the most commonly performed testing for genetic diseases in the United States

The Extraordinary State-to-State Variation in Newborn Screening Caused Great Concern



DISTRICT EDITION
TUESDAY

The Examiner.
WASHINGTON

IV rampant
South Africa

Infant screenings come down to accident of birth

What side of the Potomac your baby is born on can determine how many tests it's given

March of Dimes issues report detailing state-by-state disparities. -- SEE STORY BY SABRINA NANCE, Page 24

Hope floats for children in need of love

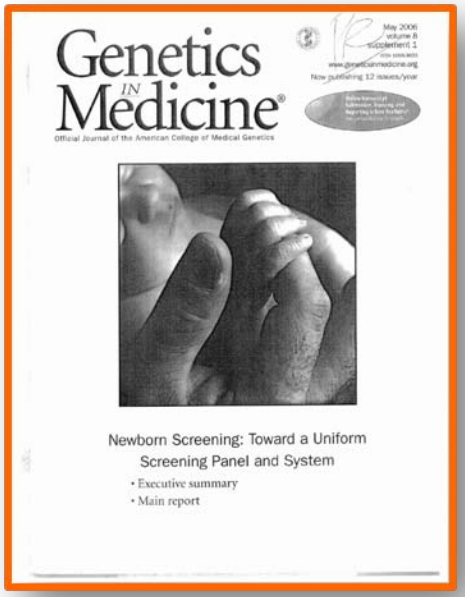
Standardization of Newborn Screening in the United States

- **In 2001, Maternal and Child Health Bureau/HRSA charged American College of Medical Genetics**
  - To evaluate the scientific and medical information related to screening for specific conditions
  - To make recommendations based on this evidence
- **Expert group convened in December 2002**
  - >70 physicians, scientists, consumers, state laboratorians, lawyers, ethicists, and others
  - Results reviewed by an independent newborn screening external review group
  - Newborn Screening: Toward a Uniform Screening Panel and System (report published in 2006)

http://www.acmg.net/resources/policies/NBS/NBS_Exec_Sum.pdf

Selection Criteria of the Uniform Screening Panel

- Incidence of conditions
- Identifiable at birth
- Burden of disease
- Mortality prevention



- Availability of test
- Test characteristics
- Diagnostic confirmation

- Availability of treatment
- Cost of treatment
- Efficacy of treatment
- Benefits of early intervention
- Benefits of early identification
- Acute management
- Simplicity of therapy

http://www.acmg.net/resources/policies/NBS/NBS_Exec_Sum.pdf

Uniform Screening Panel
29 Primary (Core) Conditions

- All result in serious medical complications (e.g., developmental delay) and/or death if not recognized early
- All children with these conditions benefit from early diagnosis and treatment

Uniform Screening Panel
29 Primary (Core) Conditions

- **20 are disorders of amino acids, fatty acids, and organic acids**
  - Detected by a sophisticated laboratory technique (tandem-mass spectroscopy)
- **3 are hemoglobinopathies (types of sickle cell disease)**
- **6 other conditions**
  - Biotinidase deficiency
  - Congenital adrenal hyperplasia
  - Cystic fibrosis
  - Congenital hypothyroidism
  - Galactosemia
  - Hearing disorders

Uniform Screening Panel
25 Secondary Targets

- **Target compounds identified by the same methods as primary targets**
- **Compounds at times present in abnormal amounts**
  - Instances when these compounds are present in abnormal amounts are not completely understood
  - Proper identification of conditions on the core panel requires that these compounds be identified and measured
  - It is recommended that these secondary targets be reported to improve the understanding of their significance

Burden of the Core Panel Conditions in the United States

- **All conditions are rare**
- **Estimated annual numbers (most common)**
  - Hearing loss: 5,064
  - Primary congenital hypothyroidism: 2,156
  - Sickle cell disease: 1,775
  - Cystic fibrosis: 1,248
  - Medium-chain acyl-CoA dehydrogenase deficiency: 239
- **About 12,500 infants are diagnosed with the core conditions each year with the current newborn screening panel**

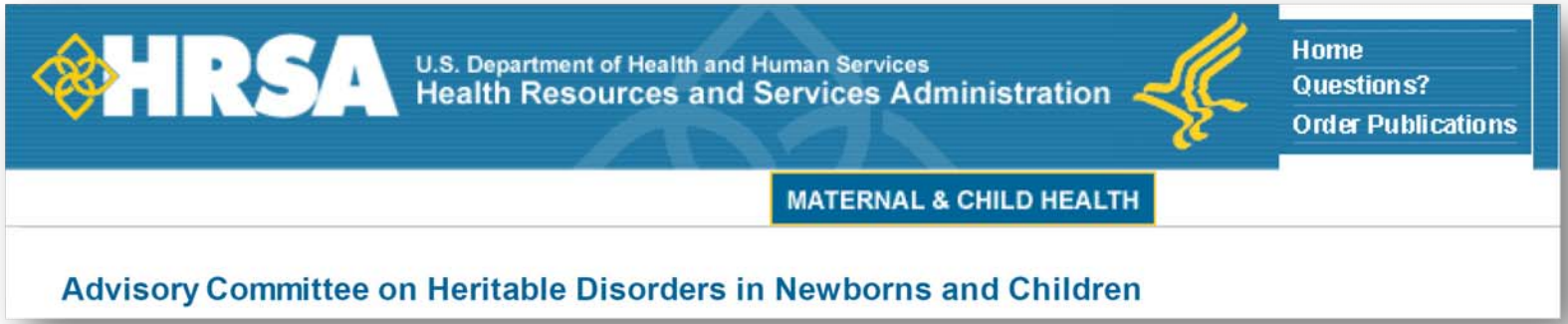
2009 Annual Data http://www.cdc.gov/ncbddd/hearingloss/ehdi-data2009.html
Impact of Expanded Newborn Screening — United States, 2006 http://www.cdc.gov/mmwr/preview/mmwrhtml/mm5737a2.htm
2011 Annual Report to Congress http://www.hrsa.gov/heritabledisorderscommittee/reports/SACHDNC2011ReportCongress.pdf

Burden of the Core Panel Conditions in the United States

- **Untreated persons suffer enormous burdens**
  - Persons with phenylketonuria have relatively normal lifespan
    - Untreated: Profound intellectual disability with IQ frequently below 20
    - Identified and treated from birth: Normal IQ
  - Persons with medium-chain acyl-CoA dehydrogenase deficiency (the most common disorder of fatty acid oxidation) are at substantial risk for sudden death

Policies and Guidelines: Authorizing Legislation

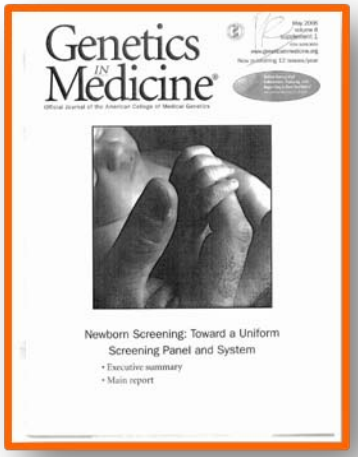
- **Title XXVI of the Children's Health Act of 2000 enacts 3 sections of the Public Health Service Act**
  - Established the Advisory Committee on Heritable Disorders in Newborns and Children (1st meeting in 2004)
  - Broad charge, but efforts to date focused on newborn screening



http://www.hrsa.gov/heritabledisorderscommittee

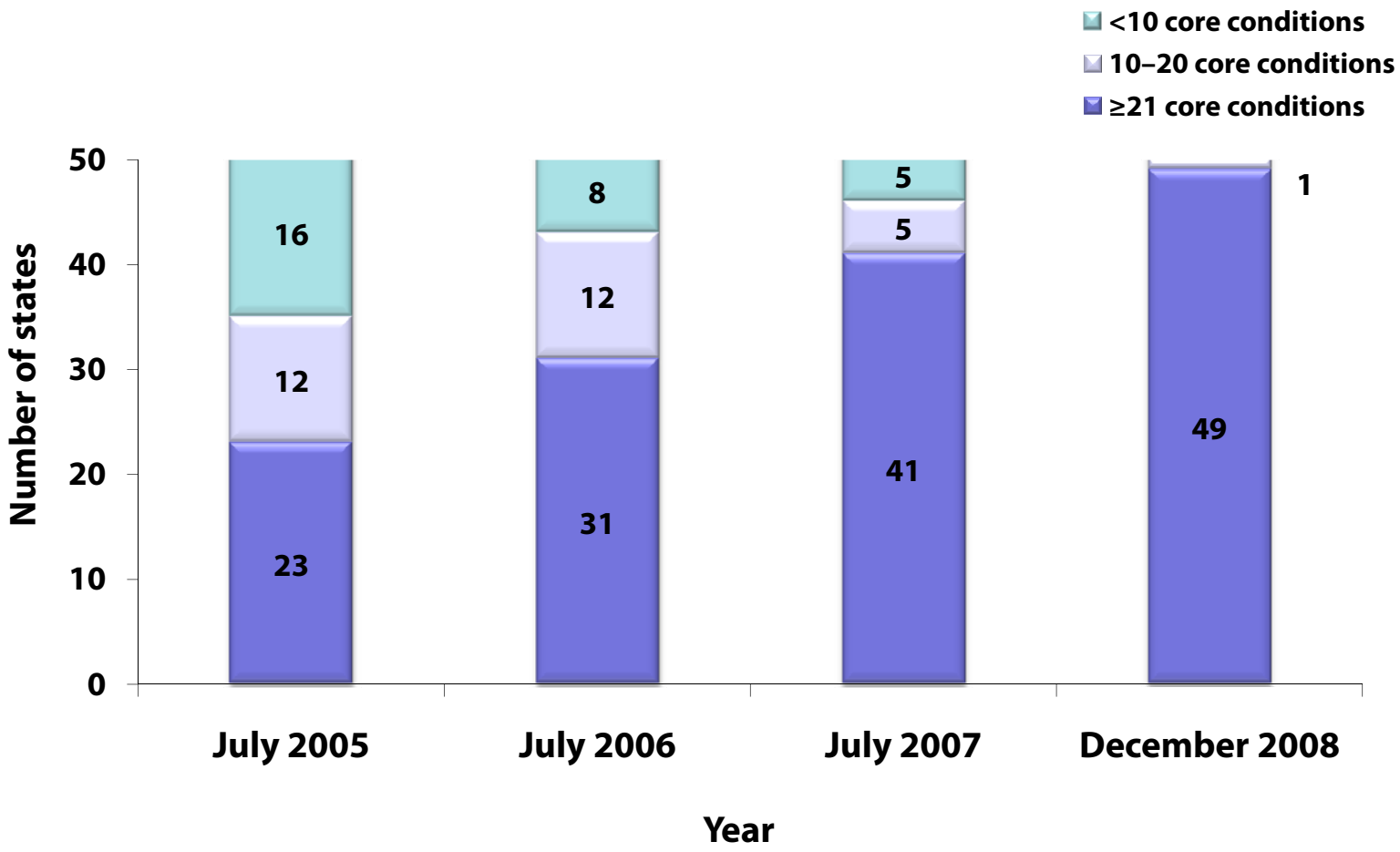
Secretary's Advisory Committee for Heritable Disorders in Newborns and Children

- **Focus on the report of the American College of Medical Genetics**
  - Unanimously accepted the report
  - Made recommendation to the HHS Secretary to adopt and implement the report
- **In time, the HHS Secretary**
  - Accepted the committee recommendation
  - Designated this Uniform Screening Panel as a national standard for newborn screening programs



HHS, Department of Health and Human Services
http://www.acmg.net/resources/policies/NBS/NBS_Exec_Sum.pdf

Newborn Screening Tests in the United States



March of Dimes. Data reported from National Newborn Screening and Genetics Resource Center
http://genes-r-us.uthscsa.edu

Nomination Form for Inclusion of Conditions into the Recommended Uniform Screening Panel

NEWBORN SCREENING UNIFORM PANEL

NOMINATION FORM FOR PROPOSED CONDITION

Name of Proponent	*(Organization, if relevant)*	Date	
Condition			
Type of Disorder			
Screening Method			
Treatment strategy			

CONDITION	Commen	OMIM or other names for disorder	
Incidence	(Determin	nces below (p.2)	
Timing of clinical onset	(Relevance of the timing of newborn screening to onset of clinical manifestations)		
Severity of disease	(Morbidity, disability, mortality, what spectrum of severity)		

Condition

TEST	Comment
Screening test(s) to be used	(High volume method, platform)
Modality of screening	(Dried blood sp
Clinical validation	(Location, dura r clinical validation)
Laboratory performance metrics	(Sensitivity, sp tive rate)
Confirmatory testing	(Reliability, availability)
Risks	(False positives, carrier detection, invasiveness of method, other. Detection or suggestion of other disorders)

Screening test

NOMINATION OF CONDITION (page 2)

TREATMENT	Comment
Modality	(Drug(s), diet, replacement therapy, transplant, other)
Urgency	(How soon after birth treatment needs to be initiated to be effective)
Efficacy (Benefits)	(Extent of prevention of mortality, morbidity, disability. Treatment limitations, such as difficulty with acceptance o
Availability	(Any limits o
Risks	(Potential medical or other ill effects from treatment)

Treatment

KEY REFERENCES (Specific citations – limit to 15)

1	
2	
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References

Submission Check list	
	Cover letter by proponent
	Nomination form
	Copy of references listed on this form
	Formal conflict of interest statement by proponent

Submit Nominations to:
Michele A. Lloyd-Puryear, M.D., Ph.D.
Chief, Genetics Services Branch
Division of Services for Children with Special Health Needs
Maternal and Child Health Bureau
5600 Fishers Lane, Room 18-A-19
Rockville, MD 20857
301-443-8604 –fax 301-443-1080 - phone

Contact information (proponent)

ftp://ftp.hrsa.gov/mchb/genetics/NominationForm.doc

Nomination Process for Inclusion of Conditions into the Recommended Uniform Screening Panel

- **Rigorous process in place for review of nominations**
- **9 nominations submitted and reviewed since 2007**
  - 6 conditions sent forward for external evidence review
  - 4 have referred back to nominators for additional studies
  - 2 recommended for addition to the Panel
    - Severe Combined Immunodeficiency (SCID) has been accepted by HHS Secretary Sebelius
    - Critical Cyanotic Congenital Heart Disease (CCCHD) is under review

Major Challenges to Newborn Screening and the Way Forward

- **Serious shortage of clinical experts in the area of inborn errors of metabolism spans most of the primary conditions detected by newborn screening**
  - Example: The American College of Medical Genetics has identified funding for fellowships in biochemical genetics
- **Public health laboratories are stretched financially at a time when important new discoveries must be brought to the public**
  - Example: Detection of multiple disorders using single tests, automation, and other cost-saving technologies

Major Challenges to Newborn Screening and the Way Forward

- **Lack of public education and understanding about the value newborn screening**
  - Example: Genetic Alliance's Newborn Screening Clearinghouse will provide a great opportunity for public education
- **Retention and use of residual dried blood spots**
  - Example: Kemper et al. Committee report: Considerations and recommendations for national guidance regarding the retention and use of residual dried blood spot specimens after newborn screening
    - Extensive professional and public input
    - Expected to provide direction for states in their own planning

Kemper, AR et al. Genetics in Medicine 2011;13:621

Family Experiences with Disorders and Screening

Sharon F. Terry, MA
President and Chief Executive Officer
Genetic Alliance

http://www.geneticalliance.org

U.S. Department of Health and Human Services
Centers for Disease Control and Prevention

Newborn Screening: What's at Stake

- **Virginia mother Jana Monaco gave birth to her 3rd child, Stephen in 1997 at a Virginia hospital**
  - Virginia only screened for 9 conditions at birth
  - Stephen had 3 years of a relatively normal, healthy life
  - On May 30, 2001, Stephen went into metabolic crisis, resulting in severe disabilities
- **Stephen was diagnosed with Isovaleric Acidemia (IVA)**
  - IVA is treated with a special diet and medication; if begun soon after birth, affected children live relatively normal, healthy, long lives
  - Had Stephen been born a few hours south, in North Carolina, the condition would have been detected at birth
    - North Carolina screened for 36 conditions, including IVA

Lessons from Jana Monaco's Story

Genetic Alliance Innovator Series – Jana Monaco
http://www.geneticalliance.org/JanaMonaco

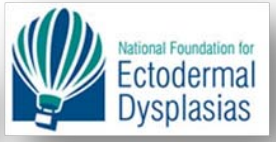
The Diagnostic Odyssey

- **At birth: 10 fingers, 10 toes**
- **"We had a gut feeling that something wasn't right."**
  - Communication with the providers: "Sick babies don't smile"
  - The family endures anxiety, fear, and uncertainty
  - Trips to multiple hospitals and specialists
  - Parents watch as their child undergoes countless procedures
- **In the end, the diagnosis is often made too late**
- **The most painful part: It could have been prevented**

SCID: Heather Smith's Story, October 29, 2010
Immune Deficiency Foundation (PrimaryImmune)
http://www.youtube.com/user/PrimaryImmune

Family Advocates Help Shape the System

- **"I became a newborn screening advocate the night my daughter died"**
  - Historically, the introduction of newborn screening at the state level depended on advocacy by parents of children with disorders
  - Disease advocacy organizations
    - Raise awareness of conditions and screening
    - Advance treatment
    - Newborn Screening Saves Lives Act (2008)
    - Children's Health Act (2000)



The organization logos included above contribute to Disease InfoSearch.org, online repository of condition-specific support organizations and resources hosted by Genetic Alliance http://geneticalliance.org/diseaseinfosearch

Other Key Community Partners

- **National advocacy/resource organizations**
- **Federal and state-funded program, examples**
  - Title V funded programs provide direct services and support for families of children with special health needs
    - Family to Family Health Information Centers; Medicaid
  - Early Hearing Detection and Intervention programs
    - Identifies infants and children with hearing loss
    - Promotes timely follow-up testing and services or interventions

http://www.geneticalliance.org
http://www.savebabies.org
http://genes-r-us.uthscsa.edu/
http://mchb.hrsa.gov/programs/
http://www.cdc.gov/ncbddd/hearingloss/ehdi-programs.html

Consumers as Collaborators

- **Genetic Alliance represents the consumer perspective on the HHS Secretary's Advisory Committee for Heritable Disorders in Newborns and Children**
  - Voice for spectrum of family experiences
  - Technical assistance for public comments: Tips, best practices
  - Technical assistance for condition nomination
  - Collaboration with advocacy organizations focused on screening implementation (e.g., Immune Deficiency Foundation)

Baby's First Test
The Newborn Screening Clearinghouse

- **In 2009, Genetic Alliance and partners were awarded a cooperative agreement to create the Newborn Screening Clearinghouse**
  - One-stop-shop of newborn screening information to raise knowledge and awareness of programs and process
  - Provides
    - Comfortable and confident web experience
    - Condition and state-specific information
    - Diverse materials available (brochures, videos, blogs)
  - Anticipated launch date: September 1, 2011

http://nbsclearinghouse.org

It is NOT Enough to Screen

- **Follow-up and management are central to keeping the promise of newborn screening**
- **Long-term follow-up activities within public health programs**
  - Lack coordination
  - Have been of low priority for funding
- **Challenges families face include**
  - Understanding intervention options
  - Connecting with new specialists or care providers
  - Interacting with public or private insurers
  - Communicating with family members, teachers, etc
  - Transitioning from pediatric to adult care services

Kemper, AR et al. Genetics in Medicine 2008;10:259-261
http://ftp.hrsa.gov/mchb/genetics/reports/LongTermFollowUpAfterNewbornScreening.pdf

Consumer Task Force on Newborn Screening Statement on Long-term Follow-up Care

- **The Consumer Task Force on Newborn Screening**
  - 9 consumers with a range of experiences in newborn screening
  - In 2009, provided public comments on long-term follow-up at the 19th meeting of the HHS Secretary's Advisory Committee
- **Key messages**
  - Balance is needed between parents responsibility to be "advocates" and their need to cope with the unfolding medical situation
  - Providers are responsible for helping families interpret information from a variety of sources in a non-directive, non-judgmental way
  - Increased federal investment in long-term follow-up care is needed

Williams, A. 19th Meeting." September 24-25, 2009, Washington, DC. http://geneticalliance.org/nbs.achdnc

Summary

- **Newborn screening prevents diagnostic odysseys**
  - Expensive for the medical system and painful for families
- **The variability in state screening panels has a significant negative impact on families**
- **Family advocates shape state screening panels and are integral to raising awareness and funds**
- **Consumers must be engaged in policy and program development**
- **It is not enough to simply screen to keep the promise of newborn screening – we must fol**

Laboratory Quality Assurance for Newborn Screening Tests

Carla D. Cuthbert, PhD

Chief, Newborn Screening and Molecular Biology Branch

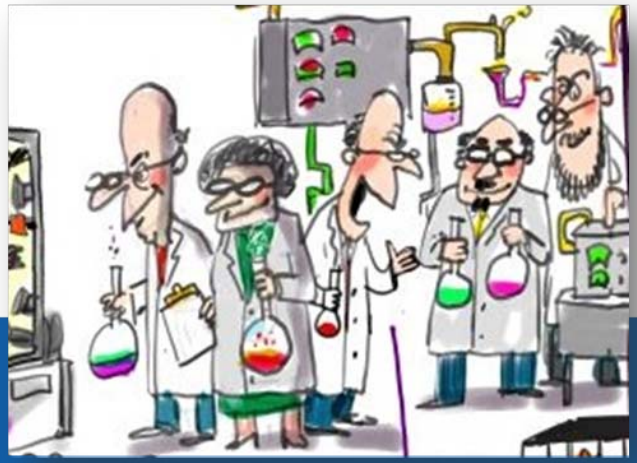
Division of Laboratory Sciences, National Center for Environmental Health

Centers for Disease Control and Prevention

U.S. Department of Health and Human Services
Centers for Disease Control and Prevention

The Laboratory is a Vital Component of the Newborn Screening System

- Conducts high-quality screening covering all mandated tests in respective states
- Communicates with other components of the Newborn Screening System
- Plays a key role in translational research by defining and designing new laboratory tests
- Committed to continuous quality improvement



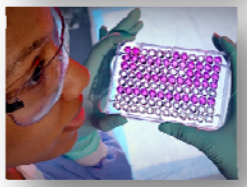
Laboratories Performing Newborn Screening in the United States

- **Most testing is performed in state public health laboratories**
- **Different testing models exist for different states**
  - Regionalization
    - Oregon tests for 5 other states and for birthing facilities in the Navajo Nation, Guam, Saipan, and Kwajalein
  - Collaboration with contract laboratories
    - California uses 7 contract laboratories
    - Perkin Elmer Genetics screens for 3 states and DC
    - Minnesota uses the Mayo Clinic for some assays
- **State laboratories are under CLIA regulatory oversight**

CLIA, Clinical Laboratory Improvement Amendments

Dried Blood Spot Measurements Use a Broad Range of Technologies

- Visible and fluorescence enzymatic assays
- Tandem mass spectrometry
- Electrophoresis and high-performance liquid chromatography
- Immunochemical and molecular assays
- Real-time polymerase chain reaction



Challenge: Identify all affected newborns babies while minimizing false positive tests

Assurance of Quality in Newborn Screening

- **Developing and improving screening tests**
- **Administering the Newborn Screening Quality Assurance Program (NSQAP)**
  - All U.S. laboratories that conduct newborn screening
  - > 450 international laboratories in 67 countries

All testing, excluding hearing screening, is done from dried-blood spots (DBS)
Blood is taken via a heal prick

NSQAP: Only program that addresses dried-blood spot measurements for all conditions

Essentials of the Newborn Screening Quality Assurance Program (NSQAP)

- **Proficiency testing**
  - Measurement of reference samples
  - Administered every 3 months for each condition
  - Report of assessment is provided on secure website
  - Timely evaluation of causes of false negative results
- **Reference materials**
  - Weekly verification of accuracy of testing in participating laboratories
- **Training and consultation**

Preparation of reference material and laboratory performance quality reports

NSQAP Website, http://www.cdc.gov/labstandards/nsqap.html

Quality Assurance Program in 2010

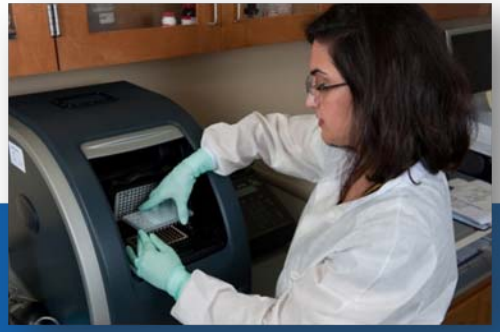
- **100%** participation of state public health laboratories
- **456** laboratories in **67** foreign countries participated
- **>700,000** dried blood spots produced and distributed
- **17** summary quality assurance reports generated
- Collaborated with APHL in support of quality issues
  - Conferences (**>400** attendees)
  - National meetings (**~200** participants)
  - Technical workshops

APHL ASSOCIATION OF PUBLIC HEALTH LABORATORIES

Introduction of New Screening Test Severe Combined Immune Deficiency (SCID)

- **Funded and administered newborn screening pilot studies**
  - First 2 states: Wisconsin and Massachusetts
  - Navajo population through the Univ. of California at San Francisco
- **Developed a novel molecular assay**
- **Serves as the only provider of national reference materials for SCID testing in dried blood spots**
- **Established a proficiency testing program**
- **Provides training and technical support**

Molecular assay for SCID by *in situ* Real-time polymerase chain reaction



SCID Screening in the United States

- **As of 2010**
  - 1 million newborns have been screened
  - 19 babies identified with SCID
  - 45 babies identified with other severe immunocompromising conditions
  - SCID incidence appears higher than previously reported (as high as 1:30,000)

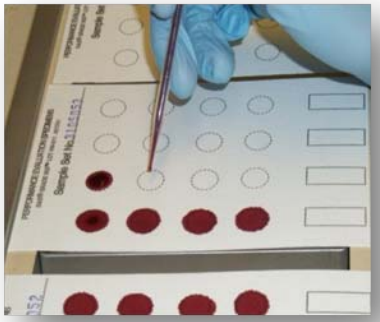
Meet Dawson!

- Appeared healthy at birth
- Failed the newborn screening test for SCID
- Received a bone marrow transplant
- Now … a thriving 3 year old

SCID, Severe Combined Immune Deficiency

Future Technical Laboratory Challenges and Considerations for Newborn Screening

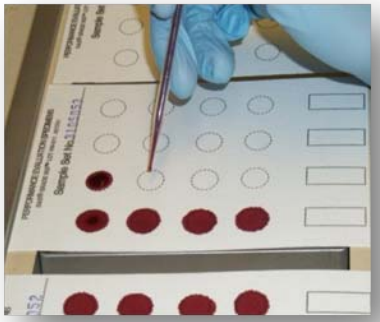
- Detection of new conditions
- Detection of multiple conditions using single tests
- Expansion of automation to reduce assay cost
- Reducing false positive results for select conditions by applying biochemical and molecular tests
- Expansion of molecular testing to other disorders



Other Challenges that Impact the Newborn Screening Laboratory

- Funding and restricted budgets
- Keeping state leaders and the public effectively informed on important newborn screening topics

Challenges present opportunities for innovation



Newborn Screening
Health Impact and Return on Investment

Scott D Grosse, PhD

Associate Director, Health Services Research and Evaluation

Division of Blood Disorders

National Center on Birth Defects and Developmental Disabilities

Centers for Disease Control and Prevention

U.S. Department of Health and Human Services
Centers for Disease Control and Prevention

Challenges in Assessing Health Impact and Return on Investment in Newborn Screening

- **Health impact**
  - Diverse conditions with varying outcomes, i.e., death, disability
  - Outcomes are hard to study in unscreened cohorts
    - Representative children are difficult to identify
  - Outcomes in screened cohorts also reflect improvements in care
    - Sickle cell disease and use of pneumococcal vaccines
- **Return on investment**
  - Preventing disability can save money
  - Preventing death, while valuable, may not reduce costs
  - Overall, newborn screening is probably cost saving – saves more money than it costs

Grosse SD. In: Khoury M et al, (eds), *Human Genome Epidemiology, 2nd Ed*. New York: Oxford U Press. 2009, 517–32
Grosse SD. In: Ungar W (ed). *Economic Evaluation in Child Health*, Oxford U Press. 2009, 113–32

Congenital Hypothyroidism Burden and Health Impact

- **Burden of congenital hypothyroidism**
  - 1 in 2,000 newborns diagnosed–about 2,000 each year
- **Health impact**
  - 20–30%: Percentage of those diagnosed with congenital hypothyroidism who had an IQ <70 before screening took effect
  - Shift to the left in IQ distribution after screening took effect
    - 20–25 points in children with clinical congenital hypothyroidism
    - 7–8 points in children with subclinical congenital hypothyroidism

Grosse SD, Van Vliet G. Arch Dis Child. 2011; 96(4):374–379

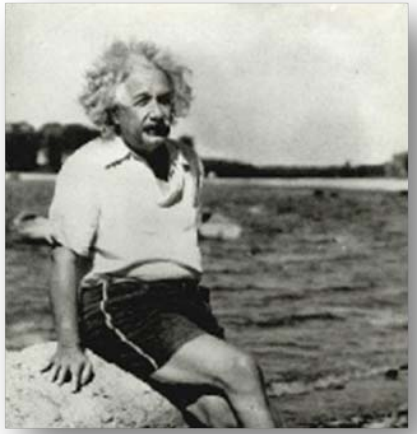
Newborn Screening is a Bargain

- **Cost of newborn dried-blood spot screening is spread over many disorders**
  - 2003 GAO report: $120 M, or $30 per infant
  - Estimate for congenital hypothyroidism alone
    - ~$5 per infant
    - Total $20 M per year for 4 million infants

Return on Investment in Newborn Screening for Congenital Hypothyroidism

Health impact

- 160 cases of intellectual disability avoided each year
- 470 other children with clinical congenital hypothyroidism
  - Total gain of 10,600 IQ points
- 540 children with mild permanent congenital hypothyroidism
  - Total gain of 4,300 IQ points
- Total health impact
  - Prevention of 160 cases of disability
  - Gain of 14,900 IQ points among 1,010 children with no disability



Grosse, SD and Van Vliet G. Arch Dis Child. 2011; 96(4):374–379

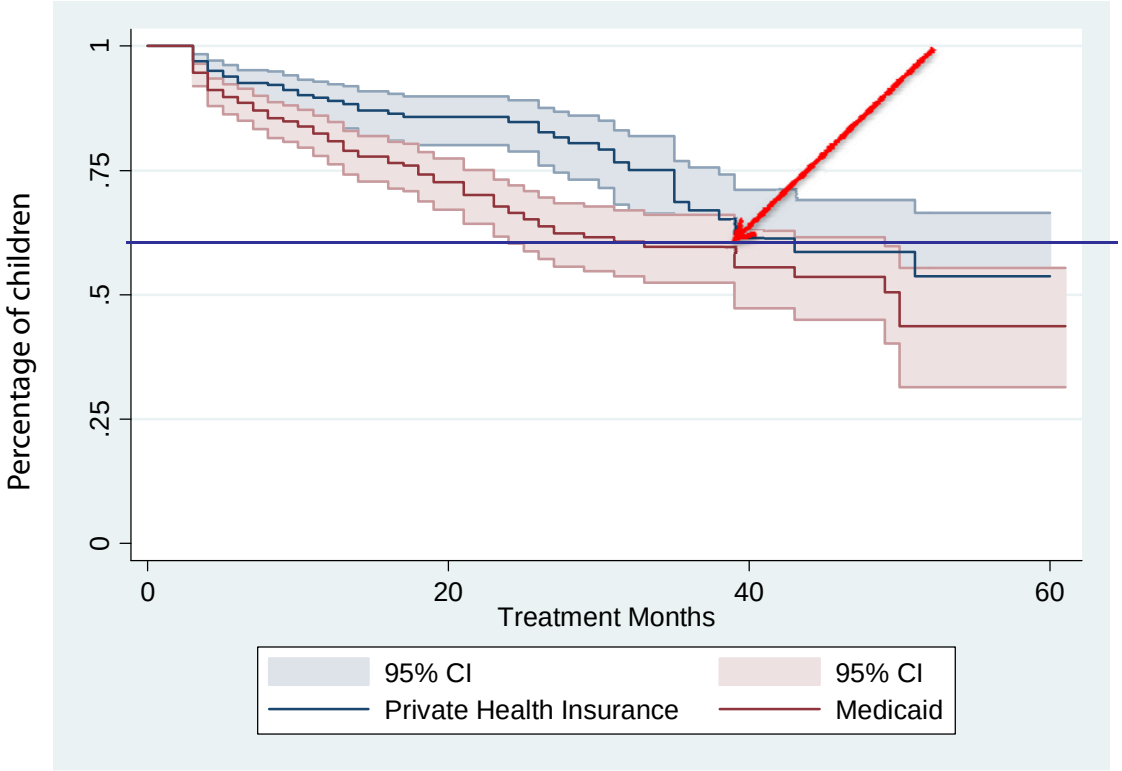
Return on Investment in Newborn Screening for Congenital Hypothyroidism

- **Economic benefits**
  - Lifetime direct and indirect cost of $1.3 M for intellectual disability
    - Avoided cost of $195 M from 160 avoided cases of disability
  - Each IQ point is worth $13,000 in increased lifetime earnings
    - $196 M per year from gain of 14,900 IQ points
  - Total $391 M per year
- **Economic benefit is almost 20 times larger than cost of newborn screening**
  - $400 M vs. $20 M

CDC. MMWR 2004; 53(3):57–59
Grosse SD. AERE Newsletter. 2007; 27(2):17-21Grosse, SD et al. Med Care. 2009; 47(7 Suppl 1):S94–S103

Need for Long-term Follow-up for Congenital Hypothyroidism: Discontinuation of Treatment

Continuation of hormone treatment for children with congenital hypothyroidism by insurance type (private or public)



Kemper, AR et al. BMC Pediatrics 2010; 10:9

Congenital Hearing Loss and Newborn Hearing Screening

- **Congenital hearing loss**
  - 2–3 in 1,000 newborns
  - >5,000 infants diagnosed each year through newborn screening
- **Health Impact**
  - Language development, school achievement, employment
  - United Kingdom of school-age children with bilateral moderate or greater hearing loss who received screening
    - Improved language, reading, and communication scores
    - Reduction in education costs (22% gross, 36% net)

Kennedy, CR et al. N Engl J Med. 2006;354:2131-41
McCann, DC et al. Arch Dis Child. 2009;94:293-7
Schroeder, L et al. Pediatrics 2006; 117:1101–12

Early Hearing Detection and Intervention (EHDI)

- **1-3-6 national goals**
  - All infants are screened at <1 month of age
  - All screen-positive infants have diagnostic evaluation at <3 months
  - All infants with abnormal hearing receive early intervention services at <6 months
- **EHDI components**
  - Screening
  - State EHDI programs
  - Medical "home"
  - Intervention therapies include
    - Training in communication options
    - Amplification (hearing aids) or cochlear implants

EHDI, Early hearing detection and intervention

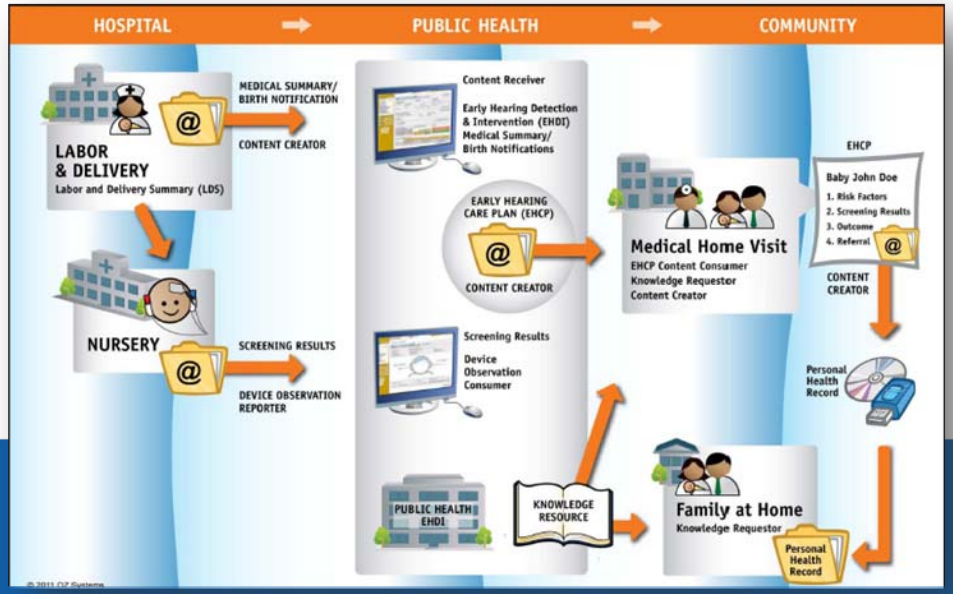
Return on Investment in Early Hearing Detection and Intervention (EHDI)

- **Cost of newborn hearing screening**
  - $40 per infant for screening and diagnostic testing
  - $10 per infant for sustaining the EHDI infrastructure
  - Total: $200 M per year for 4 million infants
- **Economic benefits**
  - Projected savings in direct costs in the United States
    - $115,600 increase in lifetime cost of education for children with permanent bilateral hearing loss
    - Savings per infant is $44,200 with 36% reduction
    - $200 M per year for 5,000 infants – break even in direct cost
  - Gains in employment and earnings
    - Could be twice the savings from education costs

Grosse SD. Volta Voices 2007;14(6):38–40
CDC. MMWR 2004; 53(3):57–59

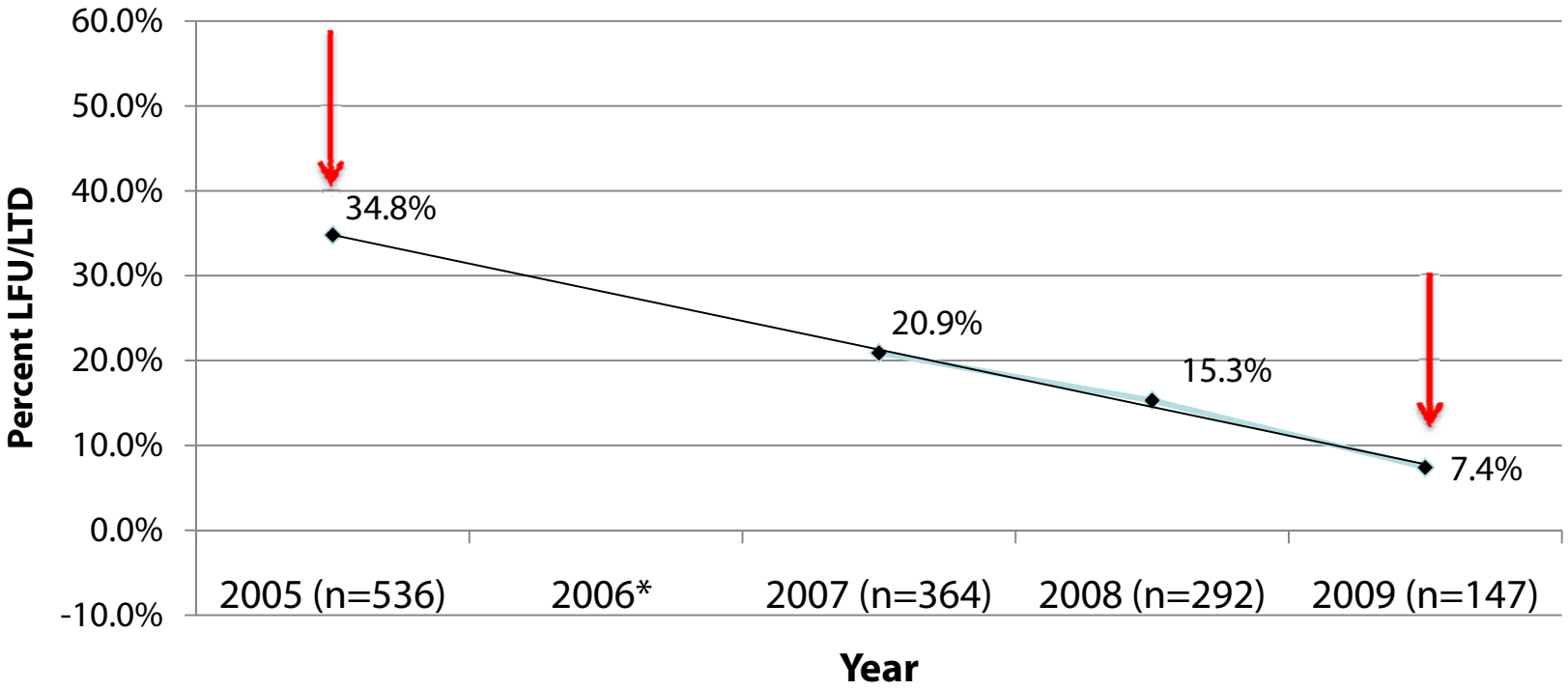
Follow-up for Early Hearing Detection and Intervention (EHDI)

- **CDC annual hearing screening and follow-up survey (2009)**
  - 56,784 infants did not pass the hearing screening
  - 25,334 (44.6%) had no documentation of further assessment without a valid reason indicated
- **Data systems, reporting, and follow-up need improving**



Need to Improve Follow-up Documentation in State EHDI Programs

Infants lost to follow-up and documentation following hearing screening, Indiana, 2005–2009



LFU, loss to follow-up
LTD, loss to documentation
2006, incomplete data reported
CDC EHDI, Early hearing detection and intervention (EDHI) hearing screening and follow-up survey

Lessons Learned

- **Newborn screening is a public health success story**
  - Saves lives
  - Prevents disability
  - Saves money
- **Better data systems and follow-up are needed**

Closing Gaps and Improving Outcomes Through Partnerships

V. Fan Tait, MD, FAAP

Associate Executive Director

American Academy of Pediatrics

http://www.aap.org

U.S. Department of Health and Human Services
Centers for Disease Control and Prevention

Long-term Follow Up

- **Goal**
  - Ensure best possible outcomes for individuals
- **Strategies**
  - Coordinate medical and multidisciplinary care for children and their families
  - Monitor progression of disorders identified through newborn screening
  - Improve information access, quality and timeliness through health information technology
  - Establish evidence-based best practices

Importance of Long-term Follow-up

- **Assess needs of children/families**
  - Chronic disease management
  - Condition-specific treatment
  - Age-appropriate preventive care throughout the lifespan of affected individuals
- **Track clinical outcomes of children**
  - Improve data quality in tracking and surveillance systems
  - Inform ongoing follow-up protocols
- **Improve quality of care**
  - Knowledge discovery, particularly for disorders with unclear natural histories
  - Develop care guidelines and clinical decision support

Barriers to Long-term Follow-up

- **Family-centered**
  - Understanding importance of follow-up
  - Timely dissemination of information
- **Condition-specific**
  - Heterogeneity of conditions
- **Health care system**
  - Slow adoption of national standard codes and vocabulary
  - Varying workforce capacity
  - Lack of interoperability of electronic health records
- **State programs**
  - Capacity and ownership

Current Status of Long-term Follow-up

- **Diagnosis and short-term follow-up remain a challenge**
  - Timely confirmatory testing and diagnosis
- **Role of pediatricians**
  - 72% feel they are the primary coordinators of care for children and youth with special health care needs
  - Only <48% developed a care plan in collaboration with other health care professionals and agencies
- **Variation in state newborn screening programs**
  - How the long-term follow-up is defined, staffed, and conducted
  - 56% of the state newborn screening programs reported they do not collect long-term follow up data

Hoff, T et al. Arch Pediatr Adolesc Med.2007;161(10):994-1000

Gaps within the Public Health and Health Care Systems

- **Co-management of complex disorders by primary care providers and pediatric subspecialists**

- **Communication between primary care providers and state newborn screening programs**
- **Gathering uniform long-term follow-up data**
  - Varying case definitions for disorders screened in newborns
  - Sparse follow-up data systems
  - State-specific data elements

Improving the Long-term Follow-up

1. Strengthen **partnerships** between public health programs and primary care providers
2. **Coordinate care** through the "medical home"
3. **Improve quality** and evidence base for treatment of disorders screened in newborns
4. Develop **education** about newborn screening and technical assistance for state programs, medical providers, and patients/families
5. Enhance federal and state **policies**

1. Partnerships between Public Health Programs and Primary Care Providers

- **National level**
  - Forum for providers, public health, and families to collaborate
  - Data collection, education, laboratory services, clinical services, and ethics
- **State level**
  - With chapters of the professional societies
- **Local levels**
  - With state newborn screening and Title V programs

1. Partnerships between Public Health Programs and Primary Care Providers

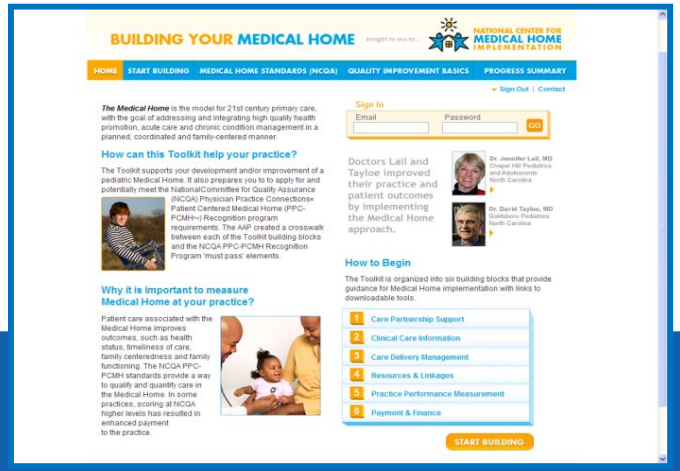
- **Resources and information available to support forming of partnerships**
  - Regional Genetics and Newborn Screening Services Collaboratives
  - NIH-funded Newborn Screening Translational Research Network
  - Genetic Alliance
  - National Newborn Screening and Genetics Resource Center
  - National Center on Hearing Assessment and Management

Regional Genetics Newborn Screening Services Collaboratives

http://www.nccrcg.org

2. Coordinating Care through the Medical Home

- **Multidisciplinary care needed**
  - Primary care, multiple specialists, physical therapies, developmental assessments and interventions, and social services
  - Public and private insurance and funding
- **Comprehensive care plan**
  - Medical summary, emergency treatment and management plans
- **Co-managed care by physicians and specialists**
  - Clear roles and responsibilities
  - Timely communication and information exchange



www.pediatricmedhome.org

3. Improving Quality and Evidence Base for Treatment of Disorders

- **Newborn Screening ACTion Quality Improvement Innovation Network**
  - Partnership with Maternal and Child Health Bureau, American Academy of Pediatrics, and American College of Medical Genetics
- **Newborn Screening Education in Quality Improvement for Pediatric Practice course**
  - Partnership with the American Academy of Pediatrics and CDC
- **Improving hearing screening and intervention services**
  - Project of the National Initiative for Children's Healthcare Quality and Maternal and Child Health Bureau

4. Developing Education, Technical Assistance, and Collaboration

- **American Academy of Pediatrics guidance documents**
  - Newborn Screening expands: Recommendations for Pediatricians and Medical Homes – Implications for the System
    - http://pediatrics.aappublications.org/content/121/1/192.full
  - Year 2007 Position Statement: Principles and Guidelines for Early Hearing Detection and Intervention
    - http://aappolicy.aappublications.org/cgi/content/full/pediatrics;120/4/898
- **National Technical Assistance Centers**
  - National Newborn Screening and Genetics Resource Center
    - http://genes-r-us.uthscsa.edu
  - National Center on Hearing Assessment and Management
    - http://www.infanthearing.org
  - Newborn Screening Clearinghouse
    - http://www.nbsclearinghouse.org
- **Genetics in Primary Care Institute**
    - http://www.medicalhomeinfo.org/GPCI

5. Enhancing Federal and State Policies

- **Align state newborn screening program capacity with long-term follow-up**
- **Standardize long-term follow-up activities**
  - Harmonize clinical definitions and nomenclature for confirmed cases
  - Establish overall data standards, e.g., common variables and data collection procedures to facilitate combination of datasets across states
  - Ultimately, promote and facilitate the use of electronic health data standards in recording and transmitting newborn screening information

Data Standards for Electronic Reporting: http://newbornscreeningcodes.nlm.nih.gov

Newborn Screening
Public Health Success

- Saves lives
- Prevents disability
- Saves money