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A program of the American Academy of Pediatrics

American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN™

American College of Medical Genetics
Medical Genetics: Translating Genes Into Health®

The Science and the System: An Overview of NBS

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Newborn Screen Positive Infant ACTion Project
Learning Session
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Purpose of Newborn Screening

- Preventive public health program
- Early detection of certain metabolic, hematologic and endocrine disorders in newborns
- Intervention to prevent morbidity and mortality

If Untreated, Disorders

- Can result in:
  - Growth problems
  - Developmental delays
  - Behavioral/emotional problems
  - Deafness or blindness
  - Retardation
  - Seizures
  - Coma, sometimes leading to death

Criteria for screened disorders

- Disorder occurs with significant frequency
- Test are inexpensive and reliable
- Effective treatment/intervention exists
- If untreated, baby may die or develop severe retardation
- Affected baby may appear normal at birth

Newborn screening is a system for identifying genetic and other health problems in newborns that leads to overall improvement in the public's health.

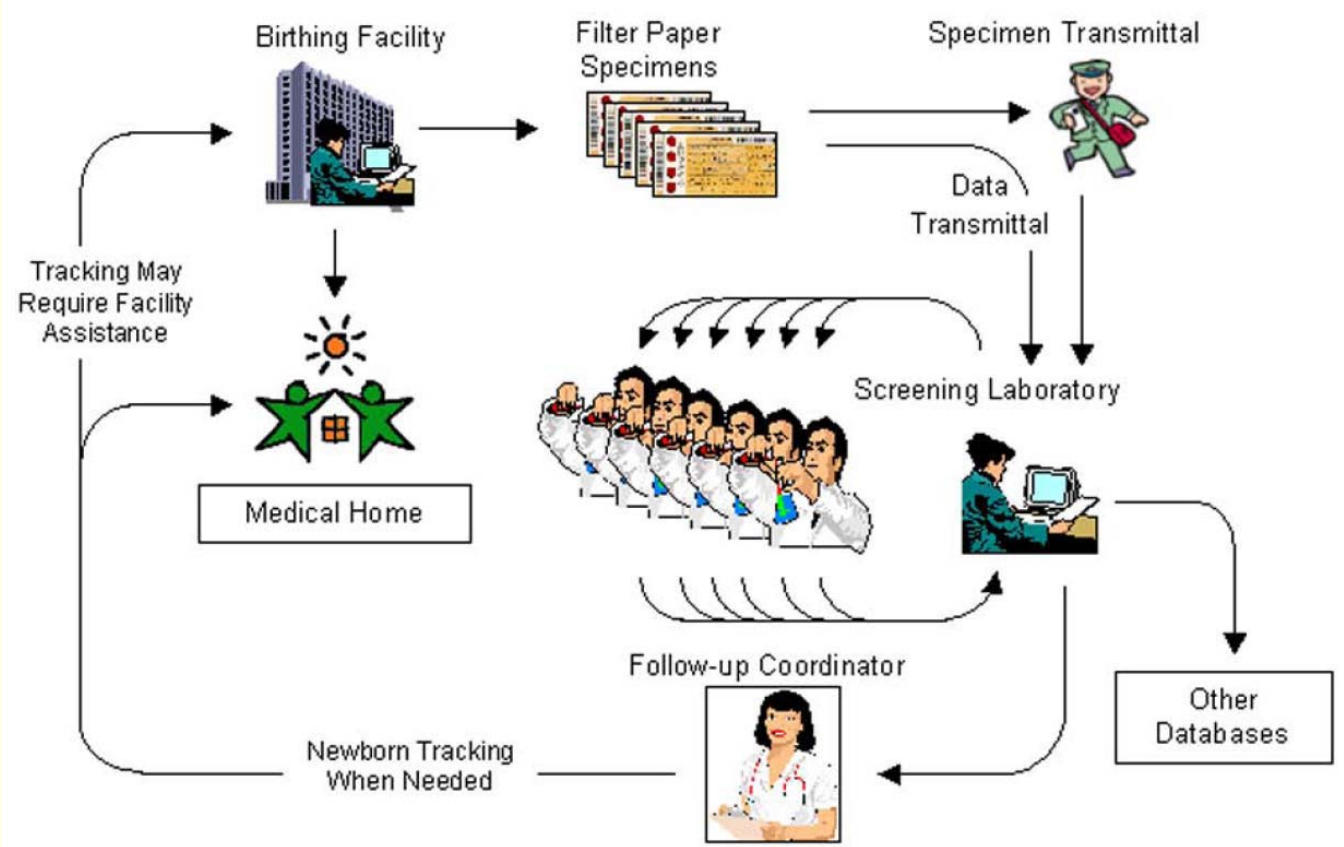

FIG. 1. Flow diagram for a typical system for the screening of newborns.

Ref. Therrell BL, David-Padilla C. Screening of Newborns for Congenital Hypothyroidism. Vienna: IAEA, 2005, p. 21.

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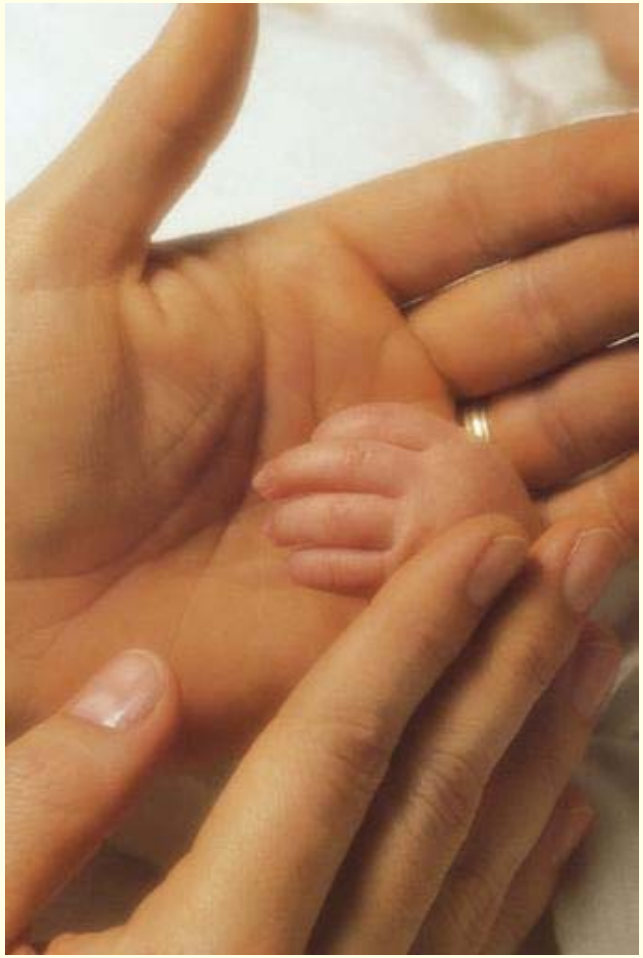
NBS is a 6-Part System

- Screening
- Follow-up
- Diagnosis
- Management
- Evaluation
- Education
- (Finance)



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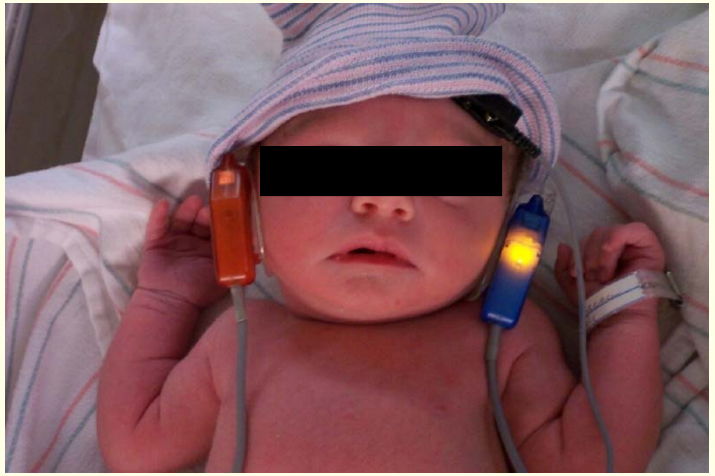
Part 1: Screening

- universal blood testing of all newborns

- Universal hearing screening of all newborns



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Tandem Mass Spectrometer (MS/MS)

- Molecules are sorted & weighed by mass
- Compounds analyzed are amino acids & acylcarnitines
  - Amino acids: building blocks for proteins
  - Acylcarnitine= Carnitine (vehicle) +fatty acid
    - Identified by size of fatty acid: short, medium, long and designated by initials & numbers

Organic Acid Metabolism Disorders

- IVA - Isovaleric acidemia
- GA I – Glutaric acidemia type I
- HMG – 3-OH 3-CH3 glutaric aciduria
- MCD – Multiple carboxylase deficiency
- MUT – Methylmalonic acidemia (mutase def)
- 3MCC – 3-Methylcrotonyl-CoA carboxylase deficiency
- Cbl A,B – Methylmalonic acidemia
- PROP – Propionic acidemia
- BKT – Beta-ketothiolase deficiency

Fatty Acid Oxidation Disorders

- MCAD – Medium-chain acyl-CoA dehydrogenase deficiency
- VLCAD – Very long-chain acyl-CoA dehydrogenase deficiency
- LCHAD – Long-chain L-3-OH acyl-CoA dehydrogenase deficiency
- TFP – Trifunctional protein deficiency
- CUD – Carnitine uptake defect

Amino Acid Metabolism Disorders

- PKU – Phenylketonuria
- MSUD – Maple syrup urine disease
- HCY – Homocystinuria
- CIT – Citrullinemia
- ASA – Argininosuccinic acidemia
- TYR I – Tyrosinemia type I

Hemoglobinopathies

- S/S – Sickle cell anemia
- S/Beta-Thal – S/ Beta-thalassemia
- S/C – Sickle C disease
- FE – Homozygous E Disease
- FC – Homozygous C Disease
- S/E – Sickle E Disease
- Others depending on state program

Others

- CH – Congenital hypothyroidism
- CAH – Congenital adrenal hyperplasia
- BIOT – Biotinidase deficiency
- GALT – Galactosemia
- HEAR – Hearing deficiency
- CF – Cystic fibrosis

Part 2: Follow-up

- Obtain test results
- Locate family (out of range, critical)
- Relay results to family
- Repeat test(s) if necessary
- Ensure diagnostic process has begun

Part 3: Diagnosis

- Assessment by Specialist
- Family consulted
- Counseling if necessary

Part 4: Management

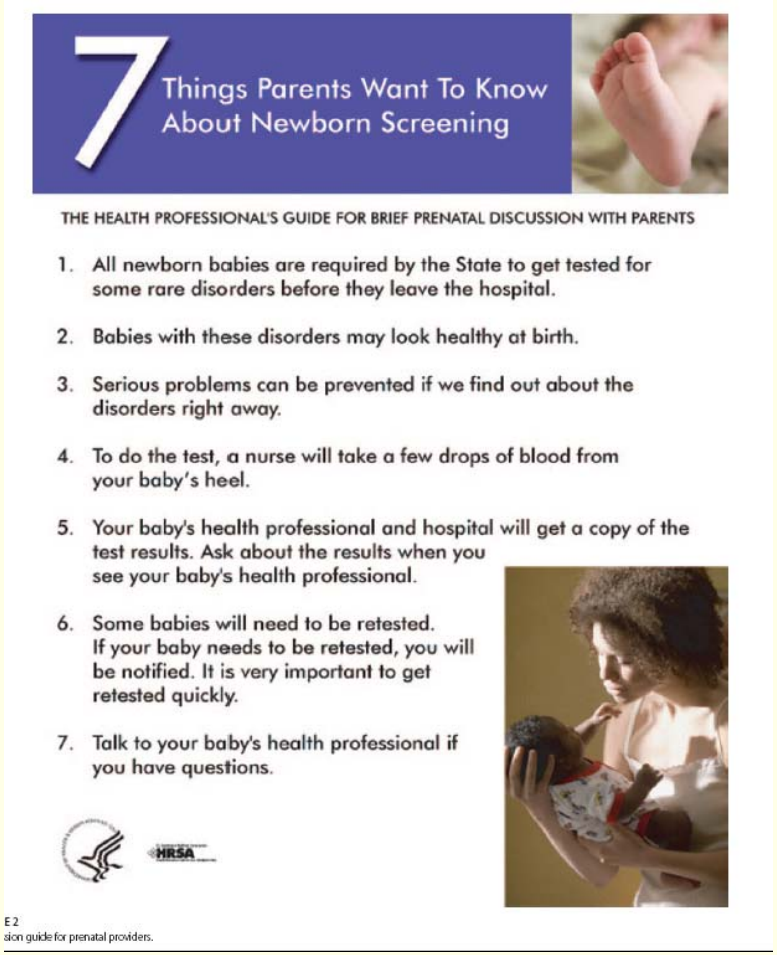
- Treatment
- Long-term follow-up
- Coordination through the medical home

Part 5: Evaluation

- Validation of testing procedures
- Program evaluation
- Outcome evaluation
- Cost effectiveness

Part 6: Education

- Parents
- Health care providers
- Hospitals
- Legislators



7 Things Parents Want To Know About Newborn Screening

THE HEALTH PROFESSIONAL'S GUIDE FOR BRIEF PRENATAL DISCUSSION WITH PARENTS

1. All newborn babies are required by the State to get tested for some rare disorders before they leave the hospital.
2. Babies with these disorders may look healthy at birth.
3. Serious problems can be prevented if we find out about the disorders right away.
4. To do the test, a nurse will take a few drops of blood from your baby's heel.
5. Your baby's health professional and hospital will get a copy of the test results. Ask about the results when you see your baby's health professional.
6. Some babies will need to be retested. If your baby needs to be retested, you will be notified. It is very important to get retested quickly.
7. Talk to your baby's health professional if you have questions.

HRSA

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sion guide for prenatal providers.

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NBS is a comprehensive system

- Private/Public Medical Practitioners
- Laboratory Personnel
- Administrative and Follow-up Personnel
- Educators
- Specialty Care Centers
- Source(s) of Payment
- Family Members
- Other Interested Individuals (Legislators, Professional Societies, CBOs)

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Resources

- National Newborn Screening and Genetics Resource Center (NNSGRC)
  - http://genes-r-us.uthscsa.edu/
- National Coordinating Center for the Regional Genetic and Newborn Screening Collaborative Groups (NCCRCG)
  - http://www.nccrcg.org//AM/Template.cfm?Section=Home5

The Regional Collaborative Groups

New England Genetics Collaborative (NEGC)
Connecticut, Massachusetts, Maine, New Hampshire, Rhode Island and Vermont

York-Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC)
Delaware, District of Columbia, Maryland, New Jersey, New York, Pennsylvania, Virginia and West Virginia

Southeast NBS & Genetics Collaborative (SERC)
Alabama, Florida, Georgia, Louisiana, Mississippi, North Carolina, Puerto Rico, South Carolina, Tennessee and the Virgin Islands

The Region 4 Genetics Collaborative (Region 4)
Illinois, Indiana, Kentucky, Michigan, Minnesota, Ohio and Wisconsin

Heartland Genetics and Newborn Screening Collaborative (Heartland)
Arkansas, Iowa, Kansas, Missouri, Nebraska, North Dakota, Oklahoma and South Dakota

Mountain States Genetics Regional Collaborative Center (MSGRCC)
Arizona, Colorado, Montana, Nevada, New Mexico, Texas, Utah and Wyoming

Western States Genetic Services Collaborative (WSGSC)
Alaska, California, Guam, Hawaii, Idaho, Oregon and Washington