The Science and the System: An Overview of NBS

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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.

NBS is a 6-Part System

- Screening
- Follow-up
- Diagnosis
- Management
- Evaluation
- Education
- (Finance)
Part 1: Screening

- universal blood testing of all newborns
- Universal hearing screening of all newborns
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
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)
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)
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