Newborn Screening – It’s Complicated

Newborn Screening and Genetics in Public Health Committee

June 2, 2018
Part 2......

Newborn Screening Laboratory Perspectives

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Texas Department of State Health Services
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Success Stories

Blood samples from Ontario newborns used to study rare diseases in babies

Gov. Northam signs two bills into law that reduce paperwork for parents

Newborn screenings helping to save lives

GREAT STRIDES: ONE FAMILY'S CYSTIC FIBROSIS STORY

Kids Day: Genetics testing keeps Nathaniel going
Addition of New Conditions

Texas Families Urge Lawmakers To Move Faster To Screen Newborns For Deadly Genetic Disease

Https://dfw.cbslocal.com, January 8, 2018
What Kind of Authorization Does Your State Need?

- Automatic inclusion – Follow national recommendation
- Department head approval
- State Advisory Committee/ workgroup approval
- State legislative authorization
- Others
Is Authorization Sufficient?

How Illinois bureaucracy robbed parents of a chance to save their children from a deadly disease
Lots Activities/Partners Involved

- Funding (various funding sources)
- Equipment and reagents
- Building retrofit
- Staffing and training
- Testing method, workflow, and algorithm
- Follow-up protocols
- Information technology
- Communication and education
- Competing priorities
- .........
Timeliness is Important

Deadly Delays: States fail to meet newborn screening goals
Critical Core Conditions

<table>
<thead>
<tr>
<th>Organic Acid Conditions</th>
<th>Fatty Acid Oxidation Disorders</th>
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<tbody>
<tr>
<td>Propionic Acidemia (PROP)</td>
<td>Medium Chain Acyl-CoA-dehydrogenase deficiency (MCAD)</td>
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<tr>
<td>Methylmalonic Acidemia (methylmalonyl-CoA mutase) (MUT)</td>
<td>Very Long chain acyl-CoA dehydrogenase deficiency (VLCAD)</td>
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<tr>
<td>Isovaleric Acidemia (IVA)</td>
<td>Long chain L-3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)</td>
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<tr>
<td>3-Hydroxy-3-methyglutaric aciduria (HMG)</td>
<td>Trifunctional protein deficiency (TFP)</td>
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<td>Holocarboxylase synthase deficiency (MCD)</td>
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<td>β-Ketothiolase deficiency (BKT)</td>
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<tr>
<td>Glutaric Aciduria, Type 1 (GA1)</td>
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**Amino Acid Disorders**

<table>
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<tr>
<th>Other</th>
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<tbody>
<tr>
<td>Argininosuccinic Aciduria (ASA)</td>
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<tr>
<td>Citrullinemia type 1 (CIT)</td>
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<tr>
<td>Maple syrup urine disease (MSUD)</td>
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</tbody>
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Critical Condition:

- A condition on the RUSP in which acute symptoms or potentially irreversible damage could develop in the first week of life, and for which early recognition and treatment can reduce risk of morbidity and mortality.

16 critical conditions

Source: SIMD Position Statement August 21, 2014
NBS System Process

Analytical

Specimen Punched → Testing Performed → Results Verified

Results Reported

Physician and Parent Notified of Presumptive Positive Result

Diagnostic Testing

Diagnosis

Treatment

Long-term Follow-up And Management

Continuous Quality Improvement

Pre-Analytical

Specimen Received

Specimen Transported

Specimen Collection

Infant Born

Parent & Provider Education

Post-Analytical

Analytical

Analytical

Pre-Analytical

Post-Analytical
Coordination with Healthcare Providers

- **Timeliness:** Collection, courier, reporting, and compatibility of electronic ordering and reporting system
- **Specimen quality**
- **Completeness and accuracy of demographic information**
- **Education:** importance of communication, training, and site visits
Ignore Science and Common Sense?

MKE DE SISTI / MILWAUKEE JOURNAL SENTINEL

A JOURNAL SENTINEL SPECIAL REPORT

The price of being wrong

Milwaukee Journal Sentinel, December 9, 2016
Testing Method, Workflow, and Algorithm

• Screening vs diagnostic testing
• Determination and evaluation of appropriate cutoff used for risk assessment
  • Various age, weight, ethnicity, status, feed
  • Decrease false positives
  • Decrease false negatives and heavy media attention around delayed diagnoses
• Different testing platforms with various technologies, capacity, and complexity
• Compatibility with existing NBS workflow (all test the same sample and results in the same report)
• Data harmonization and normalization
Storage and Use of Residual Dried Blood Spots

Lawsuit alleges Michigan stole blood of newborn babies
By Associated Press
April 12, 2018 | 8:00pm

DNA of every baby born in California is stored. Who has access to it?

CBS News, May 13, 2018
Residual DBS

• Variability - State policies and laws
• Confidentiality
• Ethics – Need for consent?
• Storage – Duration, condition, location
• Allowable uses – Who, Why, How,….
• Process for release, destruction, and documentation
• Transparency
Be aware of what’s going on nationally – it will impact you!
Consistent, proactive communication within the entire system to foster collaboration and support