Update on NC Newborn Screening Program
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Revision of Newborn Screening Form
• Changed from two filter papers to one filter paper
  5 part form ----→ 4 part form
• Color changes every year
• Most are minor changes –
  – Updated SLPH’s web site address
  – Removed Medicaid number field
  – Collection date and time
  – Hearing screening information

Revision of Newborn Screening Form
• Instructions for hearing screening
  – If the hearing screening was not done before the specimen was sent to SLPH
  – In all cases, the hearing screening should be attempted before discharge
New Filter Paper Lay-Out

New LIMS System

• Migrated to new application in July, 2010
• More work needs to be done

New Tests Considered

• Severe Combined Immunodeficiency (SCID): Bubble boy disease
• Tyrosinemia Type I
Severe Combined Immunodeficiency (SCID)

- A primary immunodeficiency disease
- Affected newborns lack T lymphocytes or white blood cells that help fight infection
- Babies with SCID appear healthy at birth, but cannot survive without early treatment

SCID

- Treatment: bone marrow transplant from a healthy donor
- Survival rate: 97% if treated within 3 months of birth

SCID

- Recommended by Health and Human Services Secretary that SCID be included in all state’s screening programs
- Recommended by NC Newborn Screening Advisory Committee in January, 2011
Proposed Implementation for SCID

• Testing Algorithm
  – DNA based testing:
    • TREC (T cell Receptor Excision Circle)
    • TRECs=Marker of recent thymic emigrants
    • Low / No TRECs = Low / No T cell production by thymus

• Follow up
• Funding for Testing and Follow up

Tyrosinemia Type I Testing

• Current MS/MS technology (measuring only tyrosine) can detect Tyrosinemia Type II & III but not Type I

• Method measures Succinylacetone (SUAC) using MS/MS technology

• Plan to implement in new lab

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