Newborn Screening for Pompe Disease and other Lysosomal Storage Disorders: 

*Introduction to CDC Reference Methods and Materials*

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*Technical Workshop on Methods to Detect Pompe Disease and other Lysosomal Storage Disorders by Newborn Bloodspot Screening*

Atlanta, April 16-17, 2015
Disclosures

Dr. Vogt is a US/HHS federal research chemist at the CDC Newborn Screening and Molecular Branch. He serves as primary investigator of the Newborn Screening Translational Research Initiative (NSTRI), a 10-year collaboration between NSMBB and the CDC Foundation.

The CDC Foundation receives donations on behalf of NSTRI from Genzyme Corporation, a Sanofi Company; Biogen-Idec; and The Legacy of Angels Foundation.
CDC Provides Laboratory Support for Newborn Screening of LSDs

- **Quality Assurance Services**
  - Produce and certify Dried Blood Spot (DBS) reference materials
  - Conduct pilot Quality Control (QC) and Proficiency Testing (PT) surveys

- **Enhanced Support for Selected LSD Testing by MS/MS Methods**
  - Qualify and distribute reagents
  - Provide training and technical support
CDC Reagent Support for MS/MS Platforms

FDA Registered Analyte-Specific Reagents (ASR)*

Available for six LSD:
Fabry, Gaucher, Krabbe, MPS-I, Niemann-Pick A/B, and Pompe

*Manufactured by Genzyme

Tested and Distributed by NSTRI/CDC

NBS labs

Diagnostic labs

NBS Kit Producers

(Zhou et al, J Pediatrics. 2011; 159:7-13)
CDC Provides Enhanced Technical Support for Newborn Screening of LSDs by MSMS

- Standard Operating Procedures (SOPs)

- MSMS Tuning Solutions
  - Produce and characterized by CDC

- Laboratory Training workshops on MSMS Methods
  - 28 Training sessions (as of 2014)
  - 39 scientists from 26 programs and 12 countries

- DBS Reference Materials characterized by MSMS
  - Dose response quality control DBS with enzyme activity at low, medium and high levels
QC Blood Pool Production

1. Wash leuko-depleted blood with saline X3
2. Add heat-inactivated serum
3. Base pool 50 ± 2% Hematocrit
   - Umbilical Cord blood 50 ± 2% Hematocrit
4. 0% cord blood 100% Base pool
   - QC BP
5. 5% cord blood 95% Base pool
   - QC Low
6. 50% cord blood 50% Base pool
   - QC Med
7. 100% cord blood 0% Base pool
   - QC High

(De Jesus et al, Clin Chem 2009; 55(1); 158-164)
Preparation of Dried Blood Spot Proficiency Testing Materials for Lysosomal Storage Disorders

1. Wash leuko-depleted blood with saline X3
2. Adjust to 50% hematocrit with heat-inactivated serum
3. Harvest EBV-transformed cell cultures, wash and cell count
4. Condition-specific blood pools (3 ~ 5 x 10^7 cells/ml)
   - Pompe
   - MPS-I
   - Krabbe

Prepare Dried Blood Spots
Thank you for your attention!

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