

# 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*  
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# 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.**

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# 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**

# **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**

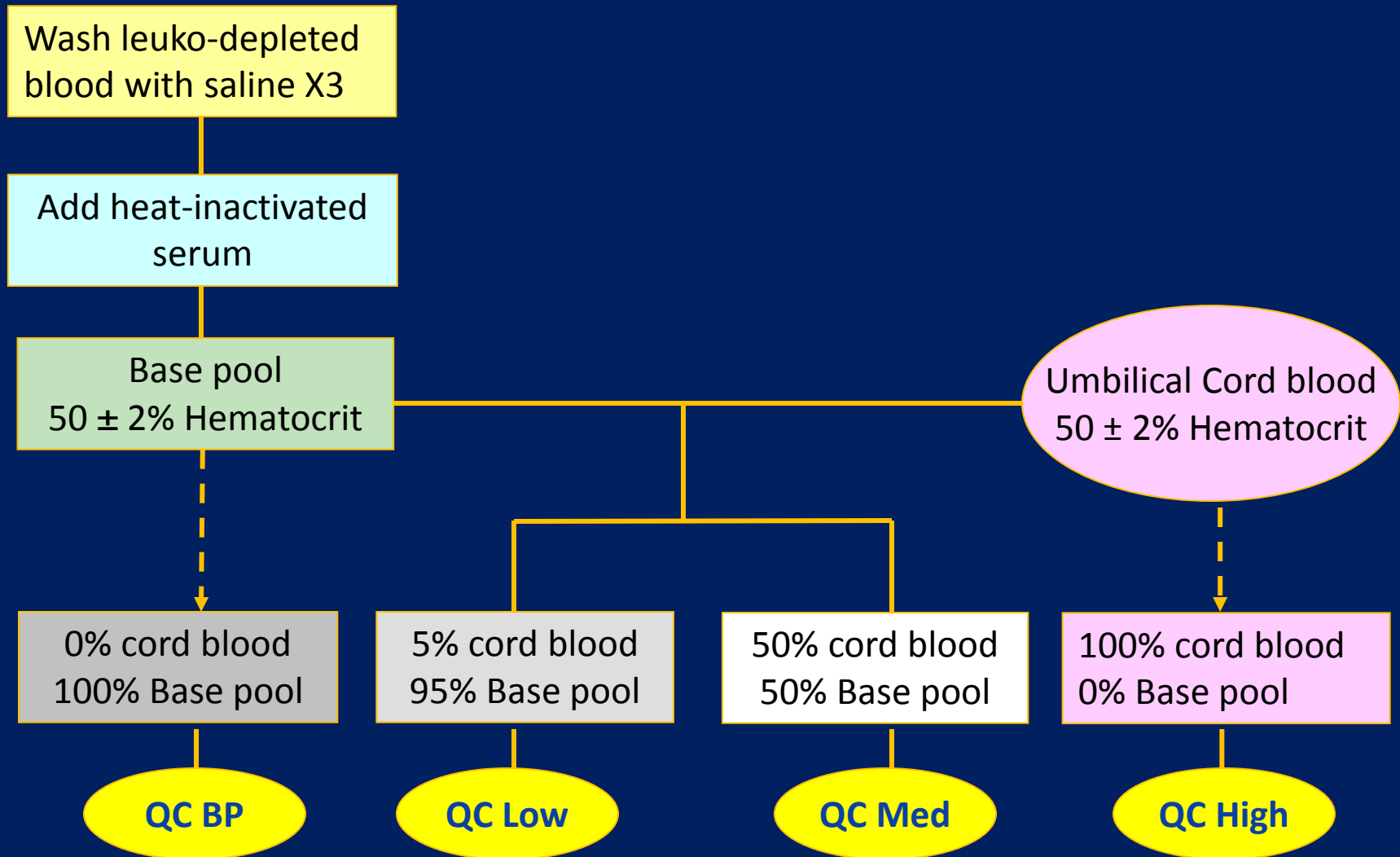
  - 28 Training sessions (as of 2014)

  - 39 scientists from 26 programs and 12 countries

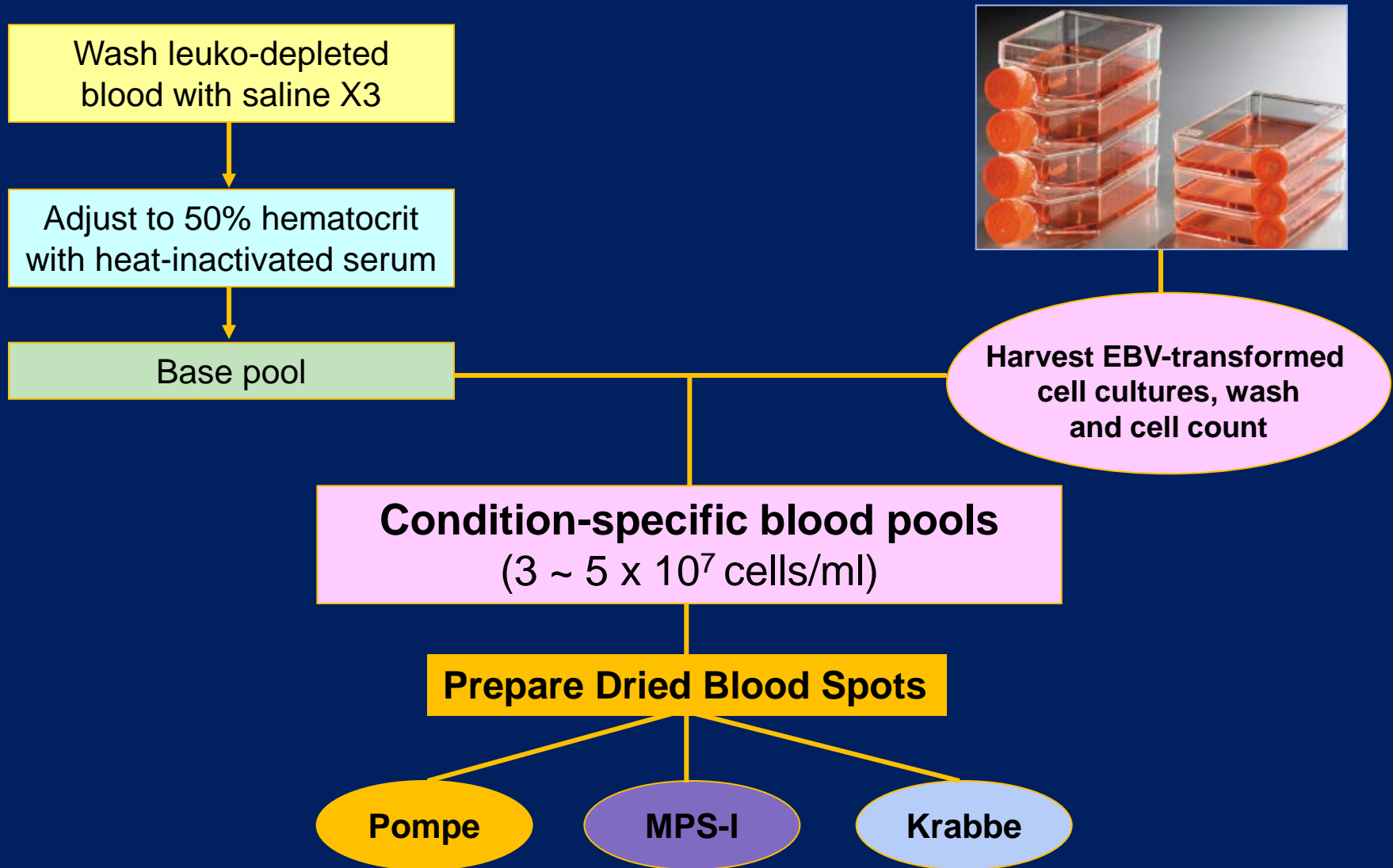
- ❑ **DBS Reference Materials characterized by MSMS**

  - Dose response quality control DBS with enzyme activity at low, medium and high levels

# QC Blood Pool Production



# Preparation of Dried Blood Spot Proficiency Testing Materials for Lysosomal Storage Disorders









# Thank you for your attention!

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