

# **Newborn Screening for Pompe Disease and other Lysosomal Storage Disorders: *Overview of Fluorometric Assays***

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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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# Quantitative Fluorescence Calibration

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## Fluorescence Calibration and Quantitative Measurement of Fluorescence Intensity; Approved Guideline

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### Abstract

Quantitative fluorescence calibration (QFC) is an empiric system to calibrate fluorescence intensity in a way that preserves stoichiometry between the concentration of fluorochrome in solutions and the equivalent molar quantity of fluorochrome on stained measurands such as cells, gels, microspheres, and microdots. This guideline describes the basic principles, reference materials, and laboratory procedures upon which QFC is based. This guideline is intended for use with reference materials and procedures developed under the National Institute of Standards and Technology (NIST) Fluorescence Intensity Standards program. While the general principles of QFC apply to any fluorescence measurement, this guideline specifically addresses analysis of cells and microspheres by flow cytometry, including cellular immunophenotyping and suspension array technology. The current and emerging uses of these laboratory methods will have an increasing impact on public health and primary care, from large-scale screening of populations to the individual profiling of each patient's disease.

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# Quantitative Fluorescence Calibration

$$i_f = [c][\epsilon][\phi][g\Omega] \int T(\lambda)Q(\lambda)s(\lambda)d\lambda$$

$i_f$



$]$

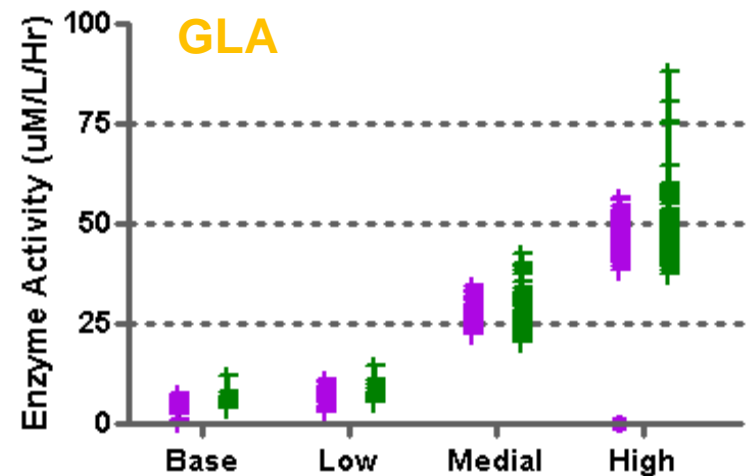
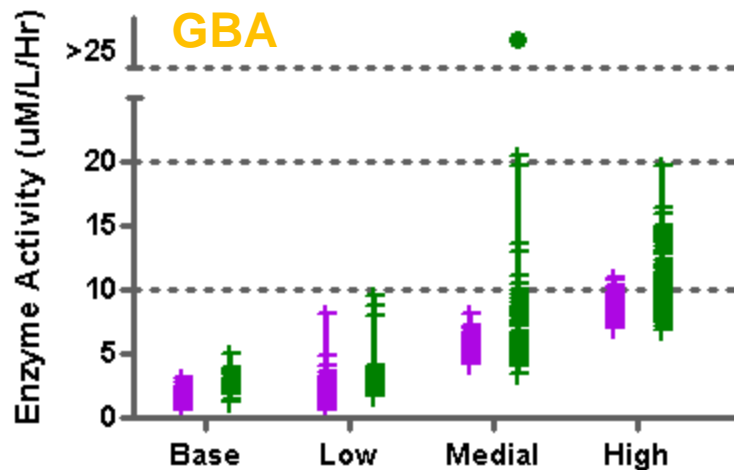
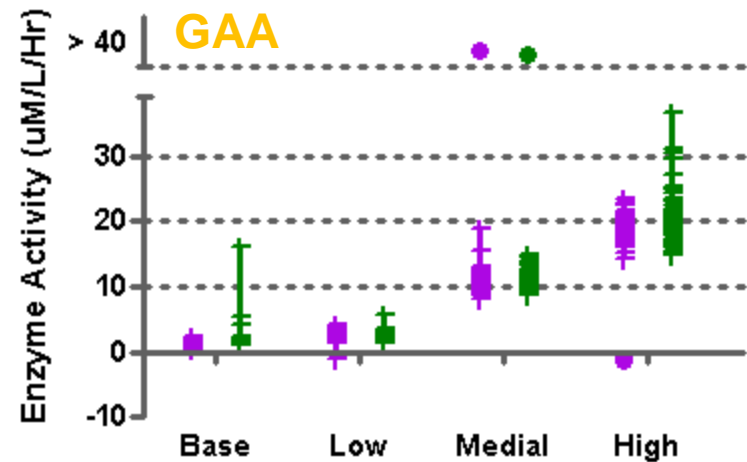
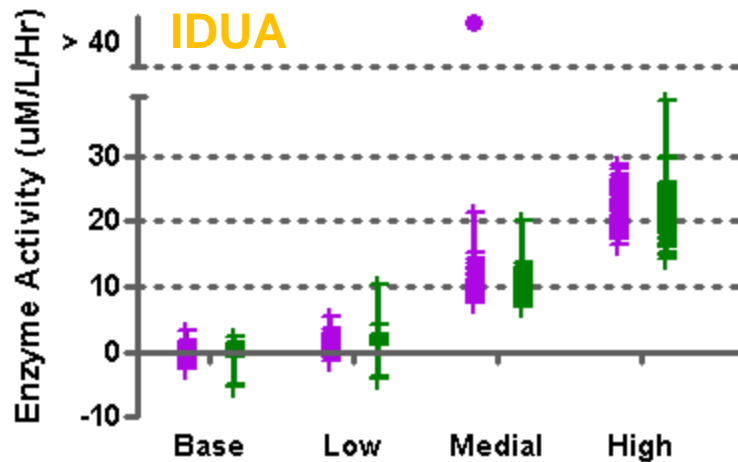
# **Fluorescence Assay Platforms Used for LSD-NBS**

**Microtiter Plate Fluorometry**

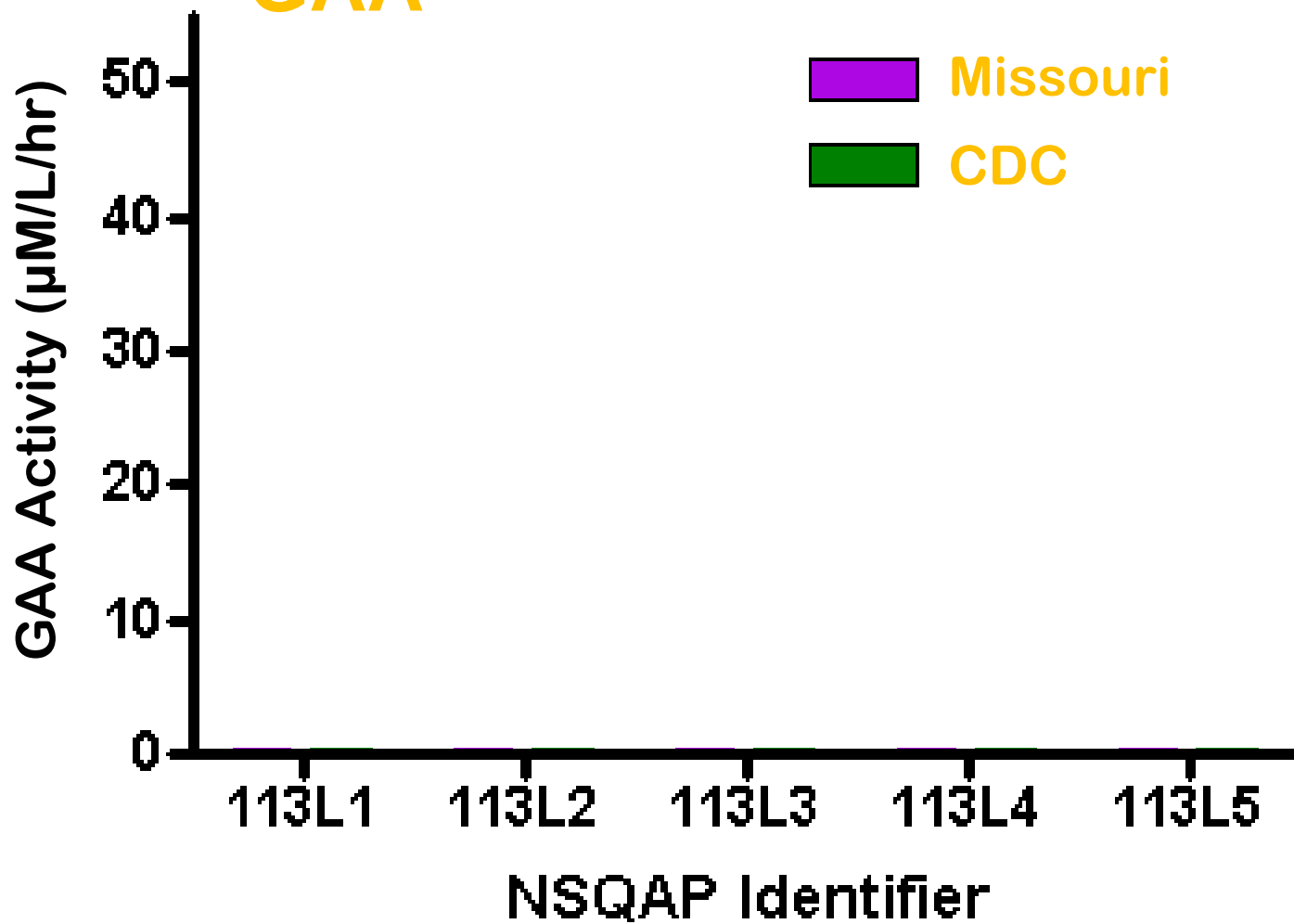
**Digital Microfluidics Fluorometry**

**(Microbead Suspension Fluorometry)**

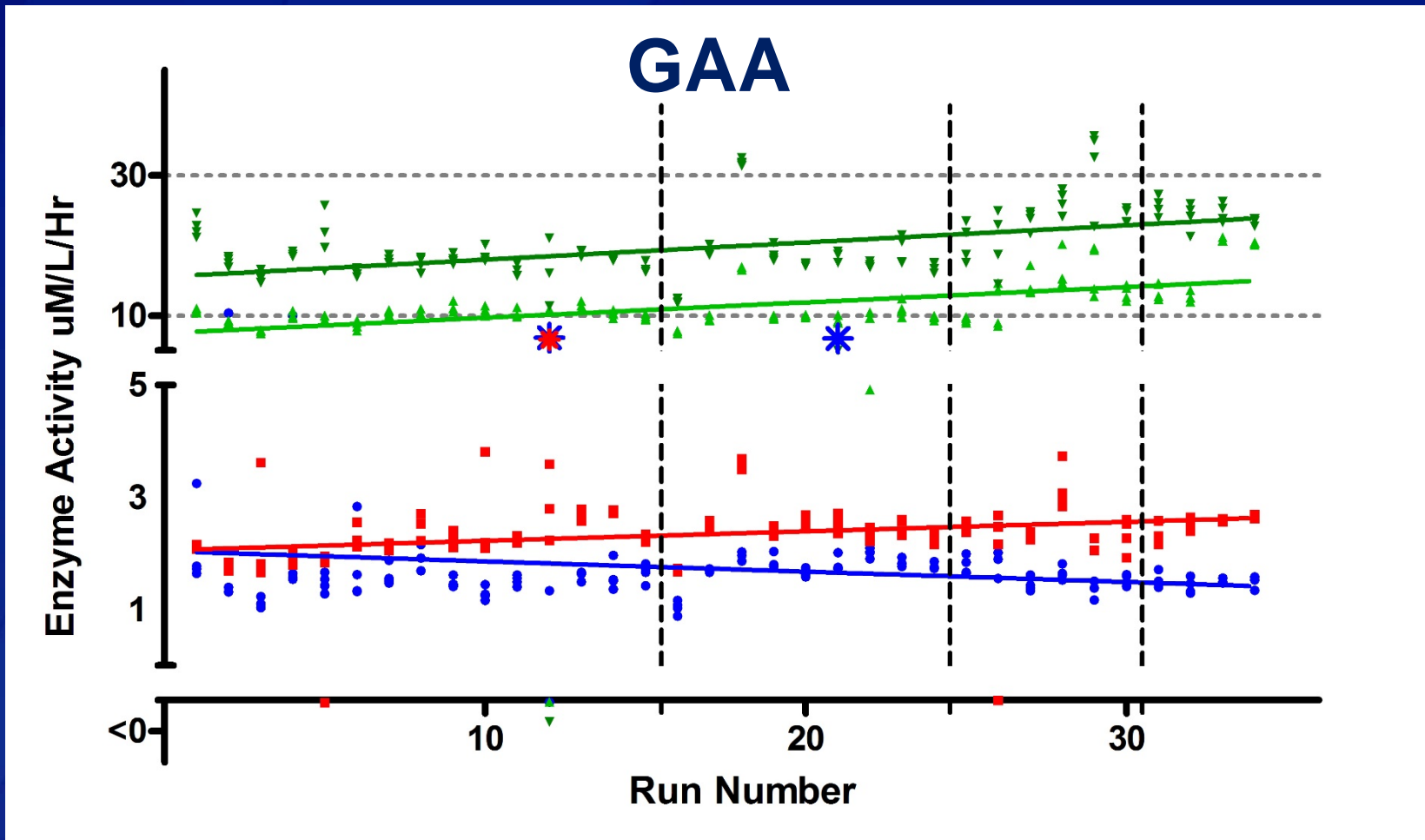
# Results from QC Blood Pools



# GAA

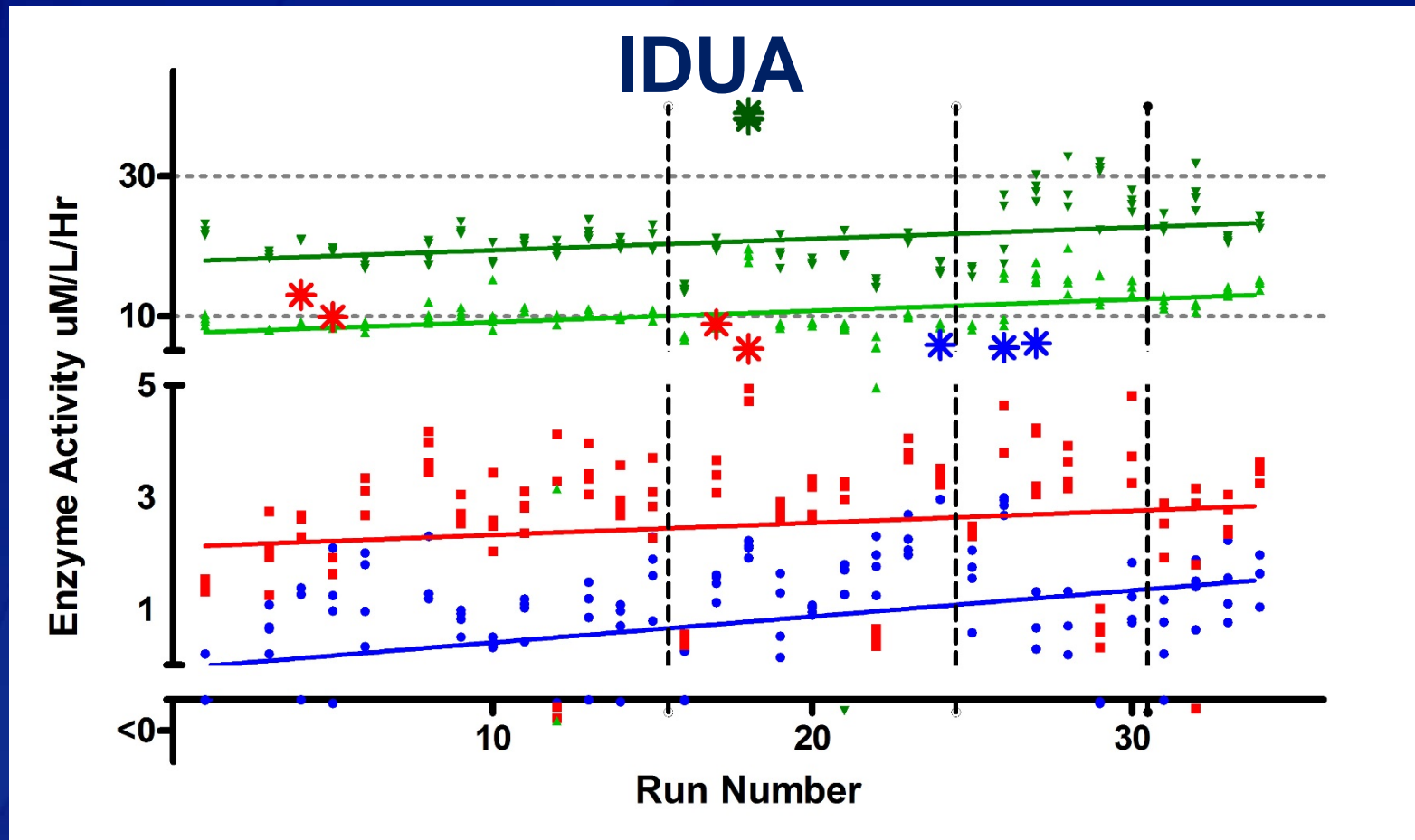


# CDC Results from QC Blood Pools (35 Runs over 3 Years)





# CDC Results from QC Blood Pools (35 Runs over 3 Years)





# Thank you for your attention!

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