# Missouri's Peer Network Resource Center For Pompe and MPS I



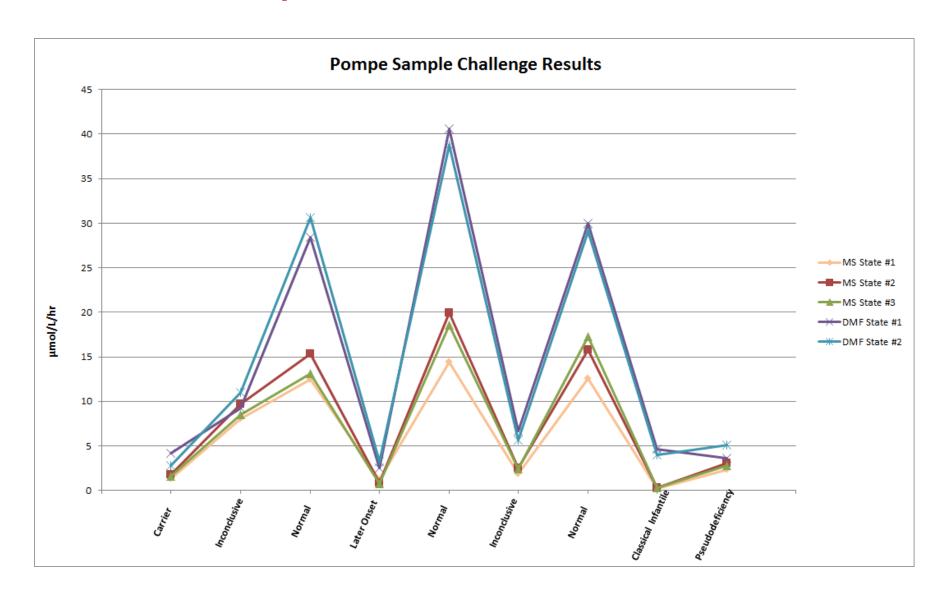
## **Summary of Services Offered by Missouri**

- Technical assistance for Lysosomal Storage Disorder (LSD) testing method validation and screening implementation
- Education for LSD screening and follow-up implementation
- First Tier Testing (for emergency/continuity of operations purposes)
- Second Tier Testing using Digital Microfluidics for four LSDs (Pompe, MPS I, Fabry, and Gaucher) to assist in method validation, pilot and implementation phases

## **Condensed/Simplified Implementation Plan**

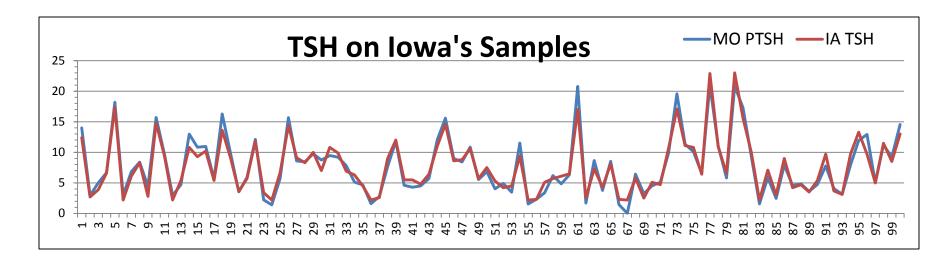
- Form an LSD Task Force
- ✓ Choose the screening method
- Obtain IRB approval for pilot study if needed
- Establish contract with vendor
- ✓ Installation, training and method familiarization
- ✓ Method validation/verification
- ✓ Pre-pilot phase (data collection on de-identified samples)
- ✓ Short Term Follow-up protocols
- ✓ Pilot Phase (statewide testing with referrals)
- ✓ Notification and education of stakeholders
- ✓ Live Testing with reporting on all NBS lab reports

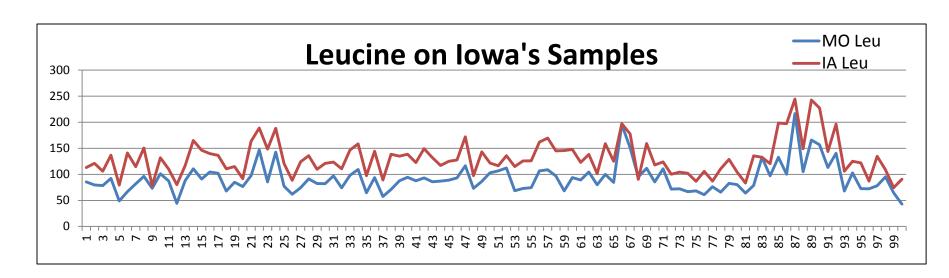
### **Samples for Method Validation**



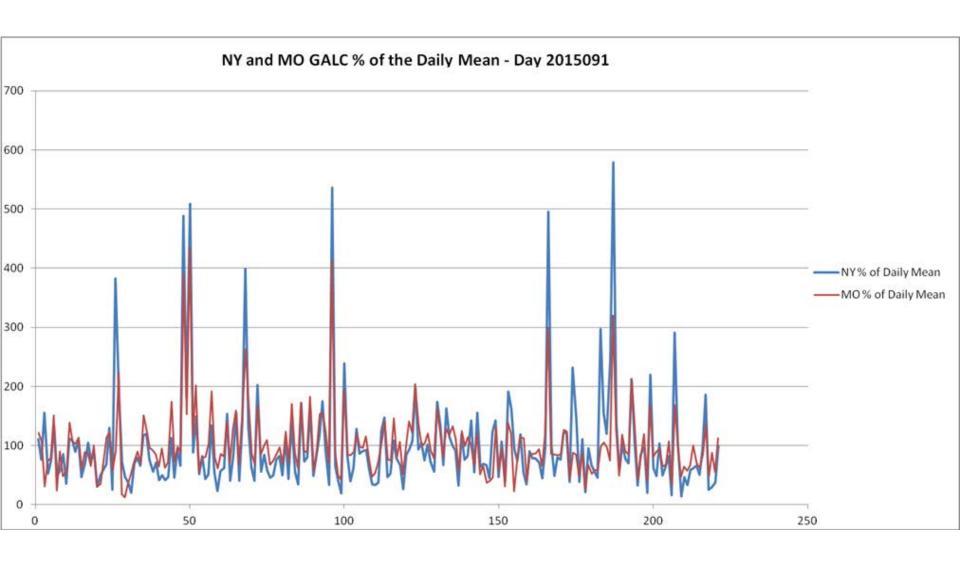
Note: We can provide them blinded or unblinded

## Pre-Tested Normal Samples for Method Familiarization and Validation





## **Results from Different Methods**



## 2<sup>nd</sup> Tier Testing to Assist With Your Validation, Pilot and Implementation Phases

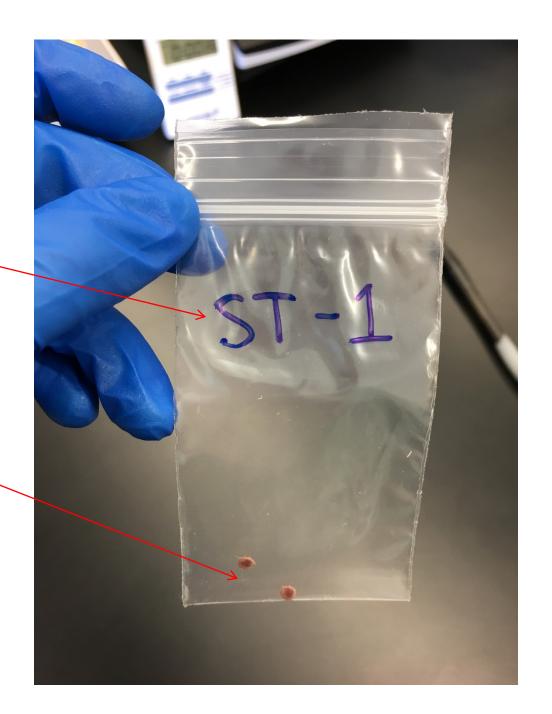
#### Samples Sent to Missouri for 2nd Tier Assessment of Pompe and MPS I

<u>Note</u>: The screening results and risk assessments below are unofficial and meant to assist the State NBS Program during their validation and pilot phase for implementing LSD screening. A normal result does NOT rule out the possibility of an underlying lysosomal storage/genetic disorder.

Sample ID	Other ID #	Age at Collection (hours)	Birth-weight (grams)	Gestation (weeks)	Race or Ethnicity	Value	IDUA WNL/ABN	Value	GAA WNL/ABN	Value	GBA WNL/ABN	Value	GLA WNL/ABN	Risk Assessment
ST-1	Example-01	24	3630	40	W	1.25	ABN	25.10	WNL	16.40	WNL	22.80	WNL	High Risk MPS I
ST-2	Example-02	29	2725	36	Α	11.25	WNL	22.02	WNL	13.88	WNL	12.89	WNL	Normal
ST-3	Example-03	25	3325	39	В	10.26	WNL	6.11	ABN	15.61	WNL	5.35	ABN	Borderline Pompe
ST-4	Example-04	32	3280	37	0	4.36	ABN	11.40	WNL	8.23	ABN	9.24	ABN	Inconclusive

Your State and Sample Starting with #1.

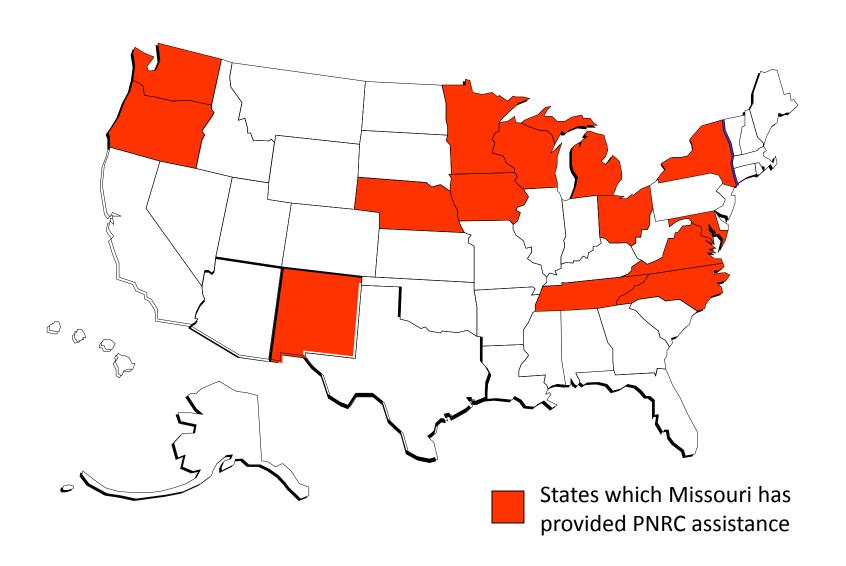
At least two punches, preferably three per baby



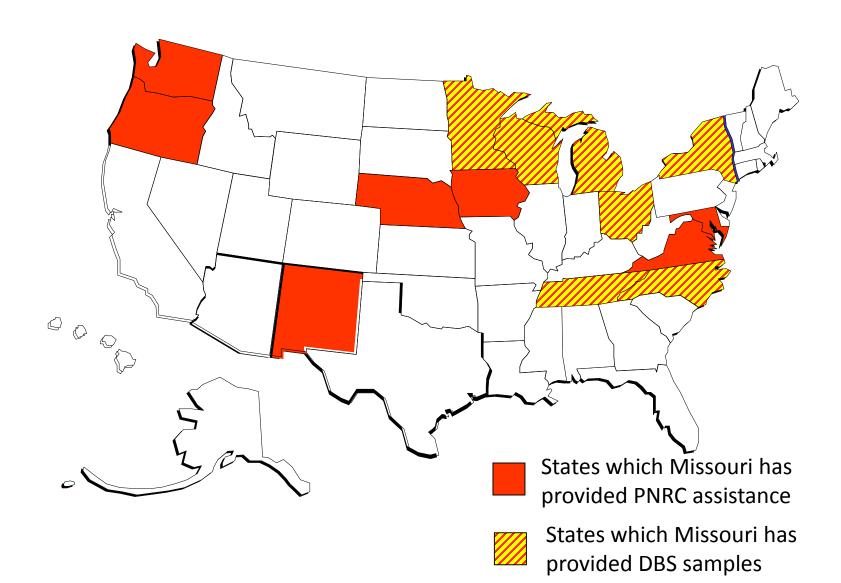
### Missouri's Packet of Informational Materials

- Frequently asked questions with suggested Do's and Don'ts
- Laboratory SOP and Method Validation Report
- Example laboratory reports with result comments
- Follow-up protocols
- Letters to PCP's from specialists
- Follow-up confirmatory data reports
- Notices sent to birthing hospitals and providers explaining our Pilot and Go-Live phases
- Spreadsheet of our LSD screening and confirmatory totals to date

## States Assisted by Missouri's PNRC This Year



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## **Acknowledgements**

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Monday, August 21, 2017

