

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



Missouri State Public Health Laboratory
Jefferson City, MO



Summary of Services Offered by Missouri

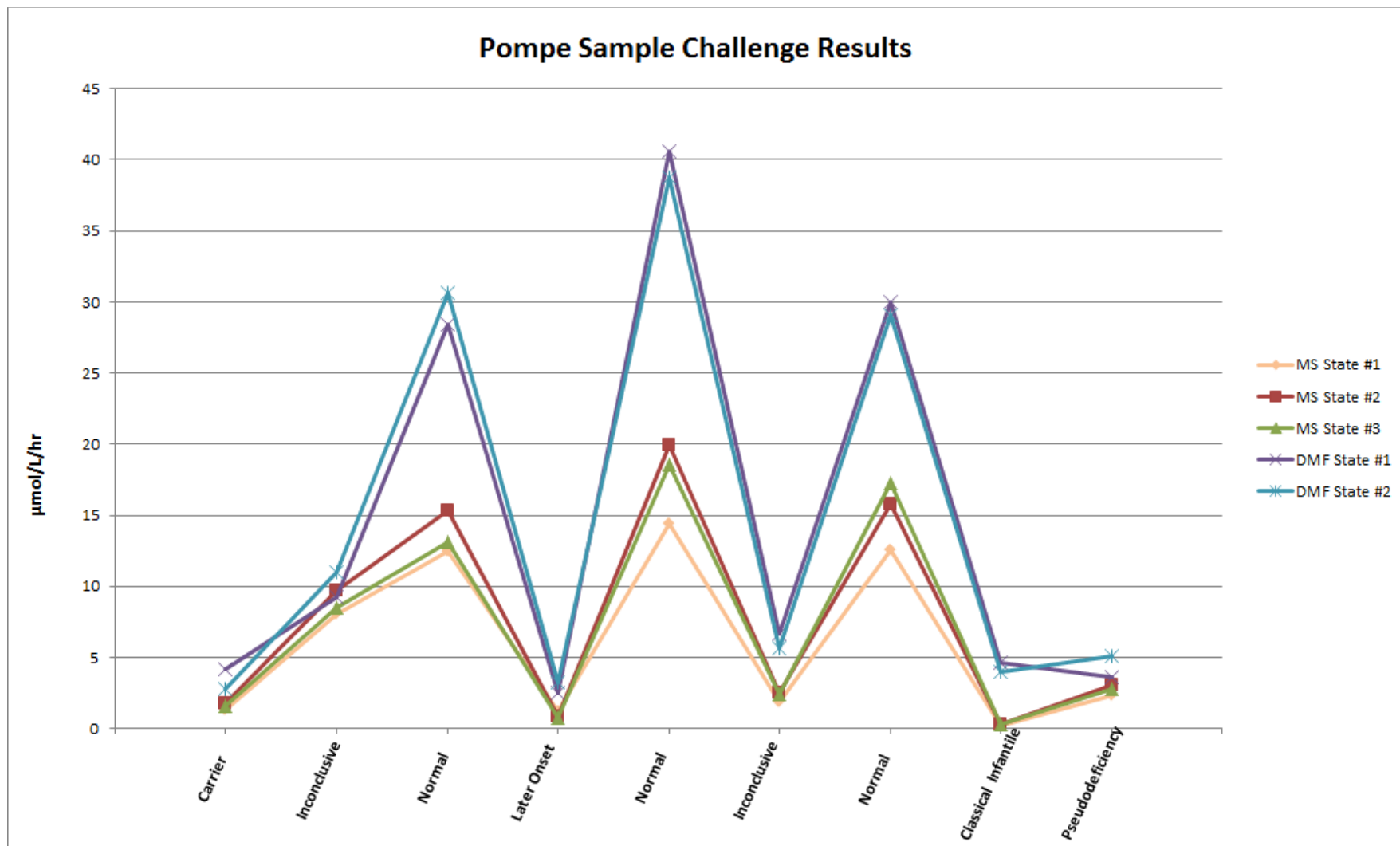
- Technical assistance for Lysosomal Storage Disorder (LSD) testing method validation and screening implementation
- Education for LSD screening and follow-up implementation
- First Tier Testing (for emergency/continuity of operations purposes)
- Second Tier Testing using Digital Microfluidics for four 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**

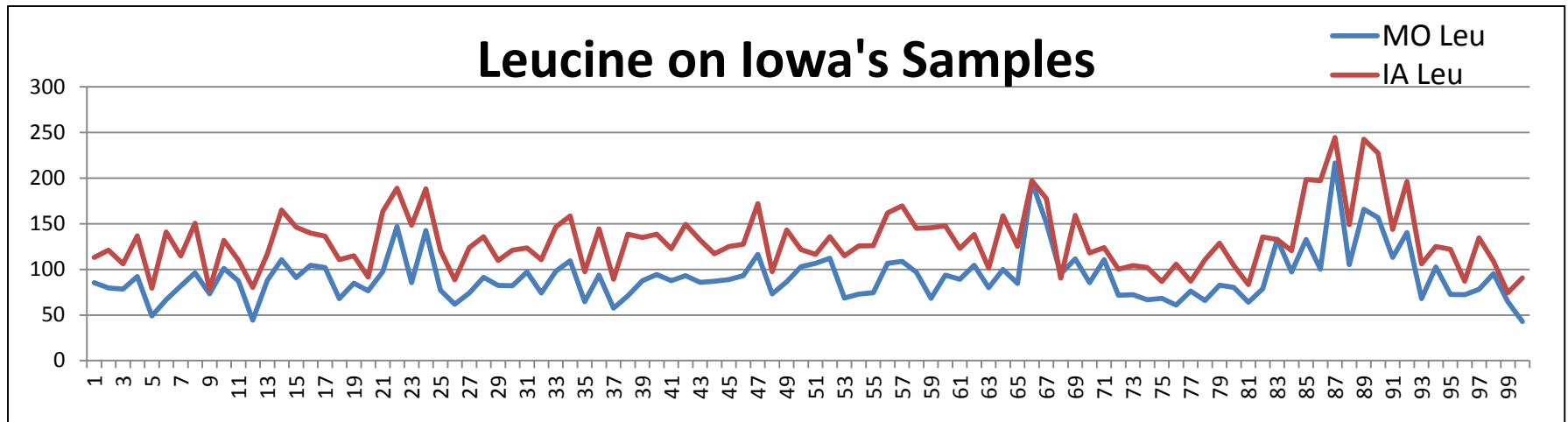
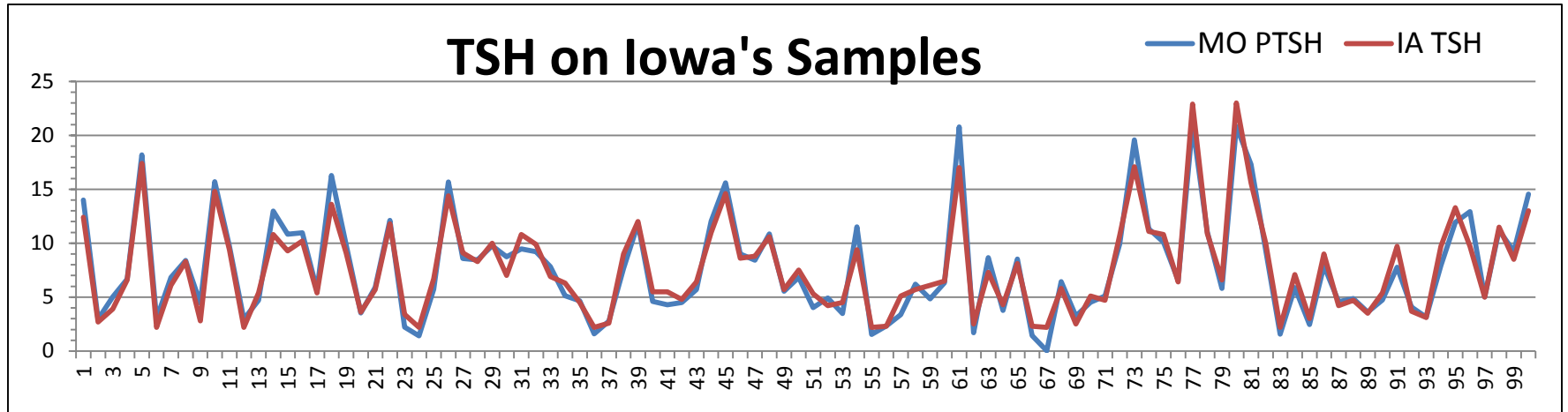
✓ **Steps we might be able to assist you with**

Samples for Method Validation

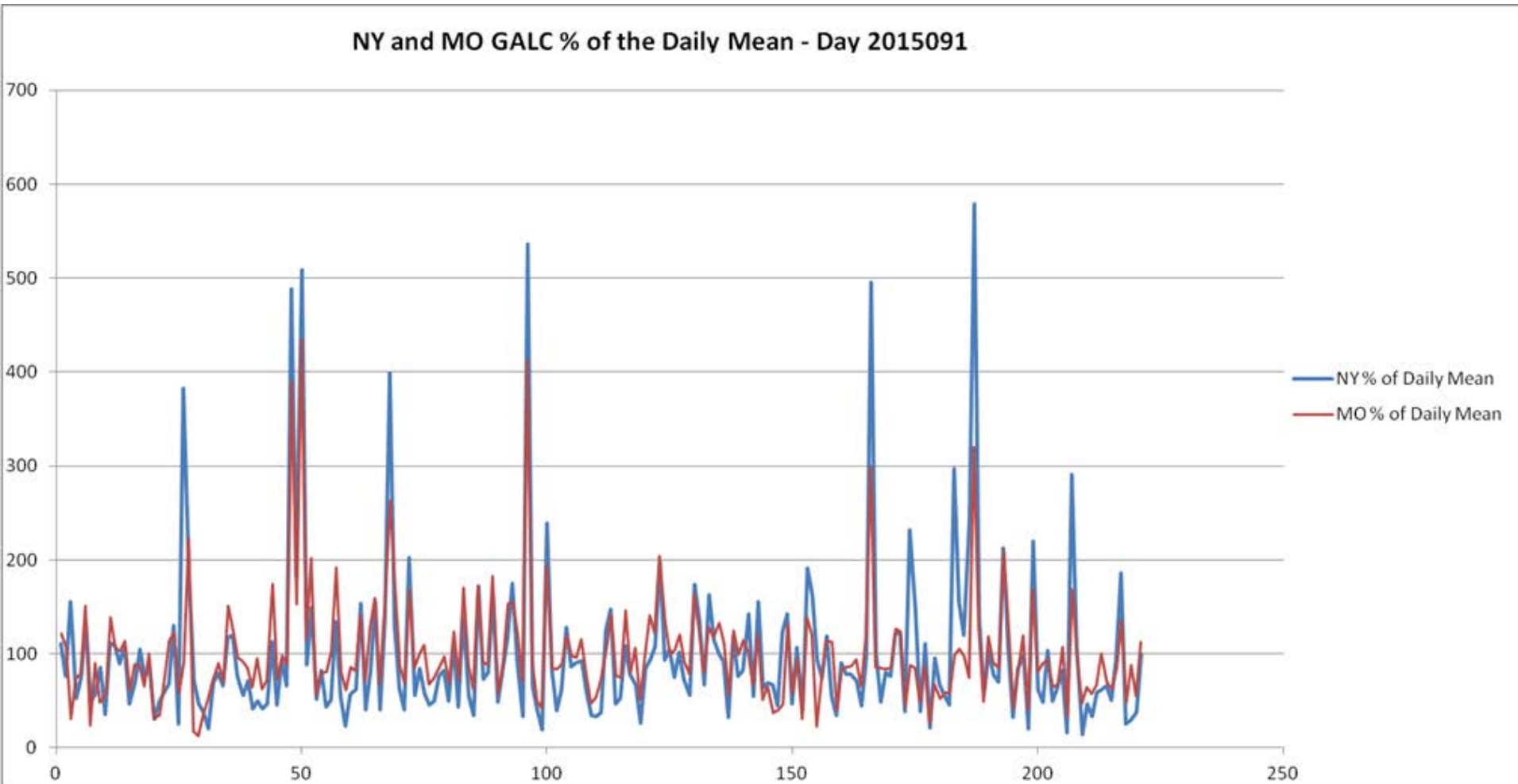


Note: We can provide them blinded or unblinded

Pre-Tested Normal Samples for Method Familiarization and Validation

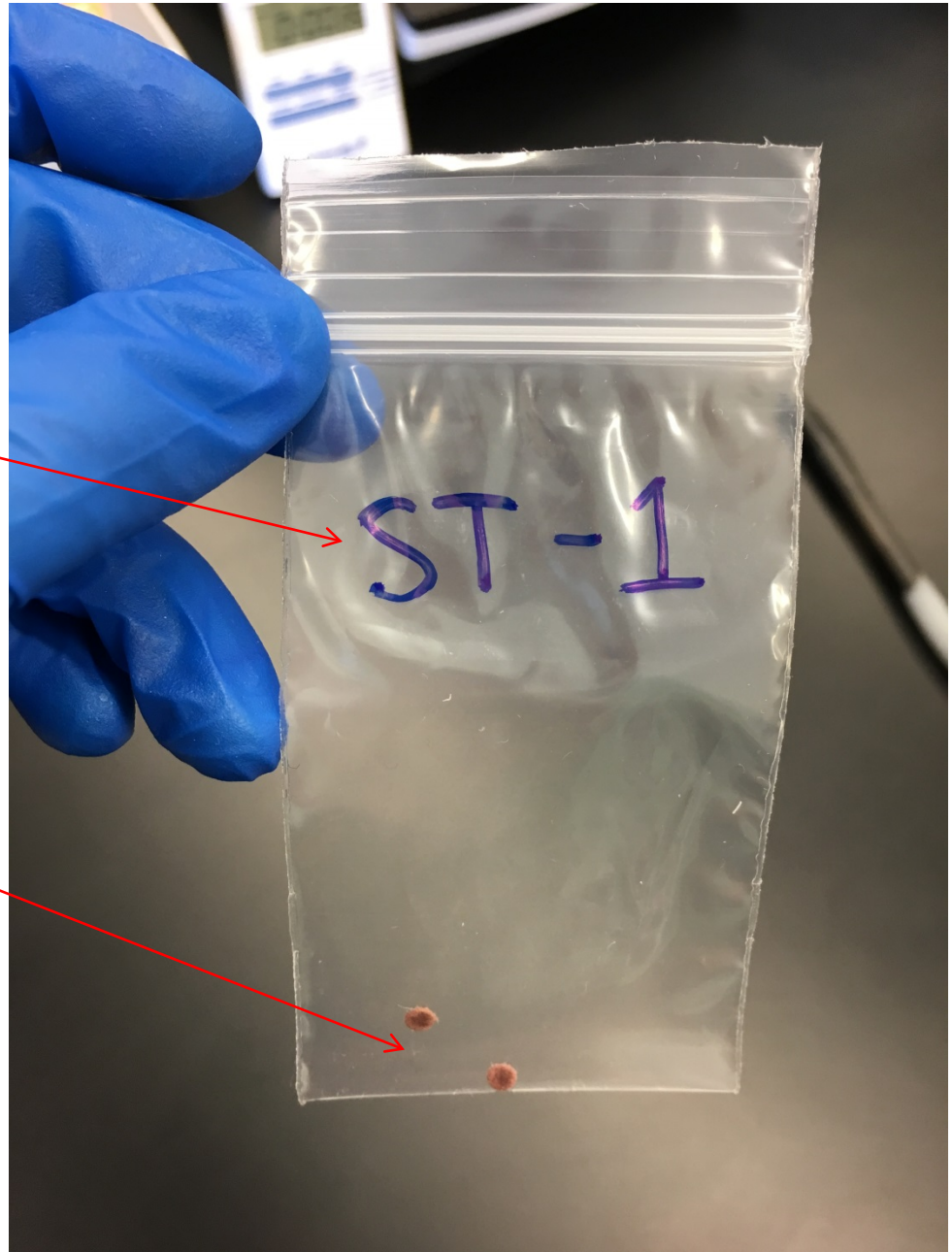


Results from Different Methods



**Your State and Sample
Starting with #1.**

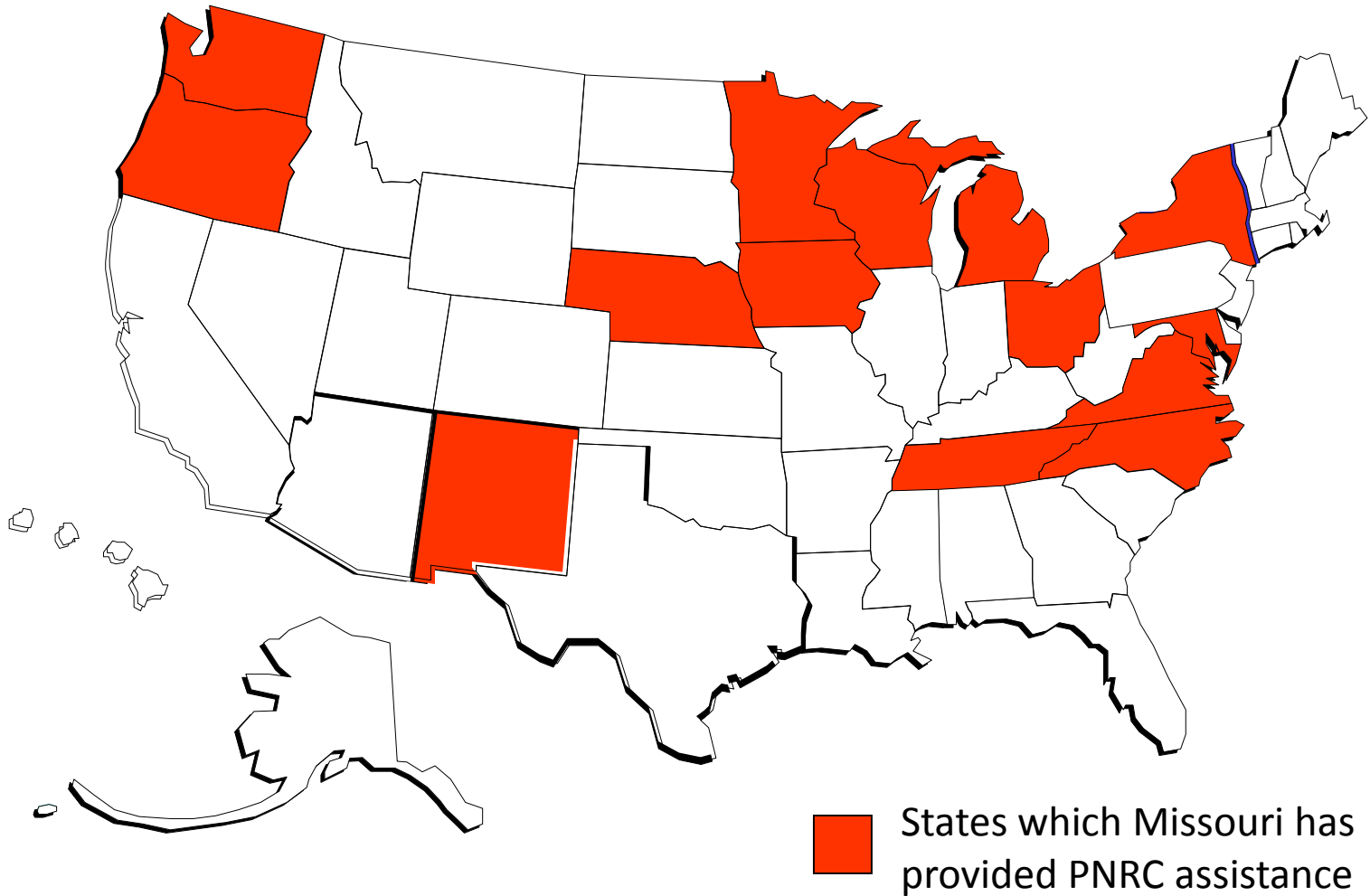
**At least two punches,
preferably three per baby**



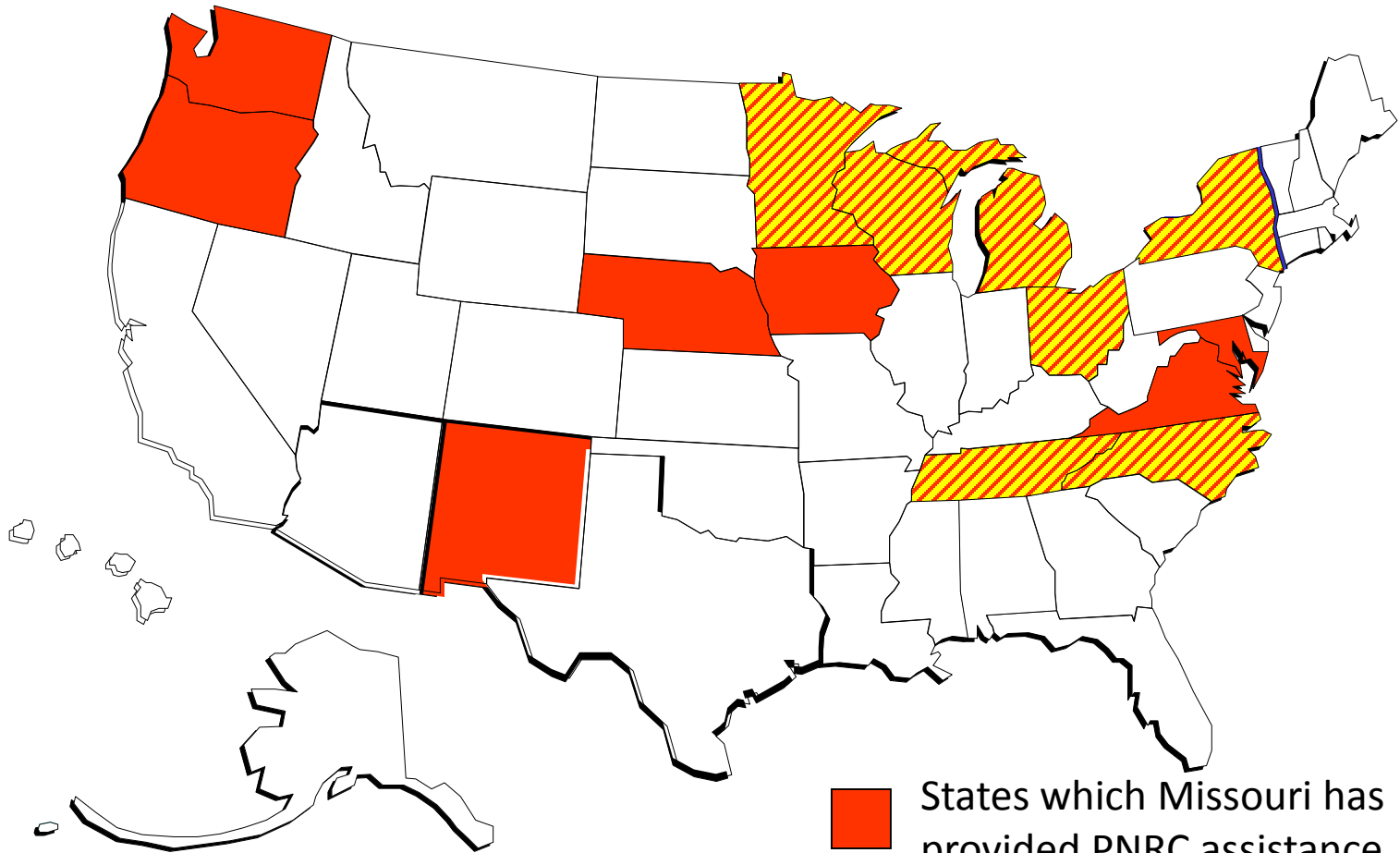
Missouri's Packet of Informational Materials



- 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



States Assisted by Missouri's PNRRC This Year



-  States which Missouri has provided PNRRC assistance
-  States which Missouri has provided DBS samples

Acknowledgements

- Tracy Klug, Lacey Vermette and the Missouri LSD laboratory team
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- The NewSTEP's Team

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Monday, August 21, 2017

