

Mountain States Genetics Regional Collaborative Center

Exchange of blood spots for educational purposes to improve quality of newborn screening by MS/MS.

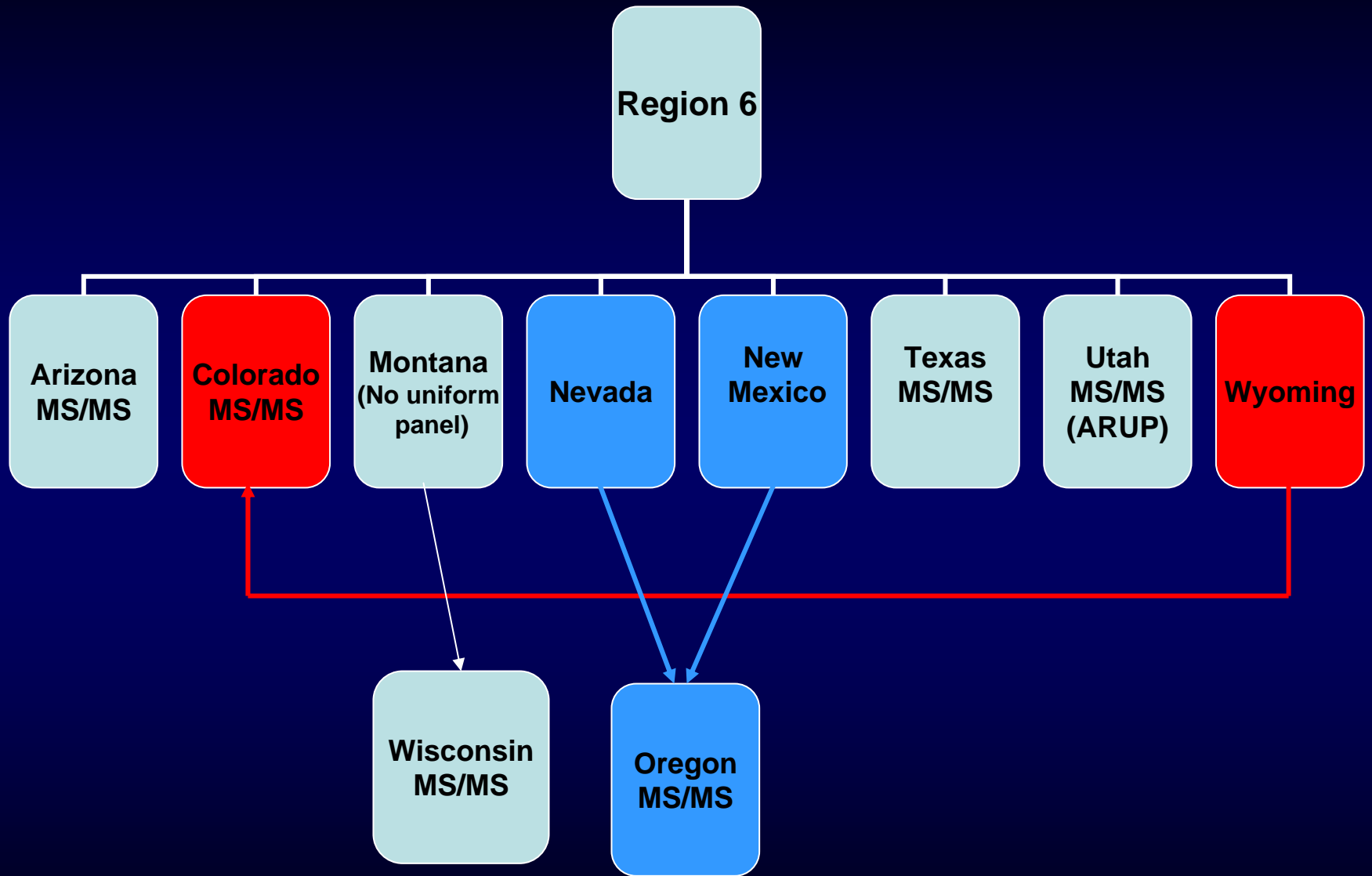
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University of Utah and ARUP Laboratories



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Background

- **Newborn screening by tandem mass spectrometry (MS/MS) has now been implemented in most states.**
- **Challenges with MS/MS cut-offs and interpretation of results have been addressed by the Region 4 Laboratory Quality Improvement activity.**



Problem/Need

- There are still some unresolved issues on how to deal with borderline/abnormal values reflecting medications, special diets, etc.
- Implementation of 2nd tier test to reduce false positives/false negatives is challenging.

How to address the problem

- **Improve recognition of abnormal patterns**
- **Promoting use of second tier tests**

Objectives

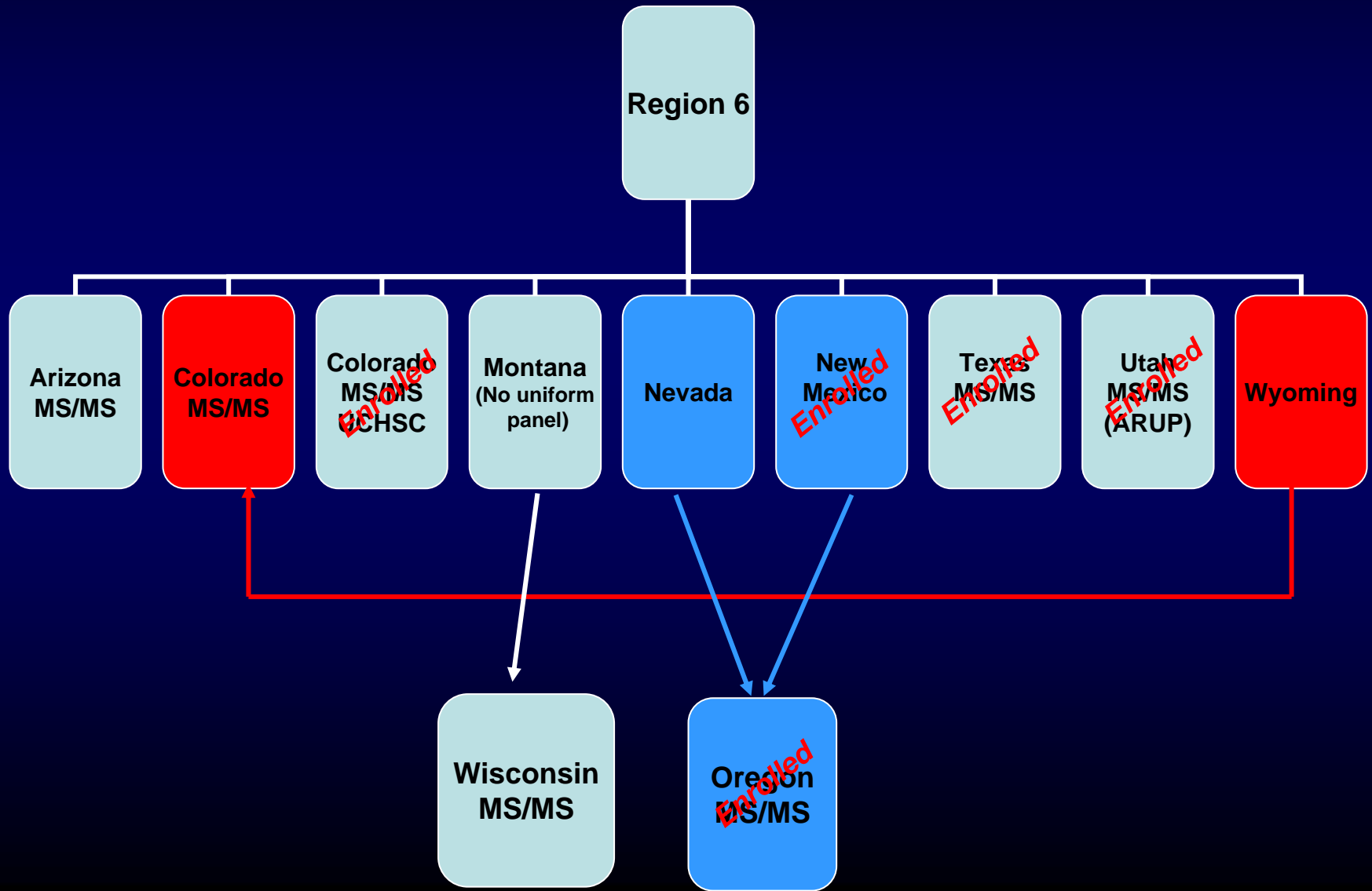
- Exchange blood spots with abnormal metabolites profile reflecting metabolic disorders or other effects.
- Provide an evaluation of results that will include information about the disease, why certain metabolites are abnormally elevated, urgency of follow-up.
- Promote the use of second tier tests to reduce the number of false positives and false negatives.

Requirements

- **Participating laboratories will be asked to:**
 - **Analyze 2-3 sets of blood spots twice per year**
 - **Complete the electronic survey**
 - **Attend one meeting/year**

Accomplishments

- First year: Enrollment of laboratories



Accomplishments

- **First year:**
 - Enrollment of laboratories
 - Development of IRB protocols to allow collection and exchange of blood spots
 - Development of questionnaires
 - Preparation of samples

Preparation of samples

- Obtained blood from the blood bank
- Processed according to CDC protocol
- Spotted on filter paper to obtain baseline values
- Spiked with analytes, spotted on filter paper
- Checked by MS/MS to see if we were on target
- Prepared the blood spots and stored them at -20 °C
- Evaluated random spots from each batch
- CV% for all analytes measured was 2.5-12.5%

Plans for 2nd year

- **September 2008 : shipment of first educational challenge**
- **December 2008 : results and educational material for the first challenge completed**
- **April 2008: shipment of second educational challenge**
- **July 2008: results and educational material for the second challenge completed**

Plans for 2nd year

- Track outcome of challenges
- Coordinate exchange of protocols for second tier tests
- Training classes
- Organize first annual meeting with the participants
- Preparation of new samples (ongoing process)

Review Panel

- **Marzia Pasquali (Utah)**
- **Nicola Longo (Utah)**
- **Steve Goodman (Colorado)**
- **Claire Leonard (New Mexico)**
- **Piero Rinaldo (Mayo/Region 4)**
- **Harry Hannon (CDC)**