

MSGRCC Newborn Screening Workgroup
Meeting Summary, Mid-Year

MSGRCC Mid-Year Workgroup Meeting – Newborn Screening
February 6, 2010
8:30pm – 4:00pm

Attendees: Kirk Aleck, Steve Amato, Becky Anderson, Sonni Aponte, Lynette Borgfeld, Jeff Botkin, Bridgette Dufour, Debra Freedenberg, Larry Goodmay, Ward Jacox, Wendie Jenkins, Joe Martinec, Marzia Pasquali, Billie Jo Peterson, Erin Rothwell, Susan Tanksley, Brad Therrell, Donna Williams, Lori Wise, Wendy Zackowicz

On conference call / webinar: Linda Beischel, Kathy Hassell, Randall Heidenreich, Denise Higgins, Carla Ortiz, Brenda Romero, Anne Seliskar, Erica Wright

Staff: Celia Kaye, Joyce Hooker, Liza Creel, Amanda Conway

Welcome and Introductions

Jeff Botkin and Susan Tanksley, workgroup co-chairs, welcomed the group and asked for introductions.

Approval of Minutes

The group agreed to accept the minutes from the July 2009 workgroup meeting.

MSGRCC Update

Celia Kaye, MSGRCC PI, presented on current MSGRCC activities. Please click here [INSERT LINK] to view the PowerPoint presentation. The following comments were made during the discussion:

- How will the RCC be involved in the Newborn Screening Translational Research Network? Celia commented that it is somewhat unclear right now. The goal of the network is to collect information that can lead to better decision making. Dr. Amato commented that this must be a two way street with participation of clinicians at the local level. Dr. Botkin mentioned that it is somewhat different from the children's oncology network because state health departments do not have treatment modalities.
- The National Coordinating Center also has a supplement on long-term follow up (LTFU). They will be distributing a questionnaire to the RCCs and states on LTFU activities and needs. Regions should include funding needs in their response.
- What is the process for any new grant opportunities? Occasionally, RCC has the opportunity to apply for supplemental funds. Currently the MSGRCC has a request in for small telehealth and distance strategies projects. RCC will circulate any federally funded opportunities that would be of interest to the region. Brad Therrell mentioned that NNSGRC typically has carryover funds at the end of the year and they look to fund projects. NNSGRC have asked to use current carryover to support Kathy Hassell's hemoglobinopathies project.

SACHDNC Recommendation on SCID

Celia Kaye gave an overview of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children's (SACHDNC) recommendation to add Severe Combined

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Immunodeficiency (SCID) to the newborn screening panel. The press release from Genetic Alliance was included in the handouts. [INSERT LINK TO RELEASE] Discussion followed:

- During the SACHDNC review, it was noted that this could create a burden on already stressed state budgets and health departments. This is especially true as follow-up must also be added and many states don't receive any state funds for newborn screening (i.e. screening fees must be increased).
- A new house bill has been proposed in New Mexico to add five new screened disorders. This could cause problems for the state because the cost is prohibitive and their current screening lab (OR) does not screen for these disorders (acid maltase deficiency or glycogen storage disease type II, globoid cell leukodystrophy, Gaucher's disease, Niemann-Pick disease and Fabry disease). A similar bill has passed in Illinois and Missouri.
- SACHDNC has reviewed several lysosomal storage disorders and has advised that more evidence is needed.
- The Texas law says that screening for disorders should occur "as funding allows". This clause means that the only way for Texas to expand is for the newborn screening program to find funding.
- Some states do not have funding from state appropriations, but only from newborn screening fees.
- RCC will send out formal SACHDNC report when available.

State Updates

The co-chairs asked the participants to provide an update on current newborn screening activities in their state and any needs that have been identified.

- Colorado – Erica Wright and Laura Taylor (written summary submitted and described below:
 - Web-based integrated data system up and running - Colorado's web-based integrated data system is now operational for newborn hearing screening data management. Currently Colorado has a paper-based system that requires all data entry be done by state health department staff. This system will allow hospital-based newborn hearing screening personnel and audiologists that see children who fail the newborn screen to record results directly into the database. This will save staff time at both ends, as well as reducing paper use. A similar system for newborn metabolic screening will be in place within the year.
 - *2009 Newborn Screening Program Report*. - This report, written by the Colorado laboratory, with input from the follow-up program, will be on the Colorado lab's website. In the meantime, a PDF of the report can be provided by email. If you are interested in obtaining a copy of the report, send your request by email to laura.taylor@state.co.us.
 - The Colorado Lab is getting new servers that will give the lab more storage and better performance. The lab will be adding a PerkinElmer "web access" feature that will allow certain entities (such as submitting laboratories and contracted subspecialists) to view newborn screening results through the Internet. The lab will pilot the system with the laboratories of the largest birthing centers. The Inherited Metabolic Diseases Clinic, the Sickle Cell Treatment and Research Center, The Division of Pediatric Endocrinology at Children's Hospital and

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Pediatric Endocrine Associates, and the CF Center, as contractors of the health department, will have access to results. There will not be access for other physicians at this time.

- PerkinElmer improved their testing kits for congenital adrenal hyperplasia. The lab started using the new kits at the beginning of December. Even with a significantly lowered cut-off, the number of abnormal screens for CAH has dropped from 4-5 *per day* to 2-3 *per week*. The large number of false positive screens for CAH has been a strain on staff resources, as well as a source of contention with providers.
- The Inherited Metabolic Diseases Clinic at Denver Children's Hospital, and the state health department have been awarded a large grant from HRSA (\$400,000 each year for three years) to develop IT solutions for improving screening, diagnosis, and short- and long-term follow-up capabilities. Utah has also been awarded this grant, so more detail may be provided in their report. The two states' goals and objectives aren't identical. An abstract of Colorado's grant can be obtained by email from laura.taylor@state.co.us.
- Arizona – Ward Jacox
 - The budget situation in the state is still very difficult.
 - Recently received a data integration grant from CDC (focus primarily on hearing screening)
 - Also received a HRSA supplement to reduce loss to follow up in the hearing program
 - Have had one false negative in CF screening
 - Have discovered that some clinicians aren't referring CF patients to CF Foundation approved clinics for sweat tests because they are out of network. The program is meeting with Medicaid and trying to educate providers on Medicaid provisions for referral.
 - Brad Therrell commented that the Mountain States region has a unique opportunity to share data on issues related to new technologies (e.g. PerkinElmer's new CAH kit). Marzia Pasquali said that she had also talked with Piero Rinaldo about collecting 2nd screen data. She offered to help identify the data that would be helpful for states. Others were also interested. Celia recommended that the MSGRCC organize a small group on data sharing and asked that anyone interested in participating let Liza know and we will organize a conference call. The following volunteered:
 - Marzia Pasquali (UT)
 - Wendie Zackowicz (AZ)
 - Lynette Borgfeld (TX)
 - Denise Higgins (MT)
 - Others to invite:
 - Dan Wright (CO)
 - Patti Ramos (TX)
 - Cheryl Hermerath (OR)
 - Norm Brown (UT)
 - Gary Halfmann (OR)

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- New Mexico – Carla Ortiz
 - The state currently contracts with the Oregon lab to screen for 26 disorders
 - Have a new information system for case management.
 - Midwives are now required to sit-in on NBS training and updated licensure test with newborn screening questions.
 - New emergency preparedness guidelines are distributed to hospitals. In the event of an emergency, patients will receive a card indicating that they need to be tested at another facility. Needs clarification. Seems a little late to me.
 - State currently stores samples for one year, and then they are destroyed.

- Utah – Jeff Botkin
 - One year since implementation of CF screening and more kids have been identified than predicted. The kids are sicker than predicted as well. The state has also had two false negatives and will be reviewing the data soon. They expect to meet soon to discuss sending kids for sweat testing based on high IRT levels.
Questions from the group:
 - Is there a possibility that there is a unique mutation in the population?
 - Are the clinicians getting a full CF sequencing for those children not detected?
 - Texas, Colorado and Utah are conducting a roundtable at the May APHL NBS Symposium to share strategies on CF screening including their different approaches using IRT-IRT-DNA testing.
 - State is continuing Clinical Health Information Exchange (CHIE) now with newborn screening information (via HRSA effective follow-up grant).
 - New bill proposed that would provide support for families that don't have insurance reimbursement for nutritional supplements.

- Wyoming – see Colorado notes

- Montana – Denise Higgins submitted a written report (below)
 - Montana expanded screening to include the 29 nationally recommended tests (including hearing) in January of 2008. The Montana legislature provided some funding for a follow-up program to serve for infants identified through the screen and their families. Shodair Children's Hospital in Helena was awarded the contract following a competitive bid.
 - Newborn Screening Program highlights for 2009 include:
 - Development of contracts for pediatric endocrinology, pulmonology, hematology and a metabolic sub-specialist.
 - Hosted Montana's first "Metabolic Day" in August of 2009. Approximately 40 patients, family members and providers participated; the day provided opportunities for family to family sharing, lectures, cooking demonstrations and recipe sharing, as well as activities for the children. The second Metabolic Day will be August 14, 2010.
 - Launching a Facebook site to facilitate communication between families.
 - Provision of educational opportunities re NBS for primary care practitioners and nurses at professional conferences.

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- Expansion of NBS clinics to include Cystic Fibrosis clinics. Montana contracts with three health care facilities to provide the basis for regional clinics – we do not have a medical school.
- Transfer of Newborn Screening Follow-up coordinator into the Laboratory to facilitate short-term follow-up activities
- IRB exemption from Montana IRB to participate in Utah 2nd-Tier testing project
- Utilizing new PerkinElmer CAH kit for screening through Wisconsin State Laboratory of Hygiene.
- Barriers encountered in newborn screening program
 - Travel restrictions at State level, that most likely will not change in the near future, limit the participation in some activities. Opportunity for MSGRCC to explore alternative ways to offer interactive participation
 - Lack of funding to offer courier services to all birthing facilities across the state. Currently only 80% of birth facilities receive courier services
- Texas – Debra Freedenberg
 - Hiring freeze currently in place for nonessential positions.
 - All state agencies have been asked to look at how they would handle a 5% budget cut.
 - Emergency preparedness program is ongoing.
 - Have a new LTFU database where at least one hospital is submitting demographic data via HL-7 transfer. Others submit reports online. They receive approximately 5-6% of demographics electronically.
 - Currently working on full implementation of state laws related to nbs and blood spot storage. There has been increased press related to the 2009 lawsuit. As part of lawsuit settlement, state will destroy all previously stored spots. Moving forward, parents can opt out of the storage. The state is receiving more opt out requests than expected. It is taking about 5FTEs to process.

Short Term Follow-up Presentation

Brad Therrell presented on the short-term follow up recommendations of the SACHDNC long-term follow up subcommittee. Click here for his PowerPoint presentation [INSERT PPT]. A summary follows:

- Should newborn screening conditions be reportable? The consensus of a small workgroup was “no”; however, this may be revisited at some point.
- The LTFU committee recommended to SACHDNC that SACHDNC write a letter to the Secretary of HHS with the following recommendation: The newborn screening serial number should be included as a required field on the standard national birth certificate. SACHDNC discussed at length, and the outcome was that Colleen Boyle will work on a small position paper in support of this recommendation, which will come back to SACHDNC and then to the Secretary. It was noted during the discussion that 10 states currently have the nbs serial number on the birth certificate, and 4 others are planning to add it. Texas formerly had the serial number and stopped doing this in 2005.

Discussion on NNSIS Data Submission

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Brad Therrell gave an overview of the NNSIS system and its history. The purpose of the database is to have data to back up federal budgets for newborn screening services and for state budget requests. In addition, it can be helpful for state quality assurance efforts. Last fall the RCCs were asked to work with states to identify barriers to timely data submission. Most states are now current. States currently submit data under their own definitions. The NNSIS system is part of the new Newborn Screening Clearinghouse but the actual linkages are currently unknown. Discussion was opened and is summarized here:

- There is still a need for consistent definitions, especially when comparing to other states.
- There will be a special session on NNSIS and data issues on the final day of the May Newborn Screening Symposium. The group requested that the session be broadcast via webinar so those unable to travel can still participate.
- The following data/information is needed:
 - Positive Predictive Values
 - Standard on what to report when a marker can indicate more than one disorder
 - How to report positive test results vs. actual cases
 - An understanding of how this data relates to the data being submitted to Region 4
 - An understanding of use of “not normal” report
 - Improved definitions so states can compare apples to apples
- States also identified a need for more data automation and resources to improve submission.
- The data sharing subgroup will explore identifying consistent definitions and central data points for the region.
- Dr. Amato asked if CDC still has public health physicians that will visit states to help with these issues. Dr. Therrell indicated that they do, but they aren’t typically used for newborn screening.
- The LTFU survey that will be conducted by the National Coordinating Center will ask states what they need to fulfill these data submission requirements.

Lunch Presentation

During lunch, Jeff Botkin showed a film the University of Utah’s Promoting Public Dialogue on the Use of Residual Newborn Screening Samples Project. The video is being used to introduce focus group participants to newborn screening blood spot storage issues.

Update on Other Projects in the Mountain States Region

Celia Kaye asked for updates from participants in other newborn screening and genetics projects in the region. The following projects were presented:

- Colorado Sickle Cell Care Network – Kathy Hassell [ATTACH ABSTRACT]
- Texas Newborn Screening Performance Measures Project – Susan Tanksley [LINK TO PPT]
- Evaluation and Implementation of Second Tier testing for Disorders Identified by MS/MS in Newborn Blood Spots in the MSGRCC – Marzia Pasquali [ATTACH ABSTRACT]
- Utah Newborn Screening Clinical Health Information Exchange – Billie Jo Peterson [ATTACH ABSTRACT]

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- Promoting Public Dialogue on the Use of Residual Newborn Screening Samples – Jeff Botkin [LINK TO PPT]

Evaluation Update

Liza Creel presented on first year data from the National Coordinating Center's (NCC) evaluation.

Next Steps

- Send out hours of operation for region's newborn screening labs to the full group
- Work with Marzia Pasquali to organize a presentation on her analyte stability project during the 2010 Annual Meeting
- Create a better linkage between the NBS Workgroup and the Genetic Alliance
- Organize and schedule first call of the data sharing subgroup
- Encourage participation in May NNSIS meeting via webinar
- Distribute LTFU survey from the NCC when available
- Explore expansion of Texas Newborn Screening Performance Measure Project
- Send evaluation report to full workgroup
- Ask for posters from regional projects for the Annual Meeting

The meeting was adjourned.

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