

TNSPMP MEETING AGENDA AND NOTES



TNSPMP PROJECT TEAM MEETING
 DRURY INN & SUITES - SAN ANTONIO
 FRIDAY - JUNE 27, 2008
 TNSPMP FACILITATOR: ROBIN SCOTT
 MEETING NOTES

ATTENDEES:

<i>Sandra Billings</i>	
<i>George R. Buchanan</i>	
<i>Kari Casas</i>	√
<i>Donna Claeys</i>	√
<i>Robert Crumb</i>	
<i>Margaret Drummond-Borg</i>	√
<i>Alice Gong</i>	√
<i>Jose L. Gonzalez</i>	
<i>Charleta Guillory</i>	√
<i>Cheryl Hermerath</i>	√
<i>Scott D. McLean</i>	√
<i>Francisco Ramirez</i>	√
<i>John Saito</i>	√
<i>Stuart K. Shapira</i>	√
<i>Eileen Sheridan-Shayeb</i>	
<i>Reid Sutton</i>	√
<i>Larry Sweetman</i>	√
<i>Lois Taylor</i>	√
<i>Brad Therrell</i>	
<i>Sister Mary Nicholas Vincelli</i>	√

<i>Morgan Walthall</i>	√
<i>Don P. Wilson</i>	√
<i>Jerald L. Zarin</i>	√
<i>Margaret Bruch</i>	√
<i>Sherry Clay</i>	√
<i>Mirsa Douglass</i>	√
<i>Eldridge Hutcheson</i>	√
<i>Daisy Johnson</i>	√
<i>David R. Martinez</i>	√
<i>Jann Melton-Kissel</i>	
<i>Susan Neill</i>	
<i>Sharon Newcomb-Kase</i>	√
<i>Susan Tanksley</i>	√
<i>Simran Tiwana</i>	√
<i>Donna Williams</i>	√
<i>Susan Snyder</i>	
<i>Lisa Kalman</i>	√
<i>Colleen Buechner for NNSGRC</i>	√

TIMELINE

- 07/03/08 Send TNSPMP Round Table discussion notes to Mirsa Douglass
- 07/17/08 Distribute the TNSPMP June meeting minutes and Literature Review survey questionnaire
- 07/25/08 Deadline to provide comments and input on TNSPMP June meeting minutes
- 09/04/08 TNSPMP quarterly meeting in Austin begins (DSHS, Boardroom)
- 09/28/08 TNSPMP TX PEAT drafted with outline and high level concepts by end of first year project period



ROUNDTABLE DISCUSSIONS ON CAH

After Simran Timvana presented findings on Congenital Adrenal Hyperplasia as it relates to time to treatment, roundtable discussions took place to suggest performance measures.

Comments are grouped by discussion table.

The following suggestions were provided by Charleta Guillory, Don Wilson, Donna Williams, Yvonne Caimanque, Judy Chrisman, John Saito, and Morgan Walthall.

Suggested Performance Measures

- The number of facilities entering both birth date and time
- The number of facilities entering birth weight
- Time to receipt in the laboratory for facilities with long transit times; consider surveying follow up activities to pinpoint the problem
- Time from collection station to mailroom
- Time from mail pickup to receipt in laboratory
- Time it takes for mom to bring baby for follow-up after contacted
- The number of children with CAH positive screen referred to a pediatric endocrinologist
- The length of time children have to wait for an appointment with a pediatric endocrinologist
- The time from birth to treatment
- The time from birth to confirmation
- The number of physicians following standard procedures in caring for child with abnormal screen e.g. Endocrine Society, American Academy of Pediatrics
- The number of specialists following standard procedures in caring for child with abnormal screen e.g. Endocrine society, American Academy of Pediatrics
- The number of pediatric endocrinologists in Texas per health region; focus on regions with higher CAH rates
- The number of children diagnosed with CAH still on appropriate treatment after x number of months/years
- The number of infants who were already on therapy before screen result received (In Texas, approximately 7 out of 15 cases in the last few years were already on therapy before screen result was provided)

General Comments

- Consider different laboratory cut-offs for male and female
 - Capture gestational age on collection cards
 - Specify definition of “age of life”. Vital statistics considers 0 to 24 hours of life as day zero
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- We need to know Texas data on mortality of CAH

The following suggestions were provided by Susan Tanksley, Scott McLean, Stuart Shapira, Lisa Kalman, Francisco Ramirez and David Martinez.

Suggested Performance Measures

- Average age of diagnosis and distribution of ages when diagnosed
- Time confirmatory laboratory results available
- Time it takes for medical personnel or responsible party to receive results
- Time to clinical contact with family and physical evaluation
- Time to treatment;
 - track date infant put on treatment
 - tracking should be done by staff in hospitals, by PCPs (Primary Care Physicians) or endocrinologist
- Track long term indicators such as compliance with treatment
- Track mortality
- Track the time when specimens for confirmatory testing are submitted and the time when confirmatory results are received
- For pre-analytical, track the age of the child at time of collection; if ambiguous genitalia were present; time to receipt at laboratory; and time to start of analysis at laboratory

General Comment

- Link performance measures to ACT sheet

The following suggestions were provided by Margaret Drummond-Borg, Alice Gong, Sherry Clay, Sister Mary Nicholas Vincelli, Donna Claeys and Jerald Zarin.

- Track the initiation of treatment by 7-10 days of age [age as defined by Bureau of Vital Statistics] before symptoms are present
- Identify age at initiation of treatment by gender
- Develop data collection for tracking and refinement of goal
- Track continuation of treatment at least through high school
- Track genetic/nutritional counseling at milestones where diet changes (such as moving from formula to solids, toddlers, high school)

The following suggestions were provided by Colleen Buechner, Sharon Newcomb-Kase, Margaret Bruch, Mirsa Douglass, and Simran Tivana.

- Measure quality of parent education as this is crucial for infant care
 - Measure intelligence quotient and track dietary compliance
 - Measure survival statistics
 - Age at death
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- Collect data on
 - Rate of false positives
 - DNA confirmation
 - Number of infants with symptoms
 - Type of symptoms observed

The following suggestions were provided by Reid Sutton, Eldridge Hutcheson, Larry Sweetman, Cheryl Hermerath, Lois Taylor, and Kari Casas.

- Accurate date and time of both birth and sampling of each baby is critical to assessing the measures below and must be implemented
 - Time in day of life or hours of life to specimen arriving in the Austin DSHS laboratory
 - Time in day of life or hours of life to abnormal NBS result
 - Time in day of life or hours of life to abnormal NBS result called to the medical doctor
 - Time in day of life or hours of life to patient contact
 - Time in day of life or hours of life to physician/laboratory appointment for evaluation and confirmatory testing
 - Time in day of life or hours of life to being on appropriate medications
 - Percent on treatment at the time of positive newborn screening result in DSHS laboratory
 - Percent of classical/salt wasting CAH hospitalized or on medical treatment at the time of abnormal newborn screening result

ROUNDTABLE DISCUSSIONS ON GALACTOSEMIA

After Simran Timana presented findings on Galactosemia as it relates to time to treatment, roundtable discussions took place to suggest performance measures.

The following suggestions were provided by Charleta Guillory, Don Wilson, Donna Williams, Yvonne Caimanque, Judy Chrisman, John Saito, and Morgan Walthall.

Suggested Performance Measures

- The number of infants confirmed with diagnosis who were referred to a dietician
- The number of infants confirmed with diagnosis who have access to a dietician
- The number of infants hospitalized with septicemia

General Comments

- Consider conducting a statistical analysis of Galactosemia incidence comparing pre and post differences of screening for total galactose-1-phosphate to galactose-1-phosphate uridylyltransferase
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- Put color sensor ink dot on collection cards to determine if specimens were exposed to excess heat

General notes on literature searches

- If possible, try to include natural progression of disorder without screening
- Look at the American College of Medical Genetics report on 29 disorders (online)
- Go to various websites to review the latest information on disorders:
www.genereviews.org, www.genetests.org, www.geneclinics.org

The following suggestions were provided by participants Susan Tanksley, Scott McLean, Stuart Shapira, Lisa Kalman, Francisco Ramirez and David Martinez.

- Use similar measures as suggested in the CAH round table discussions
- Age of death (sepsis or renal failure, or other major system failures that contribute to mortality)
- Age when soy diet was started
- Age at conference with dietician
- Time to DNA testing
- Time of genetic counseling
- Time of nutritional counseling
- Frequency of developmental evaluations (e.g. IQ at 1 yr, 2 yr etc)
- Long term outcomes need to be studied comparing treated versus untreated patients

The following suggestions were provided by participants Margaret Drummond-Borg, Alice Gong, Sherry Clay, Sister Mary Nicholas Vincelli, Donna Claeys, and Jerald Zarin.

- Short-term
 - Track initiation of treatment by 7 to 10 days of age [using age as defined by Bureau of Vital Statistics] before symptoms are present
 - Identify age at initiation of treatment
 - Develop data collection for tracking and refinement of goal
 - Long-term
 - Gain a better understanding of long term impact of patient outcomes for Galactosemia disorder. For example, follow and study intelligence quotients; compare intelligence quotients with age of treatment and duration of treatment to evaluate relationship between variables
 - Track how closely a galactose-free diet is followed
 - Consider genetic/nutritional counseling at milestones where diet changes (such as moving from formula to solids, toddlers, high school)
 - Need to understand diet options at all life stages
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- Identify how many children diagnosed with Galactosemia are referred to Early Child Intervention services
- Conduct an annual check by case management for information gathered on...
 - Compliance with galactose-free diet
 - Referrals to Early Child Intervention services
- Follow up on the number of children followed by a metabolic specialist through high school

NEXT STEPS ON PERFORMANCE MEASURES AND LITERATURE REVIEWS

Mirsa Douglass discussed next steps for performance measures and showed an overview of options for stakeholders who wish to participate in literature review activities.

Points made during discussion are shown below.

- Conduct literature review on fatty acid disorders to present at next meeting
 - Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
 - Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
 - Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
 - Trifunctional Protein Deficiency (TFP)
 - Carnitine Uptake Deficiency (CUD)
 - Carnitine Palmitoyl Transferase Deficiency1 (CPT1)
 - Send surveys to stakeholders team members requesting assistance on various literature review aspects
 - Recruit additional ad-hoc subject matter experts to attend TNSPMP meetings
 - Expand literature review efforts to students, fellows, and residents as they are available
 - Review Up-To-Date documents for disorders of interest prior to next TNSPMP quarterly meeting. Up-To-Date is an electronic resource which provides knowledge for clinicians and patients for a multitude of medical topics, including an overview of the classification and evaluation of inborn errors of metabolism
 - Send suggestions for presenters at face-to-face meetings. Dr. Gibson was suggested to present on fatty acid oxidation disorders with an emphasis on MCAD at the next meeting, September 4th
 - Contact authors from the Up-To-Date series to consider participating as a reviewer for newborn screening disorder literature reviews, or any recommendations made by the TNSPMP team
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MEETING PLUS/DELTA (FEEDBACK)

Participants shared thoughts about what they liked and didn't like about the meeting.

CHANGES

- No burnt restaurant
- Pre- meeting articles
- Bigger room
- Begin first day later with an organized dinner
- End earlier than 3pm on Friday. Consider 8-Noon
- Start earlier on Thursday
- Have laptops at table for entering notes for round table discussion

POSITIVE FEEDBACK

- Round table set-up
- Refreshments
- Meeting at hotel
- Having reference articles handy
- Convenient to airport
- Free shuttle
- Great discussion
- Dr. Gonzalez's presentation (bats)
- Free breakfast
- Mirsa Awards