



Handouts:

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|----|------------------|----|-------------------|----|------------------|----|--------------|
| #1 | Consented Report | #3 | Completed Surveys | #5 | MMA-HCY Interval | #7 | HCY Interval |
| #2 | Summary of Cases | #4 | Survey Status | #6 | HCY Intake | #8 | ROI Comments |

I. **Welcome and roll call:** (Arkansas (0), Illinois (Jan Askren) , Indiana (0), HRSA (0) , Berry
Kentucky (0) , Michigan (Alison Bannick, Jerry Feldman), Minnesota (Sue Berry,
Kristi Bentler, Carolyn Anderson) , Missouri (Peck) , Nebraska (0) , New Jersey (0) ,
New York (Richard Erbe, Eileen Blakely), Ohio (Nancy Leslie), Oklahoma (0) ,
Pennsylvania (0) , South Dakota (Quinn Stein, Amy Mrock), Wisconsin (0) , Region 4
staff (Sally Hiner, Maria Ostrander), other (0).

II. **IBEM-IS**

A Data Reports Berry

. Berry reminded the group of the need for consistency when reporting free-text and numeric values for cases. If there are NBS values other than a numeric values being received, please share this information with Berry & Bentler as it affects how data sets are grouped together. Berry will send out a brief survey on whether or not this is a problem in individual states. The group can then figure out how to work around it.

1. Consented (Handout #1) Ostrander
Ostrander reviewed numbers consented log with the group and encouraged members who have not yet submitted information to do so before the end of next week.

2. Summary of cases (Handout #2) Berry
Berry reviewed the graphs of the summary of cases from January – December 2010. The amount of data available continues to grow.

3. Completed surveys by entering clinic (Handout #3) Berry
Bentler provided an overview of how condition surveys are developed. If anyone is having issues/questions please contact Bentler. Goal is to review GA2 and Melonic surveys before or during the next conference call and determine which others to focus on after that.

4. Survey status report (Handout #4) Bentler
There are a total of 32 IBEM surveys completed and available for data entry; including 25 disorder specific intake surveys completed and 22 disorder-specific interval surveys. Please see handout #4 for a complete listing of surveys and their status (completed and available, pending, etc.).

B. Review data elements Bentler /Group

1. Interval MMA-HCY (Handout #5)
Bentler—reviewed interval survey updates with the group. A specific set of ophthalmology questions, mental health concerns, and other lines of questions were built into the survey. Bentler asked members to look at Handout #5, review the scope of changes, and provide input for additions.

Home monitoring questions were deleted. Urine homocystine will be added

into the biochemical line of questions.

Members suggested that the monitoring of Plasma MMA would be helpful to add.

Questions related to spinal and vascular events were discussed. Bentler asked the group if there were other specific imaging questions members anticipated.

Under Pharmacotherapy, Bentler asked the group if there is a need to add aspirin as a treatment option for tracking purposes. Members agreed to leave it in. Other medications also were added. In terms of Nutrition questions, standard protein restriction questions were maintained.

Berry asked the group to submit any final comments by the middle of next week.

2. HCY

a. Intake (Handout #6)

The group was asked to pay specific attention to MTHFR in handout #6 and submit comments/insights to Bentler. Bentler reviewed changes with the group. The value of Platelet count and diagnosis questions was discussed. Members decided to take it out. A question about maternal lab studies was added, as well as a plasma question.

While the focus is on NBS, members can enter adults in this process for those who might present symptoms. Berry asked the group to review and provide final comments by next week.

b. Interval (Handout #7)

The group discussed the question on blood pressure for older individuals presenting symptoms. Eye findings questions were also briefly reviewed. The home monitoring question was deleted from this survey. Specific biochemical testing questions were added.

A question on the imaging of vascular events was added. The medications and supplements section was reviewed with the group. A nutritional question related to Methionine intake was added.

III. RO1 Reviews (Handout #8)

Berry informed the group that comments on the RO1 grant application have been received, indicating “substantial enthusiasm” from the review board. Cameron and Berry have learned that we currently have the best score. Berry added that this could change the tenor of how we conduct our face-to-face meeting if funded for another 5 years. Please review comments (handout #8) for more detail. Hiner will post materials to the R4 website under the meeting date.

Berry

IV. Updates

Berry

A National

Berry indicated that the R4 workgroup has had substantial influence on emerging national databases. The group is looking at 80% common elements, in order to ensure we have chosen the right form and answer to the nature of the questions—based on current surveys. Berry asked Amy Brower to send questions for review to determine if those questions correspond to the P2 set of survey questions.

Berry added that the group will need to continue entering data w/DocSite (Covisint) for some time before the national dataset is up and running. The target date for project completion is summer 2011.

B. Regional

Hiner
/Ostrander

Ostrander reminded members to submit numbers consented/declined/withdrawn, if they have not already, and that for those clinics that qualify for case enrollment reimbursement, to submit their invoices to Patty Losey at MPHI by January 15, 2011.

Hiner informed the group that Maria Ostrander has accepted a new position (effective 12/29/10). Brenda Dietrich will provide support to members.

C Abstracts

Berry

1. SIMD: The abstract has been accepted for poster presentation. When the draft is available, Berry will send it out to members for their review and comments.
2. ACMG: Berry added that we should see part of this supplemental issue coming out in the next few weeks.

V. Training

Berry

Sue Berry will be providing another guided training on the use of the IBEM-IS on Wednesday, January 12, 2011 from 3 – 4:15 PM (CT) / 4 – 5:15 (ET). Details will be distributed as the date draws closer.

VI. Face to Face Meeting

Berry/
Hiner

Hiner sent out a Doodle poll to gauge P2 Workgroup members' availability. Members are asked to review dates and respond. Hiner added that the typical meeting runs from 9:30 AM – 4 PM. In the past, the meeting has been held at O'Hare Airport Hotel in Chicago; however, Minneapolis and Detroit are other options. Typically, 1 clinician per clinic is supported by R4. Travel for members outside of Region 4 is covered under their respective agreements. Berry asked the group to also think about data mining opportunities that could be discussed at the next meeting should the R01 funding come through.

VII. Announcements & Adjourn

Group

Berry asked the group if there were any other questions, comments or concerns they would like to raise.

Berry reminded the group about the need for consistency when entering data in the surveys.

Nancy Leslie asked the group if anyone had experience submitting an IRB request to enter retrospective data without consent on a patient who has passed away. Nobody reported prior experience, but as long as no personal identifiers existed, members thought it may be acceptable to their IRB panel. Leslie will report back to the group on her efforts to obtain an amendment to their IRB.

Meeting adjourned at 2:56 PM.

Next meeting is scheduled for Friday, January 21, 2011 2 – 3 PM ET.

Notes by Hiner and Ostrander