



Handouts:

#1 Summary of Cases	#2 Completed Surveys by Entering Clinic
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I. Welcome & Roll Call – by State & HRSA

Participating: Arkansas (0); Illinois (0); Indiana (Kathy Wood); Kentucky (0); Michigan (0); Minnesota (Kristi Bentler, Missouri (0); Nebraska (0); New Jersey (Rhonda Plager sp?, Karen Gonzales); New York (Richard Erbe, Eileen Blakely); Ohio (Nancy Leslie); Oklahoma (0); Pennsylvania (0); South Dakota (0); Wisconsin (Sandy van Calcar); HRSA (0); Region 4 staff (Sally Hiner, Maria Ostrander); Other (0).

Rhonda Plager of New Jersey has a Clinical Genetics Department of 1.5 geneticists 8 genetic counselors. They look forward to participating in the workgroup.

Richard Erbe of New York indicated that their clinic is in the process of submitting their IRB application, and will be ready to go as soon as IRB approval received.

Eileen Blakely of the NBS follow up center for NY, indicated that they have submitted their IRB application and are hoping to receive expedited review/approval in the coming week.

Berry welcomed the new members and encouraged them to contact either her or R4 staff for support.

II. IBEM-IS

A. Data Reports (Handouts 1 & 2)

Two reports have been prepared: a linear list of cases by disorder, and graphs of cases added. Down tick in cases entered could be related to the time that went into preparing for the RO1 grant.

B. Enrollment reports and invoices for Quarter Ending November 30

Ostrander sent out a reminder on 11/15/10. Follow-up reminders and updates will follow.

Hiner reminded all participants to please submit numbers as those numbers are used for national tracking as well. Ostrander will send reminders quarterly.

C. Ongoing QI

- **Data entry concerns**

Berry indicated that there have been inconsistencies with how the case number is being determined. This element is in two places. It is the first element on the enrollment survey and is entered again on the demographic page. Ostrander will send out a reminder and the link to how to assign the unique patient identifier to group members.

If members are having problems w/ access or data entry, they are encouraged to contact Berry or Bentler directly.

- **Data element concerns**

None

- **IRB concerns**

None

- **Other**

None

D. Review of list of disorders needed to complete the surveys and request for participation

MMA + Hcy Interval - pending

- **The following surveys still need development**

Arginemia
Argininosuccinate acidemia (ASA)
Citrullinemia Type I (argininosuccinate synthetase)
Citrullinemia Type II (citrin deficiency)
Homocystinuria (all combined - Cbl G, Cbl E, CBS, etc)
Hypermethioninemia -
Defects of bipterin cofactor biosynthesis/regeneration
Hyperphenylalaninemia/phenylketonuria

Dienoyl-CoA reductase deficiency
Glutaric acidemia Type II
Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
Medium/Short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD)

Malonic Acidemia

Galactokinase (GALK) deficiency galactosemia
UDP-galactose-4-epimerase (GALE) deficiency galactosemia

Berry solicited group opinions/ideas on working on enrollment/interval surveys for remaining disease-specific disorders. The original goal was to finish the list of disorders by end of the 5 yr. grant cycle. It looks like we may accomplish this early, by the end of 2011.

Members interested in working on any of the surveys should contact either Berry or Hiner. Leslie volunteered to assist with Malonic Acidemia, as they recently have seen a patient with the disorder.

E. DocSite Update

DocSite is now Covisint, a subsidiary of Compuware. Julia Flanagan remains our technical support contact. John Haughton has moved to a different position, and we are waiting for a new administrative contact to be identified.

Hiner was able to glean from Flanagan that the cost of a site license pays for the following: 1) access to the information system; 2) implementation; 3) survey site set-up; 4) standard releases to updates & software; 5) access to user manual; 6) any and all standard training materials.

Hiner and Flanagan are exploring ways to address funding issues, including looking into the possibility of splitting site-license costs for some of the non-R4 clinics who may enter smaller numbers. There are some HIPAA/IRB concerns that would have to be addressed, but more information will follow.

The problem with the data exporter capabilities of Docsite has been addressed. It will now allow for the export of data from multi-check boxes, instead of having to go into files one-by-one as previously.

III. Updates

A. NIH Grant Submission

Our collaborative proposal received a score of 20. Berry & Cameron have sought information on the scoring of proposals to ascertain how we ranked compared to other submissions. Of the 11 that went into the pool, ours was highest. Berry remains optimistic that this may go forward, and that there is potential to continue this project into the next 5 years, w/continued support of R4 staff.

B. National Projects

Berry reported that the NBS Translational Research Network & Clinical Centers Workgroup of the NCC are working on creating long-term data sets for all NBS diseases. R4's process for completing interval/enrollment surveys has been copied in national data sets for creating disease-specific elements.

Berry solicited questions from the group, and invited them to participate if interested.

C. Abstracts

1. SIMD (submitted 10/30)

Berry indicated that this abstract has been successfully submitted. Berry asked the group if there was interest in preparing a similar abstract for ACMG.

2. ACMG (due 12/10)

Berry indicated that she expects this would be a similar focus on long-term follow up as with past submissions. Berry solicited comments/thoughts from members, and will draft an abstract for members to review.

D. Region 4

Hiner reminded the group to save September 13-15, 2010 for the next Regional meeting in East Lansing, MI. Hiner will solicit additional feedback from members via email.

Dates for the P2 face-to-face meeting are being explored with R4 staff. Berry added that, although much of the content for that meeting may be driven by whether or not we receive the RO1 grant, additional ideas/topics for discussion are welcomed. Our partners from outside R4 are welcome to participate, as well. More info will follow once a date is selected.

The next meeting is currently scheduled for Friday, December 17th, 2010 @ 2 PM. A confirmation email will be forthcoming on whether or not this meeting will stand as scheduled.

IV. Agenda additions

There were no additional items for discussion.

V. Adjourn

Meeting adjourned at 2:48 PM.