



Priority 2 Workgroup Telemeeting
Agenda ~ Friday, April 17, 2009 @ 2 pm ET/1 pm CT
1/866/489-0573 at the prompt, enter *4545164*

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|------|--|------------------|
| I. | Welcome & Roll Call | Anderson |
| | <i>Please announce yourself, your state, and, if applicable, the clinic you are representing and provide a brief update of where your center is in the IBEM-IS process (waiting for approval, finalizing IRB application, enrolling, consenting, etc.)</i> | |
| II. | IBEM-IS Update | Anderson/Bentler |
| | A. New surveys available | |
| | B. IRB updates / consents | |
| | C. Filling out reporting information | |
| | D. Status of survey entry in DocSite (handout) | |
| III. | National and cross regional collaboration | Anderson |
| | A. Challenge grant – DEADLINE to submit forms April 17, 2009 | |
| IV. | Update on Family Functioning Groups and State Stakeholder meetings | Hiner |
| V. | Next disorders for group review – May, 2009 | Anderson/Bentler |
| VI. | Items for follow-up of this meeting | Members |
| V. | Adjourn | Anderson |



Region 4 Priority 2 Workgroup
Telemeeting Notes - Friday, March 20th 2009

Participating: Susan Berry Co-Lead, Carolyn Anderson, Co-Lead; Kristi Bentler, Anne Jurek MN; Barbara Burton, Barb DeLuka, IL; Stephanie Gurnon, Kathy Wood, IN; Jan Askren, KY; Ayesha Ahmad, Allison Bannick, Jerry Feldman, MI; Nancy, Leslie, OH; Bill Rhead, WI; Sally Hiner, Region 4 Genetics Collaborative Coordinator.

I. IBEM-IS Participation Updates:

- Anyone desiring IRB assistance should contact Sally (shiner@mphi.org)

State/Lead	Center	Status
IL/Burton	Children's Memorial Hospital	No change
	University of IL	No change
	Rush University Medical Center	No change
IN/Hainline	Riley Hospital for Children	No change
KY/Gowans	University of KY	No Report
	University of Louisville (Askren)	Getting closer to complete application. No change
MI/Feldman	Wayne State	No change
	University of MI	No change
MN/Berry	University of MN	No change
	Mayo	No change
OH/ Leslie	Cincinnati Children's Hospital	No change
	Case Western Reserve	No change
	Nationwide Children's, Columbus	No change
	Akron	No change
	Dayton	No change
WI/Rhead	Waisman Center - WI center #2	No Report
	Medical Center of WI - WI center #1	No Report
NE/Skrabel	Nebraska	Working on application
IA/Copeland	University of Iowa	No report

II. IBEM-IS Update - Building the Information System

- A. Monthly Report: Sue provided a monthly report for the workgroup on IBEM-IS implementation progress. She revised the report to include the unique center identifier of the registry ID (last four numbers).
- B. Disorders Pending:
 - a. Currently enrollment surveys for 29 disorders are in DocSite. DocSite is finalizing interval surveys for two disorders.

- b. Disorders needing finalizing by the group
 - i. Biotinidase was provided to DocSite and added because we don't anticipate many, if any changes. Group reviewed and some changes were recommended. Kristi will forward changes to DocSite.
 - ii. Tyrosynemia – Tabled until all of the interval surveys are completed for disorders for which enrollment surveys are already done.
- c. End of life survey – Sue will check resources to see what legal implications adding an end of life survey might have. Sue and Kristi will draft some questions for the group to consider.
- d. Adding Disorders
 - i. Region 4 is in the process of developing budgets for next year. To assist, Sue did some research to be able to project the possible number of surveys we might want to add next year by looking at the ACMG recommended uniform panel.
 - ii. Does the group want to tackle PKU? There is a large PKU population. There definitely are benefits – but we need to look at the cost/benefit. PKU would not be as medically extensive, so less data to enter. It would be a terrific gain to have that data. There is some interest and support for doing this.
 - iii. FAOD's – we have all the primary targets, but are lacking some of the secondary targets. There are several, but they are really rare. One solution would be to have an "other FAOD" group. GAI might be good as a stand alone
 - iv. Organic Acidemia – we are missing a couple from the primary and secondary targets. C5 is the next target. We can do an OA other to capture the remaining targets. Again, they are very rare so don't require their own survey, but we should collect the information.
 - v. UCDs should be part of future consideration.
- e. Draft reporting form (see what was sent with the agenda). Consensus to use. Reports will be collected every six months.

III. National Expansion

- a. Mountain States – Minimum Data Sets
 - i. The Mountain States Regional Collaborative has renamed their "care plans" minimum data sets. This is because they don't want to imply that the data sets are consensus documents of how to treat or provide care for children with the particular disorder.
 - ii. Mike Watson, through funding of ACMG and the Secretary's Advisory Council on Heritable Disorders and Newborn Screening in Children brought together CDC, NIH, and HRSA to discuss direction of long term follow-up. CDC has received applications from 4 states building long-term follow up, NIH is interested in the research activity, and HRSA is interested in quality improvement, so it make sense for all of these funders to come together.
- b. Publication

- i. Carnitine – RO1: The group agreed to work on a grant to study carnitine. Susan Berry will put together a workgroup from the P2WG to begin working on a response for an RO1. P2WG members interested in proposal development, please email Sue.
- ii. The group formulated a query question for the DocSite data. Sue will share results of the query so the group can think about developing an abstract.
- iii. The group also is interested in developing an abstract on the process to date and current experience.

IV. Announcements:

a. Presentations:

Sue reminded everyone to advise Sally of any presentations about the IBEM-IS or Priority 2 workgroup. Sharing slides used also is always appreciated.

b. Stakeholder Meetings

Stakeholder Meetings are scheduled to occur in all 7 Region 4 states between March 23 and May 15th. Sally will be contacting registered workgroup members about presenting the workgroup updates. Sue has provided a draft set of slides for presenters to work with.

c. Next Meeting – April 17th.

Sue is not available as she will be participating in the Advisory Group for National Data Collection Meeting in Washington DC. The group will meet and Carolyn will chair the meeting.

d. Face-to-face Meeting – Fall 2009

Several of our new partner states (outside Region 4) inquired about face-to-face meetings as they prepare their budgets. We anticipate the next meeting of clinicians to be held in Fall 2009.

Notes by Hiner

AgeDeID	Conditions	Permission t	Permission t	MedicalRecordI	State	Surveys
3	MCADD Interv No		10/17/07	270610035	MN	MCADD Interval,MCADD Enrollment
2	MCADD Enrolli No		7/17/07	17061035	IL	MCADD Enrollment,MCADD Interval
3	MCADD Enrolli No		12/3/07	17051001	IL	MCADD Enrollment,MCADD Interval
2	MCADD Enrolli No		6/15/07	17071033	IL	MCADD Enrollment,MCADD Interval
5	MCADD Enrolli No		6/29/07	17031034	IL	MCADD Enrollment,MCADD Interval
2	MCADD Enrolli No		11/28/07	17071017	IL	MCADD Enrollment,MCADD Interval
8	MCADD Enrolli No		11/28/07	17001018	IL	MCADD Enrollment,MCADD Interval
2	MCADD No		11/28/07	17061036	IL	MCADD Enrollment,MCADD Interval
5	MCADD Enrolli No		12/5/07	17031037	IL	MCADD Enrollment,MCADD Interval
4	MCADD Enrolli No		12/6/07	17041038	IL	MCADD Enrollment,MCADD Interval
3	MCADD Enrolli No		12/7/07	17051039	IL	MCADD Enrollment,MCADD Interval
3	MCADD Enrolli No		12/10/07	17051040	IL	MCADD Enrollment,MCADD Interval
3	MCADD Enrolli No		12/11/07	17051019	IL	MCADD Enrollment,MCADD Interval
4	MCADD Enrolli Yes		3/27/07	270410001	MN	MCADD Enrollment,MCADD Interval
2	MCADD Enrolli Yes		1/17/07	270610002	MN	MCADD Enrollment,MCADD Interval
2	MCADD Enrolli Yes		1/17/07	270610003	MN	MCADD Enrollment,MCADD Interval
2	MCADD Enrolli Yes		2/1/07	270710004	MN	MCADD Enrollment,MCADD Interval
2	MCADD Enrolli Yes		5/2/07	270610020	MN	MCADD Enrollment,MCADD Interval
2	MCADD Enrolli Yes		3/22/07	270710005	MN	MCADD Enrollment,MCADD Interval
3	MCADD Enrolli No		12/14/07	270510055	MN	MCADD Enrollment,MCADD Interval
4	MCADD Enrolli No		10/17/07	270510044	MN	MCADD Enrollment,MCADD Interval
3	MCADD Enrolli No		4/4/07	270610011	MN	MCADD Enrollment,MCADD Interval,Neurop:
4	MCADD Enrolli No		7/18/07	270410027	WI	MCADD Enrollment,MCADD Interval,Neurop:
20	MCADD Enrolli No		4/4/07	278810012	MN	MCADD Enrollment,MCADD Interval
2	MCADD Enrolli Yes		3/21/07	270610006	MN	MCADD Enrollment,MCADD Interval
2	MCADD Enrolli No		2/21/07	270610010	MN	MCADD Enrollment,MCADD Interval
6	MCADD Enrolli Yes		4/4/07	270210013	MN	MCADD Enrollment,MCADD Interval
1	MCADD Interv Yes		2/14/08	270710067	MN	MCADD Interval
7	MCADD Enrolli Yes		1/23/08	270210064	MN	MCADD Enrollment,MCADD Interval
11	MCADD Enrolli Yes		1/23/08	279710063	MN	MCADD Enrollment,MCADD Interval
15	MCADD Enrolli Yes		1/23/08	279310062	MN	MCADD Enrollment,MCADD Interval
8	MCADD Interv Yes		4/25/07	270010021	MN	MCADD Interval
5	MCADD Enrolli Yes		4/11/07	270310014	MN	MCADD Enrollment,MCADD Interval
14	LCHAD Enrollment,LCHAD Interval			279410068	MN	LCHAD Enrollment,LCHAD Interval
6	MSUD Enrollm Yes		2/14/07	270310007	MN	MSUD Interval
2	MSUD Yes		5/16/08			MSUD Enrollment
0	MCADD Enrolli Yes		4/30/08	270810089	MN	MCADD Enrollment,MCADD Interval
1	MCADD Enrolli Yes		4/3/08	270810080	MN	MCADD Enrollment,MCADD Interval
5	MCADD Enrolli Yes		3/18/08	17041002	IL	MCADD Enrollment,MCADD Interval
7	MCADD Enrolli Yes		8/18/08	390820001	OH	
3	MCADD Enrolli Yes		9/8/08	390520002	OH	MCADD Enrollment
1	MCADD Enrolli Yes		9/8/08	390720003	OH	MCADD Enrollment,MCADD Interval
3	VLCAD Enrollment,VLCAD Interval			270610015	MN	VLCAD Enrollment,VLCAD Interval
2	SCAD Enrollment			270610059	MN	SCAD Enrollment
5	MCADD Enrolli Yes		10/13/08	390320004	OH	MCADD Enrollment,MCADD Interval
4	MCADD Yes		11/4/08	390420006	OH	MCADD Enrollment
8	MCADD Enrolli Yes		10/13/08	390120005	OH	MCADD Enrollment
11	SCAD Enrollm Yes		10/23/08	17971011	IL	SCAD Enrollment
0	MCADD			17081003	IL	MCADD Enrollment,MCADD Interval
4	VLCAD Enrollment			17051007	IL	
15	MSUD			17941006	IL	MSUD Enrollment,MSUD Interval
7	MSUD			17021004	IL	MSUD Enrollment,MSUD Interval
11	MSUD			17971005	IL	MSUD Enrollment,MSUD Interval
10	TFPD Enrollme Yes		3/7/08	17981009	IL	TFPD Enrollment,TFPD Interval
4	SCAD Enrollment			17041010	IL	SCAD Enrollment
24	VLCAD Enrollment			17841008	IL	VLCAD Enrollment,VLCAD Interval
3	VLCAD Enrollment			17061012	IL	VLCAD Enrollment,VLCAD Interval
51	MSUD Interval Yes		2/26/07	275710008	MN	MSUD Interval
21	MSUD			17871013	IL	MSUD Enrollment,MSUD Interval
0	MCADD Yes		10/22/08	17081016	IL	MCADD Enrollment,MCADD Interval
2	MCADD			17061014	IL	MCADD Enrollment,MCADD Interval
4	MCADD			17041015	IL	MCADD Enrollment,MCADD Interval
3	MCADD Enrolli Yes		8/20/08	180510003	IN	MCADD Enrollment
1	VLCAD Enrolln Yes		2/20/09	260720001		VLCAD Enrollment,VLCAD Interval

14	MCADD			189410001	IN	
30	MCADD			187810010	IN	
2	MCADD			180610023	IN	
1	MCADD			180710002	IN	
7	MCADD			180210004	IN	
3	MCADD			180510006	FL	
1	MCADD			180810007	IN	
4	MCADD			180410008	IN	
11	VLCAD Enrollment			189710013		
11	MCADD Enrollment			180710014		
0	MCADD Enrollment			180810015		
0	MCADD Enrollment			180810017		
27	MSUD Interval Update,MSUD Enrollment			278110009	MN	MSUD Enrollment,MSUD Interval
5	TFPD Interval,TFPD Enrollment			270310016	WI	TFPD Enrollment
0	MCADD,MCAD Yes	2/25/09		260820002		MCADD Enrollment,MCADD Interval
7	MCADD			180110019		
0	MCADD			180810020		
6	MCADD			180210022		
1	MCADD			180710022		
1	MCADD			180710024		
11	MCADD	No	8/5/08	17981021	IL	MCADD Enrollment,MCADD Interval
3	IBD Enrollment	Yes	12/16/08	17051020	IL	IBD Enrollment,IBD Interval
1	MSUD			17081022	IL	MSUD Enrollment,MSUD Interval