



**Region 4 Priority 2 Workgroup**  
**Telemeeting Notes - Friday, May 22nd<sup>th</sup>, 2009**

Participating: Sue Berry, Co-lead; Carolyn Anderson, Co-Lead; MN; Barbara Burton, IL; Kathy Wood, IN; Ayesha Ahmad, Allison Bannick, Jerry Feldman, MI; Kim Regis, OH; Jill Skrabel, NE. Jill Shuger, HRSA; Sally Hiner, Region 4 Genetics Collaborative Coordinator.

**I. IBEM-IS Participation Updates: Updates were not reported this month. The following information remains unchanged from April, 2009.**

- Anyone desiring IRB assistance should contact Sally ([shiner@mphi.org](mailto:shiner@mphi.org))

State/Lead	Center	Status
IL/Burton	Children's Memorial Hospital	50 consented
	University of IL	Barbara contacted George Hoganson. He has new staff starting this July and plans on getting on board.
	Rush University Medical Center	No change
IN/Hainline	Riley Hospital for Children	No change – consented 30,
KY/Gowans	University of KY	No Report
	University of Louisville (Askren)	Getting closer to completing application. No change
MI/Feldman	Wayne State	Consenting and enrolling; getting ready to renew IRB
	University of MI	No change
MN/Berry	University of MN	Enrolled 3-4 since last call
	Mayo	No change
OH/ Leslie	Cincinnati Children's Hospital	Consenting
	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

<b>II IBEM-IS Update</b>				
<b>A Surveys Available</b>				
Disorder (30)	In IBEM-IS		Posted on R4 Website	
	Enrollment	Interval	Enrollment	Interval
MSUD – Maple Syrup Urine Disease	X	X	X	X
Glutaric Acidemia Type 1	X	* <sup>1</sup>	pending	
Isobutyryl-CoA Dehydrogenase Deficiency	X	X	X	X
Carnitine Uptake Disorder	X	X	X	X
CACT Deficiency	X	X	X	X
CPT-1 Deficiency	X	X	X	X
CPT-2 Deficiency	X	X	X	X
SCAD Deficiency	X	X	X	X
MCAD Deficiency	X	X	X	X
LCHAD Deficiency	X	X	X	X
Trifunctional Protein Deficiency	X	X	X	X
VLCAD Deficiency	X	X	X	X
3-MCC Deficiency	X	*** <sup>2</sup>	pending	
2-Methyl 3-Hydroxybutyryl CoA Dehydrogenase Deficiency	X	*** <sup>3</sup>		
Holocarboxylase Synthetase Deficiency	X		pending	
3-Methylglutaconic Aciduria Type I	X	***	pending	
Beta-ketothiolase Deficiency	X	***	pending	
3-Hydroxy 3-Methylglutryl (HMG) CoA Lyase Deficiency	X	***	pending	
Succinyl CoA-3-keto transferase (SCOT) Deficiency	X	***	pending	
Propionic Acidemia	*	***		
MMA (includes Mut-, Mut0, Cobalamin A, Cobalamin B, Cobalamin D variant 2)	*	***		
MMA + Hcy (includes Cobalamin C, Cobalamin F, Transcobalamin II Deficiency)	*	***		
Biotinidase Deficiency	X	*	pending	
GALT Deficiency	*	*		
Tyrosinemia	***	***		

<sup>1</sup>\* In process at DocSite

<sup>2</sup>\*\*\* Ready for P2WG review/discussion

<sup>3</sup>\*\* Reviewed by Group, preparing to send to DocSite

The Region 4 Genetics Collaborative is funded by HRSA/MCHB Cooperative Agreement U22MC03963.

Item	Discussion	Next steps	Responsible person(s)			
<b>II IBEM-IS Update (continued)</b>						
B Status of survey entry in DocSite (H.O. #1)	Sue reviewed Handout 1, which included number of surveys in IBEM-IS by clinic. These will be provided monthly. The numbers will be needed when completing IRB re-application. A summary by disorder follows: Total Entries = 125	Report to be provided monthly	Berry/ Jurek			
	MCADD			79	CUD	7
	MSUD			13	LCHAD	4
	SCAD			6	CPT1	1
	TFPD			4	CPT2	1
	VLCAD	9	IBD	1		
C Update on DocSite Data Retrieval	Sally has been working between DocSite and Anne Jurek to resolve data retrieval issues. Sally identified experts in both SQL and SAS who have drafted a plan to translate the IBEM-IS data in DocSite from SQL to SAS so that Anne can work with the data. MPHI is currently negotiating with John Haughton, DocSite and the Interactive Solutions Group at MPHI to have this plan implemented before the end of the fiscal year.	Monitor data translation plan process and keep group informed.	Hiner			
	To assist both DocSite and MPHI ISG in implementing the data translation plan, John H has asked this workgroup to formulate some data questions to be used in the test queries. Sue proposed data questions which also could be used to develop abstracts for presentation at upcoming conferences.	N/A	N/A			
	<p>Questions for data query Set of data about MCADD</p> <ul style="list-style-type: none"> <li>• Symptoms at time of DX</li> <li>• Genotype</li> <li>• Initial NBS values</li> </ul> <p>Clarification was asked re: NBS values – what are you using? MS/MS from NBS lab? In IN we confirm with another lab. Initial MS/MS from NBS lab is what we want.</p>	Provide questions to DocSite and ISG	Hiner			

D Brochure for IBEM-IS	Hiner presented the draft IBEM-IS brochure. The IRB information for University of MN should be viewed as a place holder, to be replaced with individual clinic IRB info. This could be done at time of printing, if printed locally, or via sticker. Pediatric specialist will be replaced with metabolic specialist. Sue will review for accuracy of revised consent protocol; and forward revised language if needed.	Request review and comment	Hiner
		Group to review and provide final comments	P2WG
		Compile comments, revise as appropriate	Hiner
<b>III National and Cross Regional Collaboration</b>			
A NIH Challenge Grant – FAOD TRN	Region 4 is partnering with 3 sites outside of the region in application for an NIH Challenge grant. If funded, the grant will expand the use of the IBEM-IS for FAODs by involving sites beyond Region 4 in enrollment and data access for research. This is virtually the same project as submitted to NIH last July. Berry extended special thanks to Hiner and Griffin (MPHI R4) for rapid response to questions and following up on needs.	Group to be notified if application is accepted for funding	Berry/Hiner
B NBSTRN	Sue provided an update on the Newborn Screening and Translational Research Network for which ACMG received funds last year to serve as the coordinating center. Both R4 Priority 1 and Priority 2 projects are represented on the national workgroup. Both the concept and the funding are complex. CDC, NIH and HRSA all are interested in supporting projects around long term follow-up. These three federal entities are working together in this process.	Group to be appraised of NBSTRN activities and progress	Berry
<b>IV Update on Family Functioning Discussion Groups and Stakeholder Meetings</b>			
A Family Functioning Discussion Groups	Discussion groups were held at all 7 Stakeholder meeting sites. The number of participants ranged from 3 – 17. Participants responded to the issue: “The impact of having a child with a heritable conditioning on family functioning”. MPHI is currently compiling responses and conducting qualitative analysis. Findings will be shared with the P2WG via Darin Erickson. (Reporting – Hiner)	Completed analysis Share findings	MPHI
B Stakeholder Meetings	Stakeholder meetings were held in all 7 Region 4 states between March 23 and May 15. Attendance was good, ranging from 21 – 60 participants. MEMSCIS generated a great deal of discussion during the meetings and Jihad Adwan, MEMSCIS Coordinator, has received a good number of follow up contacts from individuals learning about MEMSCIS during the stakeholder meetings. Emergency preparedness was discussed in each of the 7 states during the opportunities/future directions discussion. Notes and materials will be posted to the Region 4 website. A Survey Monkey evaluation is being developed to gather feedback and suggestions.	N/A	N/A

<b>V MEMSCIS</b>			
A Update	Hiner provided an update of changes to the web application that are expected to be completed by May 31, 2009. University of MN submitted a revised IRB application, changing the MEMSCIS from a research project to a clinical tool. With that change, changes in protocols were approved that allow families to complete online consent at the time of enrollment.	Notify P2WG of availability for active enrollment once website revisions are complete	Hiner
B Registering Clinics	Metabolic clinics can submit their clinic information to be added as a “designated care provider”. In MEMSCIS, this is called a Health Care Entity. This designation allows clinic specific protocols to be available and facilitates the critical component of connecting families to a clinic. Clinics can be added, even if they do not currently have families enrolling in MEMSCIS. Enrollment requires providing basic clinic information and designation of a contact person.	Provide forms to P2WG members and encourage to register with MEMSCIS	Hiner
<b>VI Fall Face-to-Face Meeting</b>			
A Update	The next face-to-face meeting of the P2WG clinicians is scheduled to occur this fall and will include clinicians from the Heartland Region. We need to select a date and location and begin planning the agenda.	Suggest dates	Hiner
<b>VII Year End</b>			
A Contracts/LOA’s for FY ’09-’10	Hiner requested clinical state leads who will be requesting honoraria and clinics participating in IBEM-IS who will be requesting enrollment stipends initiate the fiscal agreements for the next fiscal year. Then, as soon as we have our notice of grant award, these agreements can be finalized. Patty Losey and Maria Ostrander will be working from MPHI to facilitate getting agreements/contracts in place.	Initiate process within clinic for LOA/contract for next fiscal year	Clinical state leads
		Initiate process within clinic for LOA/Contract for enrollment stipend for next fiscal year	clinician
B Invoice for FY ending 05/31/09	Clinical state leads requesting honoraria for the period Jan 1 – May 31 2009 should get those invoices in as soon as possible. Clinics requesting enrollment stipends for the period Jan 1 – May 31, 2009 need to invoice as soon as possible. (Hiner reporting). An enrollment report also is due to Anne Jurek.	Invoice for honoraria	Clinical state leads
		Submit enrollment invoice to MPHI Submit enrollment report to Anne Jurek	clinicians
C Reporting Consents	Berry reminded IBEM-IS participants to complete and submit the form to report number consented by disorder. This information will be compiled and shared with the group. IRBs will want the number consented reported to them on the re-application/continuing application.	Submit number consented report	Clinicians consenting

Notes by Hiner

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