



Region 4 Priority 2 Workgroup
Telemeeting Notes - Friday, July 17th, 2009

Participating: Carolyn Anderson, Co-Lead; Kristi Bentler, MN; Barbara Burton, Claudia Nash, Lorna Pena, IL; Jan Askren, KY; Ayesha Ahmad, Jerry Feldman, MI; Nancy Leslie, Kim Regis, OH; Bill Rhead, Sandy vanCalcar, WI; Sara Copeland, IA; 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)

State/Lead	Center	Status
IL/Burton	Children's Memorial Hospital	Consenting & Enrolling
	University of IL	Lorna Pena will be representing UIC. She has initiated the IRB process.
	Rush University Medical Center	No change
IN/Hainline	Riley Hospital for Children	Consenting & Enrolling
KY/Gowans	University of KY	No Report
	University of Louisville (Askren)	IRB issues have been resolved with the exception of cost of review.
MI/Feldman	Wayne State	Consenting and enrolling; getting ready to renew IRB
	University of MI	No change
MN/Berry	University of MN	Enrolling
	Mayo	No change
OH/ Leslie	Cincinnati Children's Hospital	Consenting
	Case Western Reserve	No change
	Nationwide Children's, Columbus	Consenting
	Akron	No change
	Dayton	No change
WI/Rhead	Waisman Center – WI center #2	IRB approved and trained
	Medical Center of WI – WI center #1	IRB approved
NE/Skrabel	Nebraska	Working on application
IA/Copeland	University of Iowa	Application drafted

II IBEM-IS Development				
A Surveys Available				
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	X ¹	X	
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	*** ²	X	
2-Methyl 3-Hydroxybutyryl CoA Dehydrogenase Deficiency	X	** ³	X	
Holocarboxylase Synthetase Deficiency	X		X	
3-Methylglutaconic Aciduria Type I	X	***	X	
Beta-ketothiolase Deficiency	X	***	X	
3-Hydroxy 3-Methylglutryl (HMG) CoA Lyase Deficiency	X	***	X	
Succinyl CoA-3-keto transferase (SCOT) Deficiency	X	**	X	
Propionic Acidemia	X	**	X	
MMA (includes Mut-, Mut0, Cobalamin A, Cobalamin B, Cobalamin D variant 2)	X	**		
MMA + Hey (includes Cobalamin C, Cobalamin F, Transcobalamin II Deficiency)	X	***		
Biotinidase Deficiency	X	X		
GALT Deficiency	X	X		
Tyrosinemia	***	***		

¹* In process at DocSite

²*** Ready for P2WG review/discussion

³** Reviewed by Group, preparing to send to DocSite

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Item	Discussion	Next steps	Responsible person(s)
II IBEM-IS Development			
A. Proposed dialysis questions	The group discussed these proposed additions to enrollment surveys to collect this information for any disorder that could result in hyperammonemia/acute organ failure. The group made some recommendations for additions including: Enrollment survey – age at treatment and days Interval survey – age, months, days, type of dialysis	Revise dialysis questions	Bentler
		Disseminate for review	Hiner
		Review for finalizing	Group members
B. Proposed changes on all surveys	As Kristi and Carolyn were developing additional surveys, they identified some information for the additional disorders that might be helpful to allow the collection of/prompt for all disorders. The group agreed in cases where it is easier for DocSite to include in all surveys, they should be added and clinicians will leave blank those which are not appropriate to the patient.	Revise dialysis questions	Bentler
		Disseminate for review	Hiner
		Review for finalizing	Group members
C. Review of surveys a. MMA b. PA c. SCOT d. BK	All 4 surveys were reviewed by the group. Revisions will be made as suggested and the surveys forwarded to DocSite for inclusion in the IBEM-IS. Neuro radiology review of some of the data elements is needed. Next steps include finishing the interval surveys for disorders for which enrollment surveys are complete; then move on to other disorders	Revise as discussed	Bentler
		Obtain and include neuro review	
		Forward to DocSite	
III National and Cross Regional Collaboration			
Draft manuscript of workgroup progress	Feedback – the date for formal commenting already passed. Feedback was positive. Authorship – Sue is very interested in the group’s thoughts on how to deal with issues of authorship as we are a large, active group with many contributors. The group recommended authorship be the core group and then on behalf of group	Share group response with Sue	Anderson

IV Face to Face Meeting			
Face to Face meeting of Clinicians	The group discussed October as being a difficult month with so many conferences. The option of piggybacking our meeting on a national meeting was discussed. Many members are going to San Diego conference, but out of office time and travel arrangements already have been made. The conference in Albuquerque in March was suggested. Sally needs information about meetings so conflicting times are not proposed and/or we can target meetings to tag onto.	Advise Sally of conferences to avoid and those which are possibilities for tagging on to	All workgroup members
		Advise Sally of conflicts you already are aware of BEFORE the list of date options is posted	
		Send out a list of possible dates	Sally
		Provide your availability	Workgroup members
V. Next Meeting			
	August 21, 2009 1 pm CT / 2 pm ET		

Notes by Hiner