

IBEM-IS Clinicians Face-to-Face Meeting
November 20, 2009
Chicago, IL

Welcome & Introductions

In attendance: Anderson, Bannick, Bentler, Berry, Gowans, Hainline, Jurek, Katz, Leslie, Peck, Pena, Regis, Rhead and van Calcar; Cameron and Griffin from Region 4

National Initiatives on Long-term Follow-up – 9am-10am

PRESENTATION - IBEM-IS AND NATIONAL DATA COLLECTION INITIATIVES
(Berry)

❖ **NICHD funded projects**

- Newborn Screening and Clinical Translational Research Network (NBSCTRN)
 - Region 4 involvement
 - Sue Berry chairs the long term follow-up committee
 - Ann Jurek is a member of the IT committee
 - A CH committee is being formed – they have indicated they would like to work with the Region 4 CH group and will help fund a meeting that will include national representatives
 - September 09 Meeting
 - LTFU group met on data sets
 - Region 4 work has impacted national data collection efforts
 - ◆ Reviewed and adopted MCAD data set with one suggested change
 - ◆ Include values (would need to transport values from e-record to limit issues)
 - ◆ Modified to include more medical home concepts
 - Up next
 - Uniform language for data sets
 - ◆ Take data set and national library of medicine to integrate and develop uniform language
 - ◆ Creation of letter or report for an EMR – billable
 - Disease specific data sets
 - IT Planning (Anne to follow with more)
 - Proposals
 - ◆ RFPs that have to do with data collection and projects that have to do with data collection
 - Large amounts of data are appropriate for surveillance for Public Health
 - Others would be good for research

❖ **Secretary's Advisory Committee on Heritable Disorders in Newborns and Children**

- Sue Berry on Long Term Follow Up Subcommittee
 - Medical foods group – Kerry bill (MA)
 - Sponsor payment for foods – federally

- Action towards supporting this bill

❖ **CDC long term follow- up projects**

- Collaboration with NICHD and HRSA projects seems limited

PRESENTATION - IBEM-IS IN IT PLANNING (Jurek)

❖ **NBSCTRN - Informatics system to support patient data and biospecimen repositories**

- Meetings to date
 - Genetics in Electronic Health Systems Meeting (Feb 2008)
 - Describe current genetics content – IBEM-IS presentation
 - NBSTN Planning meeting (April 2009)
 - Facilitate planning for systems and structures underlying the NBSTRN
 - IT workgroup (October 2009)
 - NBSCTRN Infrastructure
 - Purpose - Recommendations to build and maintain the large network
- IT Infrastructure Future Directions
 - See what else has been done and build off of it
 - Looking at caBIG – NCI’s Cancer Biomed Informatics Grid
 - How can we link with surveillance data with LTFU clinical info
 - It’s important that LTFU data be linked to all the other activities; and should be linked without much conflict – continue to gather more information

DISCUSSION - HRSA funded Regional Projects (Cameron)

The NCC has received HRSA funding to hire one person to link what is happening in LTFU across the regions (Amy Brower and she will be making rounds to the regions). Right now there are multiple projects focused on LTFU, with little coordination across projects or vision as to how they might come together.

- ❖ Region 4 IBEM-IS
 - Where will funding come from after this 5 year grant cycle?
 - Will it become part of NBSCTRN?
- ❖ NYMAC – diagnostic criteria project (interesting – online)
- ❖ Mountain States – initial efforts to develop LTFU data for public health, now working on adding clinical data – built from birth defects registries but is a limited data set - only includes data on 2 disorders: MCAD and PKU
- ❖ South East region – worked with Public Health Informatics to develop a business case; identified ideal data elements for LTFU database; next step to compare LTFU systems with public health informatics identified data elements

Questions

- ❖ What happens as these projects move beyond the regions?
 - Will they stay in the region or move on to another entity?
 - Last spring Cameron provided Dr. Michelle Lloyd Puryear from HRSA with a concept of how to provide infrastructure support to a national LTFU project. Dr. Puryear has indicated that once a project is national, it can no longer be administered through a regional collaborative.

- Will funding be available after the end of this 5 year grant cycle?
- If the projects stay with the Region, how do we get funds to start new projects?
- If the projects move to another entity, will they get the same amount of support and continue to succeed?
 - ❖ It has been suggested by Mike Watson, that
 - NBSCTRN will adopt Region 4 model and use it to provide technical assistance
 - NBSCTRN will fund a meeting for R4 endocrine workgroup and incorporate national endocrinologists

Immediate concern for this group:

Cameron: We budgeted for the development of an IBEM-IS National Advisory Board for the current grant year. How do we feel about that, knowing that in the future this will go national? Should we move forward and create a boarder National Advisory Board or should we do it with the NCC or let them do it in 2.5 years?

Berry and Leslie: Region 4 should drive the boat

Berry: Keep this group moving and don't bring in the NBSCTRN LTFU group – to keep autonomy

We need to make the decisions about future and plan effectively and be successful. We can work on research issues regionally, but it will be resolved nationally. We can develop a model for national consideration.

IBEM-IS Research Discussion 10a-11am

DISCUSSION – TERMS OF ENGAGEMENT AND SHARING INFORMATION

P2 project has PHI data and we need to discuss how to proceed with requests, terms of engagement, etc. (G. Arnold (co-investigator of FOAD proposal) separately proposed another application that was reliant on the data that Region 4 has.

Internal Requests/Usage

Berry: NCC/Mike offered a meeting to discuss use of information – this may be useful

Cameron: Publications group for NCC is also meeting – Nancy Leslie would be happy to represent Region 4 on this group

Berry: Access to data is open and available to Region 4 contributors

Jurek: user can only see their own state and cannot run queries/reports

Leslie: data is in there, but you would need to send a data request

Berry: request vs. share; we will discuss; make sure we give credit, if we request

Berry: authors are writers; workgroup should be listed as other contributors

Leslie: we should be the model for those broad database pubs for the future

Berry: if what we do works well, others will follow

Berry: Include other participants in the lists – ok

Data mining

Establish study cohorts for overlaying

Berry: Should we stay with a 4 person review team or move towards ad hoc?

Leslie: could create a 4 person board; send it out and if substantial discussion, then put on the phone

Berry: only one able to do the data mining; people need equal access

DECISION – WORKGROUP MEMBER REQUESTS

- ❖ For data mining or cohort studies requested by workgroup members
 - Request to Cameron
 - Cameron to recruit 3 person review team (Public health; parent/family member or R4 parent coordinator; and clinician. Take out methodologist - Cameron will preview the request)
 - At the time the request is reviewed – let the group know; to foster interaction and keep apprised
 - When requesting data, identify which data elements they want access to
- ❖ For data mining: Jurek and Berry will mine the data after approval

Requests for data from non-workgroup members

Collaborative studies requested of non-workgroup members – MPHI will negotiate rights of access.

If they want the data, then they should contribute data; will need IRB/DocSite licenses and they would be allowed access

We don't want to restrict research possibilities for the data – it's a matter of sorting that out and how we would facilitate the access

Hainline: have we considered if a drug company would like to go through one of us, and how do you handle that?

Berry: two things there: data mining and overlay a research question, which would be a second step – clinicians would need to agree and engage their patients. This would be a fabulous opportunity to work together.

DECISION – NON WORKGROUP MEMBER REQUESTS

- ❖ All requests go to Cameron. MPHI is an honest broker in the deal
- ❖ At the time the request is reviewed – let the group know; to foster interaction and keep apprised
- ❖ The review panel will evaluate the proposal
- ❖ If the review panel approves the proposal, it will be circulated to workgroup members to ascertain conflict of interest/ways to collaborate with the researcher.
- ❖ Workgroup members will develop recommendations for collaboration to be included in a Memorandum of Agreement
- ❖ MPHI will establish a Memorandum of Agreement for collaborative interaction that will vary by request.

DISCUSSION – RESEARCH RESOURCES

vanCalCar: Are there resources available through Region 4 to support research of workgroup members?

Cameron – this is not in the budget but sometimes the region has carry forward funds that can be requested to support projects. Approval of the Region 4 Advisory group would be obtained prior to spending funds in this way. It also depends on what amount of support would be needed – up to \$5,000 is a realistic possibility; while 25,000 is probably not realistic.

- May get more proposals with seed money – but that may need to come from outside sources
- Could pilot it and get advisory groups approval
- There would need to be support from WG and approval by advisory group

DISCUSSION – CHANGES TO PROCESS & FORMS

Leslie: question 5 – IRB approved? Before IRB will approve, they need a number of subjects – Could give preliminary numbers to support IRB proposals

Researchers need to send query so feasibility can be assessed prior to submitting full proposal.

- ➔ CHANGE NAME of Data Request Form to Research Proposal
- ➔ Ask for data elements – for data mining activities

For cohort studies – if a researcher wants to pose additional questions they would have to submit a proposal for the additional survey to the group for approval and THEY would incur the cost to add survey to DocSite (the interested party would plan for that in planning and budgeting).

Jurek: Clarification on cohort – perhaps use: long term follow up or expansion study
Or “add-on studies”

- ➔ CHANGE FORM: 10 A – let group know of data mining requests

---Break 11:10-11:20---

DISCUSSION: WORKGROUP PARTICIPATION

State Rep Participation

Region 4 participants; encourage interaction between departments of health

- ➔ Action Item: Need to bring this up to the state leads to see what is appropriate

Hainline: Taking surveillance data and using in IN – have to enter it in both places; doing entry for the state

Anderson: There are different ways for interaction

Hainline: Direct connection with the state should be fostered

Anderson: In the beginning stages we need to involve the state at this time to be doing surveillance

- Need to have a generalized meeting
- Go into enroll a patient; and see that their information for surveillance data
 - Enter data for surveillance if they do not consent anyways
- This is an evolution – we need to go in that direction!

Leslie: need to refine more than state closure logs. Should be following all of them! Expansion of activity

In Georgia: Interfaced NBS database and Public School Database, it's flawed, but could find correlations

We do have a denominator problem; but one way to fix is to move forward with surveillance database

Every state has own system/administration – how collaborative can/will they be; either way this offers a tool for them and we should engage them.

Anderson: One barrier – they see this as a research tool; clinicians will need to advocate and work hard to develop those associations and educate
Starting to understand (California and Massachusetts); but not quite there yet.

Hainline: IN has a genetic counselor in state board of health – they have infiltrated!
Clinicians don't always know what the state does; and perhaps geneticists will be able to close the gap

DECISION – PARTICIPATION OF STATE REPS

Push this as a surveillance tool! Make linkage between clinicians and state department

Creating Useful Data Reports 11:10 am -1:00 pm

DISCUSSION – DATA REPORTS (Cameron)

How might the data be helpful to you outside of research?

Cameron: I make an assumption that you want all specialists to enter cases, not just those who want to do research. How do you make the data useful to those clinicians? What reports would be beneficial?

Berry: Ability to have center specific data for their own center – whether they have all had flu shots, for example; add characteristics just for your center

Leslie: Industry sponsored sites -- you can look up patient info, flag it, etc; set some limits and reminders for your patients

Berry: Example: Complete listing of patients and the surveys they are assigned
Stock reports are not available, but we can have them created

DEMONSTRATION – HOW TO CREATE A REPORT IN DOCSITE (Berry)

Berry: Are there reports that the group would like to see that Berry could report regularly?

Bentler: Tip: If you have timed out, “Measure review” has blanks where you have not entered data – if you are kicked off and have to go back to find where you left off

Priority 2 Work Plan

DISCUSSION –NEW SURVEYS

Should the imaging survey be independent from the disorder surveys?

- Leave it imbedded in the disorder surveys (AB, NL)

Considered a death survey – Mountain states, recoiled – reason: ascertaining for legal purposes

- Survey would have to be constructed without proscribing causality

Dialysis and pregnancy surveys to be developed

- IRB considerations – Berry will look into further
 - **Loren can help – will look into it as well and share
- Dialysis is done
 - A few things were changed
 - Levels of ammonia (peak studied level) and branch chain of amino acids
- ➔ Metabolite and peak level
- ➔ ADD: Free text box for why dialysis stopped
- ➔ Reason for Dialysis – should be a check box not a pick list
- ➔ If you have an “other” should have what is specified below
- ➔ Add-ons would be appropriate for these

Pregnancy survey

- Volunteers – Pregnant; perhaps use something from a maternal PKU survey
 - Bannick to help -- look and pass around

Transplant survey

Leslie could do this by giving some ideas to Bentler

- Articles or anything foundational would be helpful – send to Bentler

DECISION - NEXT SURVEYS TO BE COMPLETED

1. Tyrosinemia – almost ready to go and will be next to add
 Immediate entries available
 Hainline: Tyro. Type II – make this available
 Make sure you have eyes and skin apps in too
 Agree on interval
2. Isovaleric Acidemia
3. 3MAG Type 1 – third to be added
 ➔ Collect as a phenotype as they are – don't take at Type 1
 Remove "Type 1"
4. GA-2 should be near the top of the list as well
5. HMG
6. MMA + Hcy
 Homocystinuria
 3-4 patients – but all literature is there, so it could be easy to release survey
 Can be on the list to do
7. 2-methylbutyryl CoA dehydrogenase deficiency
 Lower on the list – being done a different way!
 Sandy has a database with 65+ Mung cases
 ▪ If you have cases, perhaps talk with Sandy (2-4 year olds especially)

Other Disorders

Urea Cycle Disorders - Consortia were excited about us adding surveys

Pull their survey, adapt to DocSite – would make faster; they would have to apply to us to get their data

Develop even though not all are newborn screened

What about NKH (Nonketotic Hyperglycinemia)

Is this NBS-able? KY does

These patients die – but our system is not based on ascertainment

Bottom of the survey – worthwhile to put one in to look at the glycines in the profiles

Could potentially add an "other" survey to cover: such as malonic acidemia

Urea cycles – do it as UCD, and then split them

See what survey looks like, Berry to share and see

OAT – "Ornithine Artho Transferase"

Ophthalmologist will pick this up
Likely more out than we know

“Rare Others” Genetics survey – to include things like LPI (lung), OAT (eye)

Hainline and Leslie (Leslie to look into LPI) will see if these should go elsewhere

We are not going to do CF -- since they have their own database - same with Endocrinopathies and Hemoglobinopathies

NL: Creatine disorders? Let’s set those aside for the moment

Make sure we have things that are well justified and significant mortality/morbidity

Upcoming Potential Diseases:

SCID – next one up; however screening test has not diagnosed anyone

WI and New England just started doing it

Summary

1. Complete
 - a. interval survey
 - b. Tyro
 - c. IVA
 - d. GA2
 - e. HMG
 - f. Hcy
 - g. 2methylbutyryl
2. Then, move towards conversations on:
 - a. UCD’s
 - b. PKU

DISCUSSION - IBEM-IS BROCHURE

- ➔ Will need to be forwarded on to IRBs
- ➔ Change IRB # on back
- ➔ Send template for sites to update!

DISCUSSION - MEMSCIS

Name change – change coming soon

Consent process – being done on the web

Fill out: Form to set up clinics as entities on MEMSCIS

Not a research activity; does not need to go through IRB

Actual information in MEMSCIS will be filled out by parents/clinicians/both

Prescriptions will be double-checked

Emergency letters – can copy/paste in

To send in MEMSCIS form – send electronically to group and add in Jihad’s info
Fax: 612.626.2993 attn: Sue or Kristi

- ➔ R4/MPHI can provide access to MEMSCIS brochures as well
- ➔ Send Hans the form (Cameron will do)

Closing Comments

We should work as a region as long as we can; not stop moving forward at any point
We need to try and resolve sustainability for data entry

NIH grant funds (?) for data entry

Mike/NBSCTRN – suggested that forms are sent to a central place and they can be entered there

IRB issues – entry errors – privacy demographics secure, send the rest

It’s not sustainable to pay for every entry – Children’s cancer data centers; how do they do it?

They DO pay -- for “Centers of excellence” to do data entry.

It has been suggested that if university/hospital views it as a QI piece they might be willing to support case entry

Use systems (EPIC) to connect – billing, etc

Data set utility; a product the clinician can really use!

MCAD paper – Leslie will assist, Hainline will read, Berry will write

Berry: encourages to ENTER DATA!