

Integrating Electronic Records Systems into National Data Collection: A Region 4 Project

Presented by:

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For the Region 4 Priority 2 Project Workgroup



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Outline

- Rationale for studying Inborn Errors of Metabolism
- Region 4 long-term follow-up project
- Data priorities and analysis
- Linking to other electronic data systems



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Inborn Errors of Metabolism (IBEM)

- Definition
 - Rare genetic conditions
 - Typically autosomal recessive
 - Enzyme abnormality produces a metabolic error
- Example
 - Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)



IBEM continued

- Symptoms are specific for each condition
- Treatment
 - No uniform or organized strategy
 - Almost no evidence-based care (beyond experience of practitioners)
 - Rare diseases
 - Primarily children
 - Very little existing data regarding long-term outcomes



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Newborn Screening (NBS)

- Each State decides which tests to do
- New HRSA-ACMG* initiatives have defined a “uniform panel”
- Many states expanded to add conditions detected by tandem mass spectrometry (MS/MS)
 - MS/MS detects a variety of IBEM



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*HRSA-ACMG = Health Resources and Services Administration-
American College of Medical Genetics

Health Resources and Services Administration (HRSA)

- Assists in improving the conditions of children with rare genetic conditions, including IBEM ascertained by NBS
- Set up regional genetics collaboratives to address these issues
- A major focus for our Region: long-term follow-up of IBEM



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Region 4 Collaborators

- Metabolic Clinicians and State Health Department NBS Specialists

- Illinois
- Indiana
- Kentucky
- Michigan
- Minnesota
- Ohio
- Wisconsin



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- All 7 states screen by MS/MS

Region 4 LTFU workgroup's charge and priorities

- Develop and implement a regional action plan to address long-term NBS follow up and evaluation of clinical outcomes
 - Create standardized diagnostic and medical management protocols for disorders diagnosed by NBS/Care plans for specific diagnoses
 - Evaluate clinical outcomes through identifying critical elements for follow up



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An Evolution: The Region 4 Registry

- Gathering uniform data and assessing clinical practice differences is a way to learn which treatment strategies are most effective.
- Started with one condition—MCADD
 - 5-7 kids/year in MN
 - 45-50 kids/year in Region 4



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MCADD: How we initiated registry activity



- Reviewed literature
- Used existing sources
 - Mountain States Collaborative & Oregon database
- Mutual agreement on essential elements
- Sections
 - Enrollment
 - Demographic
 - Short- and long-term follow-up interval surveys
 - Neuropsychological survey

Development of an IBEM- Information System (IBEM-IS)

- Expanded from MCADD registry
- Add other IBEM
- Keep core elements, add disease- and center-specific elements
- Create a large-scale follow-up record as a platform for research



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Registry Design

- Ease of data entry
- Web-based with option of paper checklist for use during clinical visits
- Access of each center for their own data
- Summary data access limited
- Allow access to Departments of Health for their state's data



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DocSite[®]:

A relational database platform

- HIPAA compliant
- Ease of entry at point of service
- Reporting functions
- Ease of adding elements for management/monitoring
- Ease of analysis



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Sites [Add](#)

Metabolic Test Site - Everyc

Patient Search [Add](#)

Last Name:

First Name:

DOB:

MRN:

Status:

[Search](#) [Clear](#)
 Search All Sites

Patient Reports

Please select a report type:

PDF XLS WORD

- [Visit Planner](#)
- [Outreach Report: Phone](#)
- [Patient Flow](#)
- [Patient Flow: All Measures](#)
- [Patient Handout](#)
- [Progress Note VP and Patient Handout](#)

Patient [Edit](#)

Demographics

Last Name: **Another** First Name: **Lexy**
 Middle Name: Date of Birth: **2/20/2007 (6 months)**
 Gender: **Female** Race: **White**
 Preferred Language: **English** Ethnicity:
 Title: Salutation:

Contact Information

Address Line1: **456 Anywhere St** Address Line 2:
 City: **South Walleye** State: **MN**
 Zip: **55455** Email:
 Phone: Alt Phone:
 Alt Phone 2:

Other Information

SSN: **Not entered** MRN: **Not entered**
 Patient Status: **Active** Principal Care Site: **Metabolic Test Site - Everyone**
 Allergies:
 Comment:

Managed Conditions [Add](#)

Managed Condition	ICD-9	Date Diagnosed	Rank		
MCADD Enrollment		unspecified	1	delete	edit
MCADD Interval Update		unspecified	2	delete	edit

Current Medications [Add By Name](#)

No medications found.

Patient Planner

https://patient.docsite.com - ReportViewer - Microsoft Internet Explorer

File Edit View Favorites Tools Help

Save a Copy Print Email Search Review & Comment Sign

Select Text 118%

Metabolic Test Site - Everyone Print Date: 8/31/2007

Visit Planner Date of Visit: ___/___/___ Page 1 of 5

Provider: Susan Berry	Patient: Lexy Another	Gender: Female	Phone:
Visit Provider:	MRN:	DOB: 2/20/2007 (0)	
Conditions: MCADD Interval Update, MCADD Enrollment		Preferred Language: English	
Comorbidities:		First Measure Date: 02/21/2007	
Allergies:			
Medications:			

Administrative	Today's Action	Last Value	Date Last	Pt. Goal	Int. Days	Due Date
! Unique Registry ID (2digFIPS/2digBirthYr/1digCenter/3digAssession)		27071fakeMN002	02/27/07		Per Visit	
Permission to contact about research - I agree to be contacted with information on potential future research applicable to my inborn error of metabolism that becomes available	Yes, No	Yes	02/27/07		Per Visit	
Permission for compensation - I agree that identifying information about me may be used or disclosed as necessary to provide compensation if I am eligible for compensation	Yes, No	Yes	02/27/07		Per Visit	
! Is patient followed by more than one metabolic center?	Yes, No, Unknown, N/A	No	02/27/07		Per Visit	
! Maternal education - highest grade level completed	1-8 years (grade), 9-12 years (no diploma), Completed high school, Training after high school (not college), Some college, College graduate, Post-graduate	College graduate	02/27/07		Per Visit	
! Paternal education - highest grade	1-8 years (grade), 9-12 years (no diploma),	Post-graduate	02/27/07		Per Visit	

8.5 x 11 in

1 of 5

Done Internet

start PatientPlanner - Mic... https://patient.docsit... Removable Disk (E:) Microsoft PowerPoint ... 3:22 PM

Sites [Add](#)

Metabolic Test Site - Everyc

Patient Search [Add](#)

Last Name:

First Name:

DOB:

MRN:

Status: Active

[Search](#) [Clear](#)

Search All Sites

Patient Reports

- Please select a report type:
- PDF
 - XLS
 - WORD
- [Visit Planner](#)
- [Outreach Report: Phone](#)
- [Patient Flow](#)
- [Patient Flow: All Measures](#)
- [Patient Handout](#)
- [Progress Note VP and Patient Handout](#)

Patient [Edit](#)

Last Name: **Another** First Name: **Lexy** DOB: **2/20/2007**

Gender: **Female** SSN: **Not entered** MRN: **Not entered**

Providers

Berry, Susan

Survey Data Entry

Select a survey: Metabolic - Interval MCADD Survey

[Save](#) [Cancel](#)

Measure Date [Set all dates](#)

Interval History/Concerns		
Measure Name	Date	Value
! Dx Code (ICD-9)	<input type="text"/>	<input type="checkbox"/> MCADD (277.85) <input type="checkbox"/> other
! Other Diagnoses (free text)	<input type="text"/>	
! Follow-up Status	<input type="text"/>	<input type="checkbox"/> Active <input type="checkbox"/> Closed <input type="checkbox"/> Deceased <input type="checkbox"/> Lost to follow up <input type="checkbox"/> Moved out of

Condition Region 4 Registry as a Research Platform



- Plan interventions that can be assessed with data in IBEM-IS
- Initial projects may examine
 - Clinical history
 - Short-term outcomes
- We request at enrollment that registry subjects consider consent to allow continuing contact, anticipating engaging them as participants in further research trials.

IBEM-IS Accomplishments



- Selected a “host” that uses Web-based EMR technology
- Engaged the majority of treating centers in Region 4 (total 17)
 - Added additional metabolic leads in larger states
- IRB approval received: MN, IL, WI, OH
- Registry entry proceeding for MCAD deficiency - new and interval
- Elements defined and added for MSUD: **adds an aminoacidopathy**
- Elements defined and added for LCHAD **(extending a category)**
- Elements defined for C3, C5OH disorders (organic acidemias) - includes biotinidase: **adds the 3rd MS/MS category**
- Enrolling all clinic subjects as they give permission
- Entering new patient data as elements are added to the database
- **Total number of disorders defined: 26**
- **Total enrolled: 89 (31 MCAD; 5 MSUD; 17 OA; 17 LCFAOD)**

Integration of Data

- Departments of Health
- Emergency services
 - Minnesota Emergency Medical Services for Children Information System (MEMSCIS)
- Other Regional Collaboratives



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Birth head circumference (OFC) (cm)

Newborn Screening

Measure Name	Date	Value
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If newborn screened, days of age at time primary or metabolic provider were notified of FIRST abnormal newborn screen for this IBEM (365 x yrs or 30 x months or counted days)

State newborn screen serial number

C6 (hexanoylcarnitine) on FIRST newborn screen (enter 99999 if N/A) (umol/L)

C8 (octanoylcarnitine) on FIRST newborn screen (enter 99999 if N/A) (umol/L)

C10 (decanoylcarnitine) on FIRST newborn screen (enter 99999 if N/A) (umol/L)

C10:1 (decanoylcarnitine) on FIRST newborn screen (enter 99999 if N/A) (umol/L)

C8/C10 ratio on FIRST newborn screen (enter 99999 if N/A)

C6 (hexanoylcarnitine) on SECOND newborn screen (enter 99999 if N/A) (umol/L)

C8 (octanoylcarnitine) on SECOND newborn screen (enter 99999 if N/A) (umol/L)

C10 (decanoylcarnitine) on SECOND newborn screen (enter 99999 if N/A) (umol/L)

C10:1 (decanoylcarnitine) on SECOND newborn screen (enter 99999 if N/A) (umol/L)

C8/C10 ratio on SECOND newborn screen (enter 99999 if N/A)

C6 (hexanoylcarnitine) on THIRD newborn screen (enter 99999 if N/A) (umol/L)

C8 (octanoylcarnitine) on THIRD newborn screen (enter 99999 if N/A) (umol/L)

C10 (decanoylcarnitine) on THIRD newborn screen (enter 99999 if N/A) (umol/L)

C10:1 (decanoylcarnitine) on THIRD newborn screen (enter 99999 if N/A) (umol/L)

C8/C10 ratio on THIRD newborn screen (enter 99999 if N/A)

Diagnostic Testing

Measure Name	Date	Value
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Midwest Emergency Medical Services For Children Information System

- Participating Hospitals
- Web Links
- Privacy
- Contact
- Take the Tour!



User ID:

Password:

Submit

EMERGENCY ACCESS

Forgot your password?

REGISTER PATIENT

A collaboration of University of Minnesota, MCHB Genetics Region 4, MN EMSC Resource Center, and the EMS Regulatory Board.

Working together to improve children's lives.



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IBEM-IS Conclusions

- Track and long-term follow up of metabolic disorders ascertained through expanded newborn screening
- Link with other electronic records
- Advance research
- Improve care for children with rare genetic disorders



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