

Update: Long Term Follow up of Newborn Screening Conditions in New England

New England Newborn Screening Program

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

February 26, 2009

Anne Marie Comeau, Ph.D.
Deputy Director, NENSP
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Building Upon the Foundation of Six New England States' Comprehensive Newborn Screening Programs for Sustainable Follow-Up

Foundations

- New England longstanding NBS collaborative networks
- Massachusetts' experience
- Centralized NBS database, continuation of existing datasets

Goals

- Public Health Quality Assurance,
 - Public Health Quality Improvement and
 - Public Health Engagement in Research

Long-Term Follow Up Workgroup

Ellie Mulcahy, Gail Boaz -- Maine Genetics Program

Barbara McNeilly, Kristine Campagna -- Rhode Island NBS Program

Cindy Ingham – Vermont NBS Program

Marcia Lavochkin -- New Hampshire NBS Program

Vine Samuels-- Connecticut's Laboratory NBS Tracking Program

Janet Farrell – Massachusetts DPH

NENSP

Roger Eaton, Inderneel Sahai

Jaime Hale

Anne Comeau

Massachusetts Regulations

Stewardship and Authority



Massachusetts Regulations Governing Blood Screening of Newborns for Treatable Diseases and Disorders

(105 CMR 270.000 MGL c.111, ss3, 4E, 5, 6, 24A and 110A)

270.010: Follow-up of Newborn Blood Testing

For the purposes of quality assurance, quality improvement and ongoing evaluation of the effectiveness of the Newborn Blood Screening Program, including the determination of those disorders and diseases that should be screened for, the attending physician shall report to the Newborn Blood Screening Program, upon request, the following information within 30 days of the request:

- (A) Diagnostic and long term outcomes for all newborns whose newborn screening results warranted diagnostic evaluation for a newborn screening disorder or disease; and
- (B) Any additional, relevant information regarding these diagnostic and long term outcomes as specified by the Newborn Blood Screening Program.



Massachusetts Regulations Governing Blood Screening of Newborns for Treatable Diseases and Disorders

(105 CMR 270.000 MGL c.111, ss3, 4E, 5, 6, 24A and 110A)

270.010: Follow-up of Newborn Blood Testing

For the purposes of quality assurance, quality improvement and ongoing evaluation of the effectiveness of the Newborn Blood Screening Program, including the determination of those disorders and diseases that should be

**purposes of quality assurance,
quality improvement and ongoing
evaluation of the effectiveness**

(B) Any additional, relevant information regarding these diagnostic and long term outcomes as specified by the Newborn Blood Screening Program.



Massachusetts Regulations Governing Blood Screening of Newborns for Treatable Diseases and Disorders

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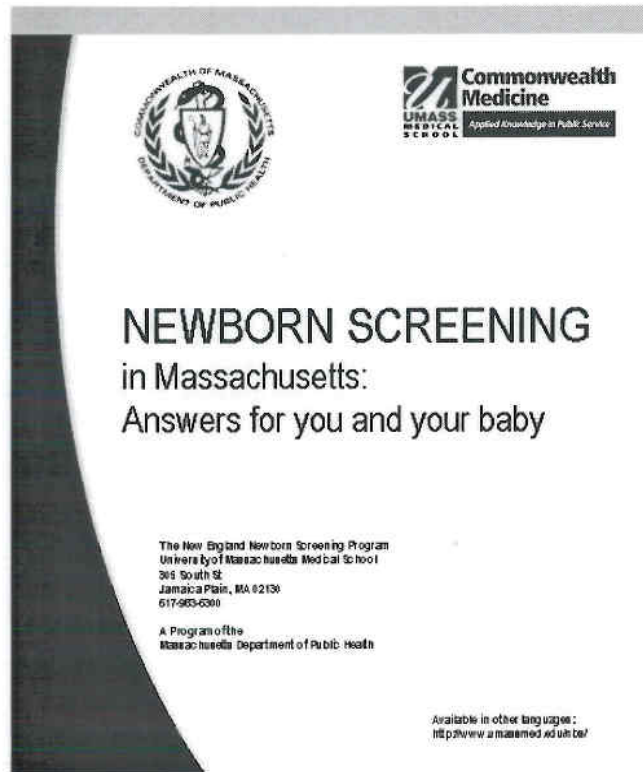
For the purposes of quality assurance, quality improvement and ongoing evaluation of the effectiveness of the Newborn Blood Screening Program, including the determination of those disorders and diseases that should be

**shall report to the Newborn
Blood Screening Program**

Diagnostic and long term outcomes

(B) Any additional, relevant information regarding these diagnostic and long term outcomes as specified by the Newborn Blood Screening Program.

Parent Notification of NBS Quality Assurance and Improvements in Massachusetts



NEWBORN SCREENING

QUALITY ASSURANCE AND IMPROVEMENTS

Newborn screening programs need to know that they are working well and need to know how to improve. This means that programs need to know whether the screening results match diagnostic results. This also means that programs need to know how babies who are diagnosed with newborn screening disorders are doing and whether they continue to get the care they need. Information on diagnoses and outcomes is collected for program-wide improvements.

Your baby's information and leftover blood may be stored for at least 10 years. Sometimes, the information or leftover blood will be used to make sure that newborn screening tests are working well. Sometimes the information or leftover blood will be used to make better tests for the newborn screening program. Other times, the information or leftover blood will be used for health studies. For any health studies, your written permission is needed before we release your baby's name to an external researcher.

In addition, if any information or leftover blood is going to be used for a study, the study has to be approved by two groups of people who make sure that your baby's rights are protected. These groups of people are called "Human Subjects Review Committees". One Human Subjects Review Committee is at the Department of Public Health, and the other is at the University of Massachusetts Medical School. The Federal Government sets the rules and regulates each Committee. For any proposed study, Human Subjects Review Committees decide whether your permission is needed. If either Committee decides that your permission is needed, the New England Newborn Screening Program will contact you before proceeding with the study.

Contacting you: We know that for many reasons, parents change health care providers and may change the name of their baby. If your baby has been diagnosed with a newborn screening disorder, or is being followed to find out if your baby has a newborn screening disorder, you may receive a letter from the New England Newborn Screening Program to ensure that your baby's information is up to date.

3 Foci for Data Collection and Best-Practice Development

Hemoglobinopathies

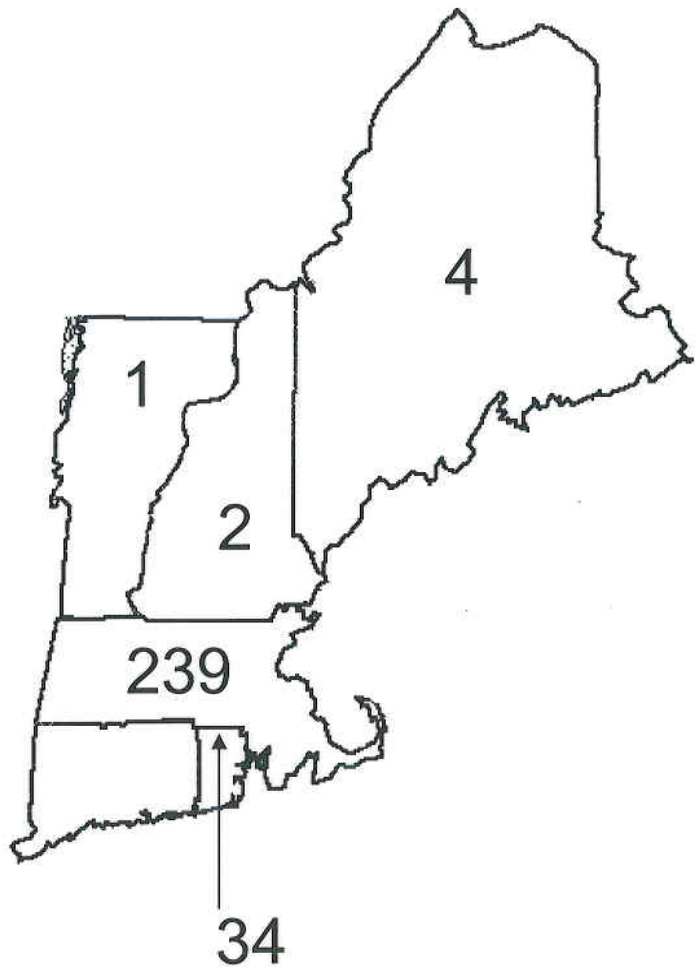
Cystic Fibrosis

Metabolic Conditions

Sickling Hemoglobinopathies



Sickling Hemoglobinopathies Detected by the NENSP in New England 2003-present



A laboratory analyst transfers newborn sample extracts for sickle cell disease screening at the New England Newborn Screening Program.

The New England Hemoglobinopathies Newborn Screening Workgroup

Dr. Anne Marie Comeau Ms. Claire Hughes

Dr. Naheed Usmani

Dr. Philippa Sprinz Dr. Anjulika Chawla

Ms. Kathleen Ryan, RN Dr. Matthew Heeney

Dr. Karla Fuentes

Dr. Maria Pelidis

Dr. Mary Huang

Dr. David Ebb

Dr. Anne Rossi

Dr. Farzana Pashankar

Dr. J. Nathan Hagstrom

Dr. Joanna Luty



Baystate Children's Hospital



Children's Hospital Boston



Connecticut Children's
MEDICAL CENTER

Floating Hospital
for Children
at **Tufts** Medical
Center



MassGeneral Hospital
for Children



Hasbro



LTFU Variables for Sickling Hemoglobinopathies

Defined by MA and Regional NBS Hgb Workgroups

Census

- Date of last clinic visit
- Date of most recent visit
- Alive; if no date & cause of death

General Demographics/Current Practices

- Height (cm) & Weight (kg) at last clinic visit
- TCD (Normal/Abnormal); date(s)

LTFU Variables for Sickling Hemoglobinopathies

continued

Clinical Status

- Clinical Stroke? if yes, date(s)
- Radiological Stroke? if yes, date(s)
- MRI/MRA Abnormal? date(s)
- Splenectomy? if yes, date

Notable Infections

- Febrile Episodes ? if yes, date(s)
- Culture + ? Sepsis? if yes, date(s)

Treatments and Clinical Episodes

- Hyper Transfusions? if yes, start & stop dates
- Prescribed Hydroxyurea in last 6 months? if yes start & stop dates
- Hospitalizations ?if yes, date(s)
- Acute Chest ? if yes, date(s)



New England Newborn Screening Program
 University of Massachusetts Medical School
 305 South Street
 Jamaica Plain, MA 02130-3597
 617-983-6300 (office) 617-522-2846 (fax)

Hemoglobinopathies LTFU Report

Last Name: _____	
First Name: _____	
DOB: _____	NBS result: _____
Initial Clinic: _____	Current Clinic: _____
Confirmatory HGB Result: _____	

Date of most recent clinic visit: _____

Height (cm) _____ Date _____

Weight (kg) _____ Date _____

Date of 1st TCD: _____ Normal Abnormal

Date of Last TCD: _____ Normal Abnormal

MRI/MRA: Normal Abnormal Date _____

Hospitalizations: No Yes *If yes, date(s)* _____

Acute Chest Syndrome: No Yes *If yes, date(s)* _____

Clinically "Overt" Stroke: No Yes *If yes, date(s)* _____

Radiologic only "Silent" Stroke: No Yes *If yes, date(s)* _____

Febrile Episodes: No Yes *If yes, date(s)* _____

Culture + Bacteremia: No Yes *If yes, date(s)* _____

Prescribed hydroxyurea in last 6 months? Yes No Hyper Transfusions? Yes No

Start Date _____ Start Date _____

End Date _____ End Date _____

Splenectomy: No Yes *If yes, date* _____

Is child living? No Yes *If no, date of death* _____

If no, cause of death _____

**Data form for
 Hemoglobinopathies
 Sent between
 NENSP and clinics**

Sampling of LTFU Hgb data

30 patients (DOB 2003-present)

Variable	n	%
At least one clinic visit in the past 12 months	25	83
At least one TCD	8	27
At least one hospitalization in their life	7	23
Splenectomy	2	7
At least one event of acute chest in their life	2	7
Taken hydroxyurea	4	13
Clinical stroke <i>(Child has had 2 clinical strokes – at 2.5 and 3 years of age)</i>	1	3

Cystic Fibrosis

The Massachusetts CF NBS Workgroup

Representatives from NENSP and the 5 MA CF Centers

Dr. Anne Marie Comeau

Dr. Richard Parad

Baystate  Children's Hospital

Dr. Robert (Bob) Gerstle



Children's Hospital Boston

Dr. Henry (Hank) Dorkin

Dr. Terry Spencer



MassGeneral Hospital
for Children

Dr. Allen Lapey



Tufts-New England Medical Center

Dr. William (Bill) Yee



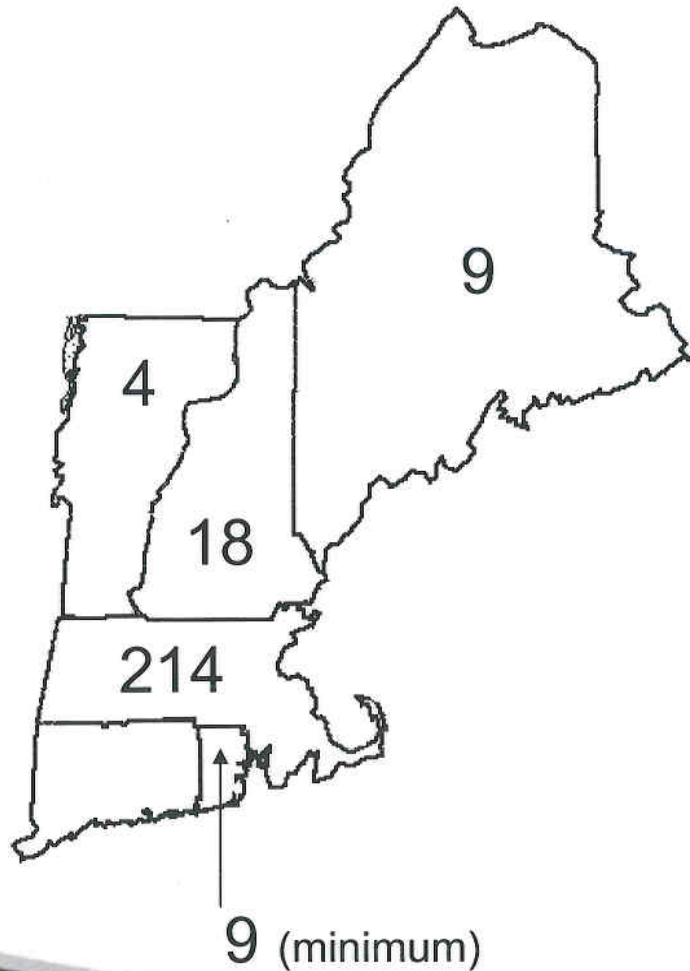
UMassMemorial
Medical Center

A Member of UMass Memorial Health Care

Dr. Brian O'Sullivan

Cystic Fibrosis Cases Detected by the NENSP in New England

(From state's start date – January 31, 2009)



State	CF NBS start date
Massachusetts	2/1/1999
New Hampshire	5/1/2006
Rhode Island	7/1/2006
Vermont	3/1/2008
Maine	7/1/2008
Connecticut	—

Notable Trends in Cystic Fibrosis Cases Detected by the NENSP in New England

Massachusetts: Decrease of CF

Hale JE, Parad RB, and Comeau AM. Newborn screening showing decreasing incidence of cystic fibrosis. N Engl J Med. 2008 Feb 28;358(9):973-4.



The NEW ENGLAND
JOURNAL of MEDICINE

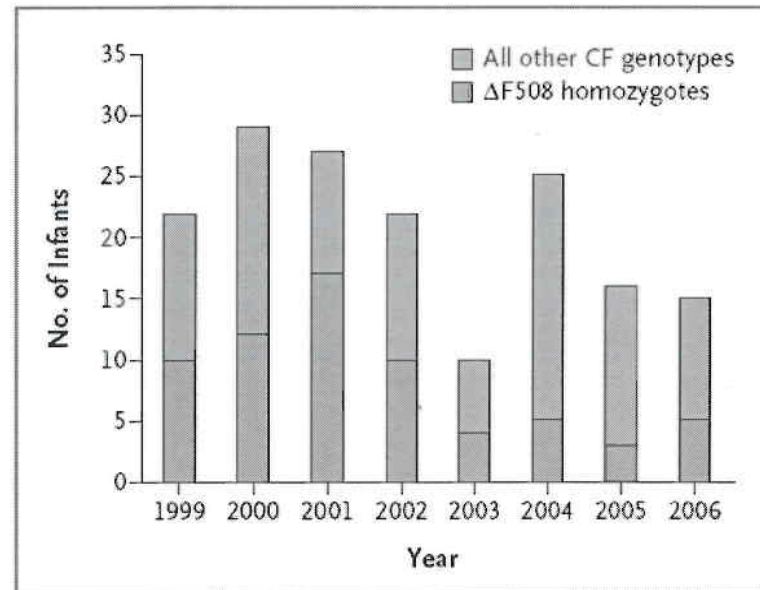


Figure 1. Newborns with Cystic Fibrosis Identified by Newborn Screening for the Disease in Massachusetts. Bars indicate the number of first persons in a family with positive results of newborn screening and a diagnosis of cystic fibrosis (CF). Additional persons in a single family were excluded.

Notable Trends in Cystic Fibrosis Cases Detected by the NENSP in New England

Maine: CF NBS Case Projections vs. Actual Number

Projected Cases per Year

Northern Maine

2 CF Births per year

Central Maine

2 CF Births per year

Southern Maine

2 CF Births per year

6 TOTAL per year



Actual Cases Detected in 7 months of screening

Northern Maine

2 CF births

Central Maine

5 CF Births

Southern Maine

2 CF

9 TOTAL in 7 months

Metabolic Conditions

The New England Metabolic Newborn Screening Workgroup

Inderneel Sahai, MD (NENSP), Chair



Dr. Madelena Martin Dr. Beverly Hay

Dr. Vivian Shih Dr. Marsha F. Browning



Dr. Mark Korson Dr. Cheryl Garganta



Dr. Gerald Berry Dr. Harvey Levy

Dr. Chanika Phornphutkul

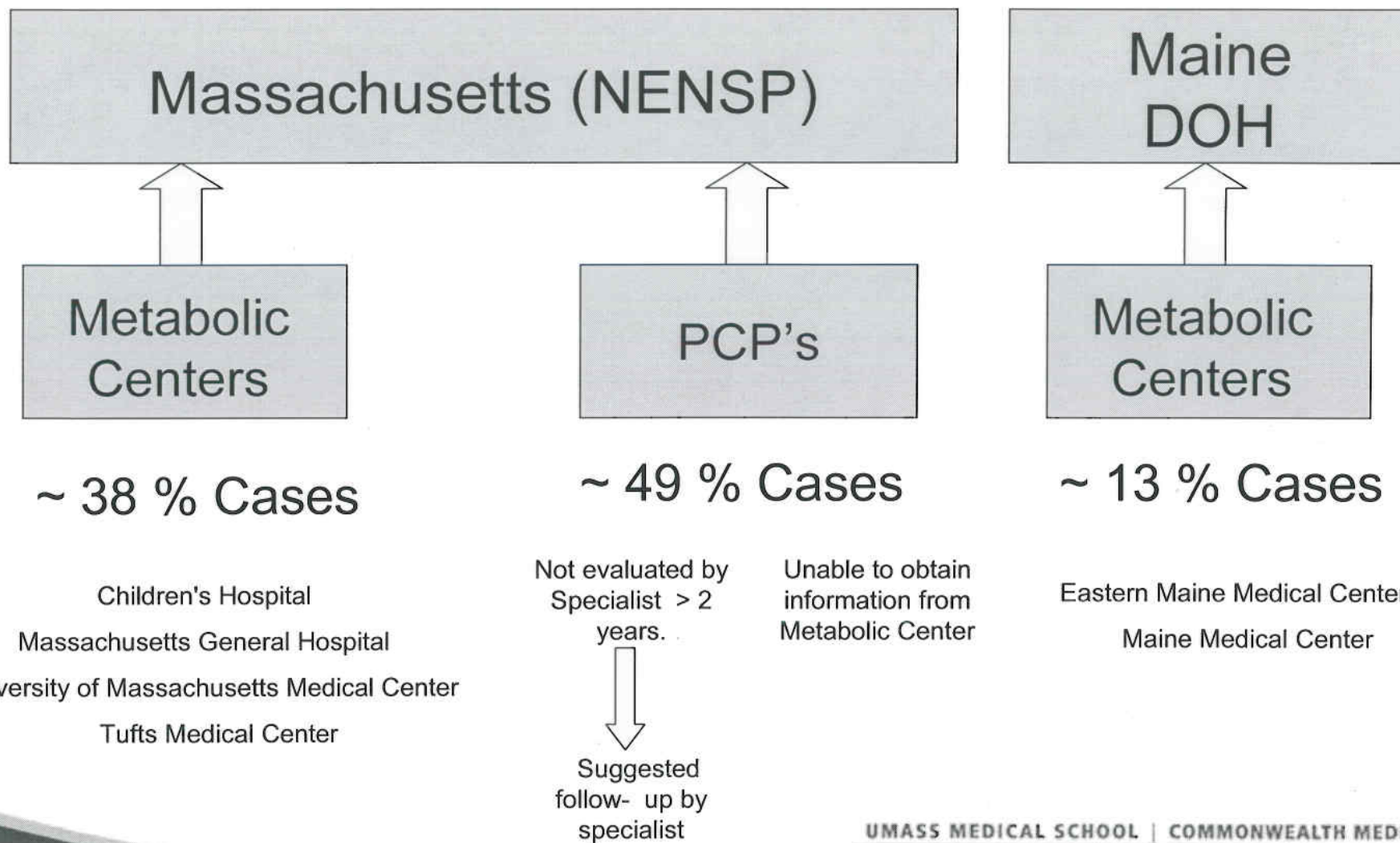


Dr. Thomas Brewster Dr. Wendy Smith

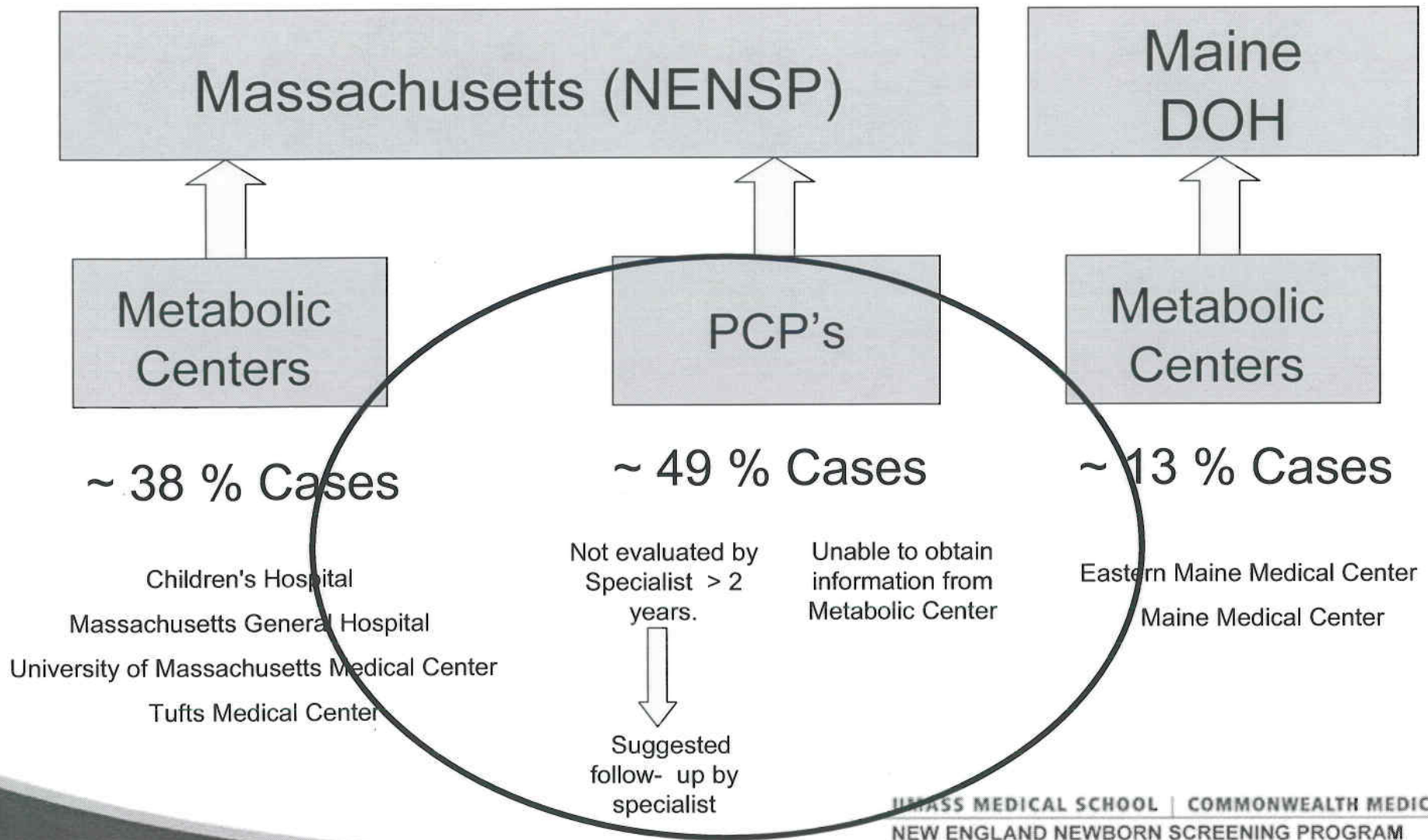
Dr. Leah Burke Dr. Greta Seashore



SOURCE OF NENSP LTFU DATA (Metabolic Disorders)



SOURCE OF NENSP LTFU DATA (Metabolic Disorders)

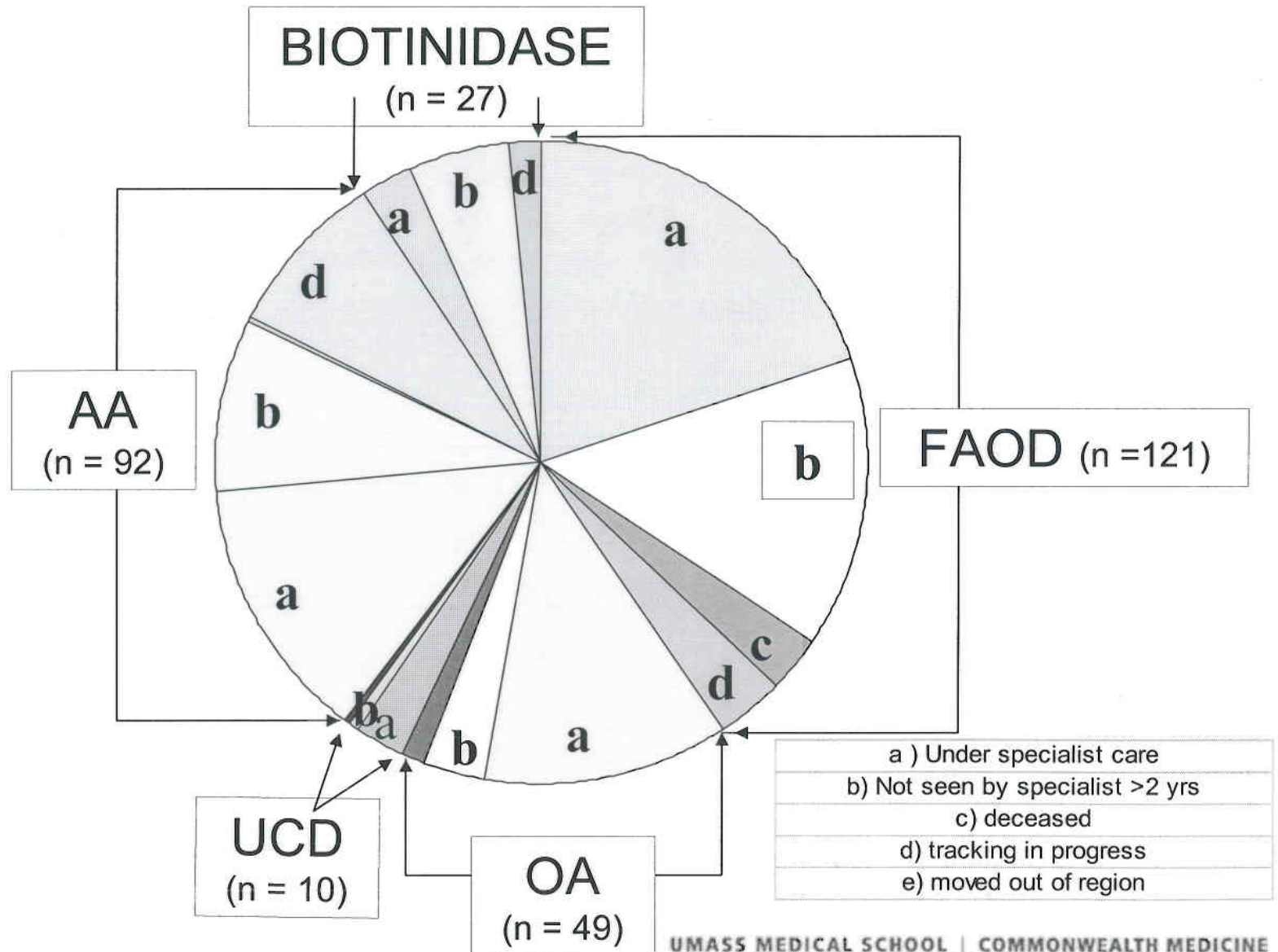


LTFU Status of Metabolic cases detected by NENSP in MA and ME

	Total n	Deceased	Moved	Tracking	LTFU Data	Not seen by specialist > 2 years
Fatty Acid Oxidation Defects	121	8	0	10	103	44
Organic Acidemias	49	0	3	0	46	9
Urea Cycle Defects	10	0	1	0	9	1
Amino Acid Disorders	92	1	0	25	66	26
Biotinidase Deficiency	27	0	0	5	22	15
TOTAL	299	9	4	40	246	95

95/246 children with LTFU data have not been seen by specialist in > 2 years

FOLLOW-UP SPECIALIST CARE



DISORDERS WITH LIMITED FOLLOW-UP WITH METABOLIC SPECIALIST

FAOD	<ul style="list-style-type: none">•MCAD (19/39)•VLCAD (6/21)•CUD (4/10)•SCAD (15/28)
OA	<ul style="list-style-type: none">•MCC (9/16)•MBCD (2/2)
UCD	<ul style="list-style-type: none">•Citrullinemia I (1/4)•CPS (1/1)
AA	<ul style="list-style-type: none">•Atypical PKU (6/40)•Hyper-PHE (17/18)•MAT (3/4)
Bio	<ul style="list-style-type: none">•Profound (3/5)•Partial (12/17)

Reasons Stated for Follow-up Outside of Specialty Care

- Limited information provided by specialist (uncertain spectrum)
- No specific treatment provided
- Child has remained asymptomatic without treatment
- Unnecessary travel

Center-to-Center Transfers

Metabolic: ~ 48%

Cystic Fibrosis: ~20%

Hemoglobinopathies: <5%

LTFU of Metabolic Disorders Identified in Massachusetts & Maine (1999-2008)



Metabolic Case Outcomes.
Sahai et al

MCAD

Our records show that you are the primary care physician for a baby with MCAD deficiency

summarizes our experience to date, and alerts you of two late-occurring deaths among babies identified at birth in New England with MCAD.

Although most children with MCAD deficiency diagnosed by newborn screening have done well clinically, there have been two deaths, at age 11 months and 33 months. Both children had regular pediatric care and had no other medical conditions. One child had vomited several times but ate a snack before falling asleep. He was found unresponsive the next morning and could not be resuscitated. The other child had also vomited several times before becoming lethargic.

These deaths underscore the importance of close monitoring for early signs of illness and immediate medical attention to prevent severe hypoglycemia.

PEDIATRICS®

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

Spectrum of Medium-Chain Acyl-CoA Dehydrogenase Deficiency Detected by Newborn Screening

Ho-Wen Hsu, MD^a, Thomas H. Zytkevich, PhD^a, Anne Marie Comeau, PhD^a, Arnold W. Strauss, MD^b, Deborah Marsden, MD^c, Vivian E. Shih, MD^d, George F. Grady, MD^a, Roger B. Eaton, PhD^a

^aDepartment of Pediatrics, New England Newborn Screening Program, University of Massachusetts Medical School, Jamaica Plain, Massachusetts; ^bDepartment of Pediatrics, University of Cincinnati College of Medicine, Cincinnati, Ohio; ^cDepartment of Pediatrics, Children's Hospital Boston, Boston, Massachusetts; ^dDepartment of Neurology, Massachusetts General Hospital, Boston, Massachusetts

ABSTRACT

Objective: We describe the clinical spectrum of medium chain acyl-CoA dehydrogenase (MCAD) deficiency detected by routine newborn screening, assess factors associated with elevations of octanoyl carnitine (C8) in newborns, and characteristics associated with adverse clinical consequences of MCAD deficiency.

Pediatrics 2008;121;e1108-e1114

Back to the Foundations: The Databases

Metabolic Database

102aGetGuthrie

LAST FIRST DOB GUTHRIE_NUMBER
 MOTHER'S LAST BABY_SYSKEY QJ1207050534
 RF1210050301
 SC1213051562

Create, Edit, View New Contact, Baby Hx_Clinical | Create, Edit, or View Medical Visit and Labs | Long Term Follow-Up | Conversion to Metric

102bContacts by Specimen, from Metabolic Elevations Tbl subform

Specimen-Driven and Specific Contacts

The contact information in this box concerns only the first contact made in response to -> QJ12-07-05-0534 this specimen.

Date 12/8/2005 To what: Dr. Office, etc. DR MCCAHAN
 Is This First Contact Ever on This Baby? Name of Person Spoken With: DR MAFFEI
 Contact Made By (XY) JCB Tele# Fax#
 Contact THROUGH (Person) (direct) FK_BABY: 20366153
 Contact TO (Category) Cov PCP

102cMiniVisitForm NOTE: Visits Already Entered

VisitCategory	FacilityCategory	Visit/Sample I
First Specialist	Specialist	12/15/2005

The information below refers to the whole baby (is not specimen specific)

102dBabyList subform

Baby Syskey	Tracking Aids	Action Requested by NENSP:	Req'd Date	Still Waiting for:	Current Specialist:
20366153	<input type="checkbox"/> Lost to F/U <input checked="" type="checkbox"/> Tracking Done	<input type="checkbox"/> Follow-Up FP <input type="checkbox"/> 1st PCP Eval <input type="checkbox"/> 1st Referral To Specialist <input type="checkbox"/> Other Request (see comments)	<input type="text"/> 12/8/2005 <input type="text"/> 12/8/2005 <input type="text"/> 12/8/2005	<input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/>	<input type="text"/> HAY Current PCP from Core: <input type="text"/> MCCAHAN <input type="text"/> 5088364884

Check to add to Baby List

Record: 14

Dx Candidate Disorder MCAD DxStatus case Date Of Diagnosis 1/5/2006

102eCLINICAL_TBL

Is There Significant Family History? Consanguinity

Fam Hx Comment: NEG

Neonatal Course

Full Term? (check if yes) Gestational Age (wks):
 Prenatal Complications? Pren'l Comp Comments: TION, PROGESTERON RX 1ST TRIMESTER
 Obstetrical Complications? Obs Comp Comments:

Major Milestones

	Yrs	Mos
Crawling	<input type="text"/>	<input type="text"/>
Walking	<input type="text"/>	<input type="text"/>
First Words	<input type="text"/>	<input type="text"/>

Delays ("ever")

Motor
 Receptive Skills
 Speech

Metabolic Database

102aGetGuthrie

MOTHER'S LAST | C BABY_SYSKEY | 20366153 QJ1207050534
 RF1210050301
 SC1213051562

Create,Edit,View New Contact,Baby Hx _Clinical Create,Edit,orViewMedicaVisit and labs Long Term Follow-Up Conversion to Metric

Specimen-Driven and Specific Contacts

102bContacts by Specimen,from MetabolicElevations Tbl subform

The contact information in this box concerns only the first contact made in response to --> QJ12-07-05-0534 this specimen.

Date: 12/8/2005 To what Dr. Office, etc.: DR MCCAHAN

Is This First Contact Ever on This Baby? Name of Person Spoken With: DR MAFFEI

Contact Made By (XY): JCB Telet: Fax#: FK_BABY: 20366153

ContactTHROUGH(Person): (direct)

Contact TO (Category): Cov PCP

102cMiniVisitForm **NOTE: Visits Already Entered**

The information below refers to the whole baby (is not specimen specific)

VisitCategory	FacilityCatego	Visit/Sample I
First Specialist	Specialist	12/15/2005

102dBabylist subform

Baby Syskey 20366153	Tracking Aids	Action Requested by NENSP:	Req'd Date	Still Waiting for:	CurrentSpecialist:
Check to add to Baby List <input checked="" type="checkbox"/>	Lost to F/U <input type="checkbox"/>	Follow-Up FP	12/ 8/2005	<input type="checkbox"/>	HAY
	Tracking Done <input checked="" type="checkbox"/>	1st PCP Eval	12/8/2005	<input type="checkbox"/>	Current PCP from Core rbelv
		1st Referral To Specialist	12/8/2005	<input type="checkbox"/>	MCCAHAN
		Other Request (see comments)		<input type="checkbox"/>	5088364884
		LastContactDate:	3/2/2006		

Record:

102eCLINICAL_TBL

Is There Significant Family History? Consanguinity

Fam Hx Comment:

Neonatal Course

Full Term? (check if yes) Gestational Age (wks):

Prenatal Complications? Pren'l Comp Comments:

Obstetrical Complications? Obs Comp Comments:

Major Milestones

	Age	
	Yrs	Mos
Crawling	<input type="text"/>	<input type="text"/>
Walking	<input type="text"/>	<input type="text"/>
First Words	<input type="text"/>	<input type="text"/>

Delays ("ever")

- Motor
- Receptive Skills
- Speech

Clinical Signs/Conditions in Baby ("ever")

- CARDIAC
- PULM
- RENAL
- HEPAT
- NEURO
- SEIZURES
- Baby Not in Cohort
- Clinical Attention 1st
- ID
- GI
- ENDO
- ANOMALIES
- CHROMOSOMAL
- ACIDOSIS
- KETOSIS
- JAUNDICE

Outcomes

- # hospitalizations
- COMA
- Developmental Delay
- IQ(##)
- DISABILITY
- DISABILITY COMMENT
- DATE of DEATH

clinical comment

Treatments/Interventions

Background (probably unrelated to screening)

- | Meds | Feeding | Other |
|-----------------------------------|------------------------------------|--------------------------------------|
| DOPAMINE <input type="checkbox"/> | HYPERAL <input type="checkbox"/> | ECMO <input type="checkbox"/> |
| IODINE <input type="checkbox"/> | CARNITINE <input type="checkbox"/> | VENTILATOR <input type="checkbox"/> |
| ABX <input type="checkbox"/> | | SURGERY <input type="checkbox"/> |
| | | TRANSFUSION <input type="checkbox"/> |

Background Comments:

Screening/Dx-Related Intervention/Rx

Comments from CORE

COMMENT
12/8/2005 ICB REPORTED

Diagnostic Labs

Plasma
Acylcarnitines

102aGetGuthrie

RF1210050301
SC1213051562

Create/Edit/View New Contact, Baby Hx, Clinical | Create, Edit, or View Medical Visit and Labs | Long Term Follow-Up | Conversion to Metric

103bMedicalVisitForm

Visit/Sample Date: 12/15/2005 | Doctor LastName: HAY | Doctor Address 1 (Optional):
 Facility Category: Specialist | Doctor First Name: | Doctor Address 2 (Street):
 First Visit?: First Specialist | Doctor Telet: | Doctor Address 3 (Town, State, Zip):
 Baby Sick at Visit?: | Doctor Fax#: |
 Results/Eval Needed?: | Visit Comment: |

Lab Results of Samples Obtained at this Visit

Plasma Acyl Carn | Plasma AA | Urine Acyl Gly | Urine Organic Acids | Urine AA | Routine Labs | Specialty Labs

103cPAC_RESULTS_FORM

Plasma Acylcarnitines | PAC-Protocol: ARUP | Report Date: 12/15/2005
 All Plasma AcylC WNL: | NEED_PAC_RESULTS:

Interp	Results	CutOff	Ratio	Interp	Results	CutOff	Ratio
C2				C12:1			
C3				C12:0H			
C3:1				C14			
C3DC				C14:1			
C4				C14:2			
C4:0H				C14:0H			
C4DC				C14:10H			
C5				C16			
C5:1				C16:1			
C5:0H				C16:0H			
C5DC				C16:10H			
C6	P	0.87	0.16	5.44	C16DC		
C6:0H				C18			
C6DC				C18:1			
C8	P	3.41	0.21	16.2	C18:2		
C8:1				C18:0H			
C8DC				C18:10H			
C10	P	0.6	0.26	2.31	C18:20H		
C10:1	p1.			C18:1DC			
C12				Free C _n			
				Total C _n			

Plasma AcylCarn Comments: F/T = 1.2 (0.2-0.8)

Diagnostic Labs

Urine Acylglycines

102aGetGuthrie

PlasmaAcylCam PlasmaAA **UrineAcylGly** UrineOrganicAcids UrineAA RoutineLabs SpecialtyLabs

103dUAG_RESULTS_FORM

Urine Acylglycines UAG-Protocol **Mavc** NEED_UAG_ REPORT_DATE 12/15/2005

UAG_ALL_WNL

	Interp	Results	CutOff	Ratio		Interp	Results	CutOff	Ratio
EthylmalonicAcid					n-Hexanoylglycine	P	13.03	1.9	6.858
2-EthylmalonicAcid					n-Octanoylglycine				
2-MethylsuccinicAcid					3-Phenylpropionylglycine				
GlutaricAcid					Suberylglycine				
Isobutyrylglycine					trans-Cinnamoylglycine				
n-Butyrylglycine					DodecanedioicAcid				
2-Methylbutyrylglycine	P	0.24	0.3	0.8	TetradecandioicAcid				
Isovalerylglycine					HexadecandioicAcid				

UrineAcylGly Comments:

Record: 1 of 1

Diagnostic Labs

Specialty Labs



102aGetGuthrie

PlasmaAcylCarn | PlasmaAA | UrineAcylGly | UrineOrganicAcids | UrineAA | RoutineLabs | **SpecialtyLabs**

103gSPECIALTY_RESULTS_TBL

Specialty Testing Need any Specialty Labs Results from this visit?

DNA

	Protocol	Need	Date of Report	Results(0,1,2)	Allele 1	Allele 2
MCAD PCR	NBS	<input type="checkbox"/>	12/7/2006	1	985A.G	
MCAD Sequence	VANDERBILT	<input type="checkbox"/>	1/5/2006			G799A
VLCAD PCR		<input type="checkbox"/>				
VLAD Sequence		<input type="checkbox"/>				
DNA_LCHAD		<input type="checkbox"/>				
LCHAD Sequence		<input type="checkbox"/>				
DNA_TRIFCT		<input type="checkbox"/>				
Trifunct Sequence		<input type="checkbox"/>				

DNA_COMMENTS: VU MUTATION HAS BEEN NOTED PREVIOUSLY IN SYMPTOMATIC AND ASYMPTOMATIC PATIEN

FIBROBLAST ENZYME

ENZYME_FAOD_PROTOCOL	<input type="checkbox"/>	Need	Results
----------------------	--------------------------	------	---------

ENZYME_COMMENTS:

Record: 1 of 1

Long-term Follow-Up

LAST <input type="text"/>	FIRST <input type="text"/>	DOB <input type="text" value="2002"/>	GUTHRIE_NUMBER
MOTHER'S LAST <input type="text"/>	BABY_SYSKEY <input type="text" value="479317"/>		▶ GA0326021109
			A00403025004

[Create,Edit,View New Contact,Baby Hx_Clinical](#) |
 [Create,Edit,orViewMedicalVisit and labs](#) |
 [Long Term Follow-Up](#) |
 [Conversion to Metric](#)

NCHS - United States Clinical Growth Charts

[Age 1 Year](#) |
 [>1 to 3 Years](#) |
 [>3 to 7 Years](#) |
 [>7 to 13 Years](#) |
 [>13 to 18 Years](#) |
 [>18 to 25 Years](#)

FU07a [BoysGrowthChart 3-20](#) [GirlsGrowthChart 3-20](#)

Growth indicators at Yrs + Mos

Height (cm)	Weight(Kg)
<input type="text" value="110.6"/>	<input type="text" value="19"/>
Percentiles from Chart: <input type="text" value="90%"/>	<input type="text" value="75-90%"/>

Visits recorded for this age range -->

Visit/Sample D.
▶ <input type="text" value="6/7/2005"/>
<input type="text" value="6/7/2005"/>
<input type="text" value="12/29/2005"/>

Record:

Below Info Is Current As Of (if for some reason a blank below should not be assumed as "normal" as of this date, write "UNKNOWN" in appropriate comment)

Frequent Hospitalizations? Number of Hospitalizations in Time Frame (see Tab)

Reasons for hosp

SpecialNeeds?

Review of Systems (check if there was issue/problem with system during time frame(as of above date), and add comments)

Neurological

seizures? motor delays? receptive skills? Speech Delay?

Pulmonary

Cardiac

Renal

GI

Hepatic

ID

Endo

Metabolic

Other

NEW ENGLAND NEWBORN SCREENING PROGRAM

Cystic Fibrosis Database

LAST FIRST SEX DOB
 MOTHER'S LAST MOTHER'S FIRST BABY_SYSKEY
 MOTHER'S ADDRESS STATE
 MOTHER'S CITY
 MOTHER'S STATE MOTHER'S ZIP

Guthrie Numbers of all Specimens

GUTHRIE_NUMBER
<input type="text"/>
<input type="text"/>
<input type="text"/>

Enter CF in Babylist subform (in candidate disorder) so culture results can be entered!

NENSP Category and DNA Results

Category
 CF DNA1
 CF DNA2
 CF Poly T

Positive screen on repeat specimen? (initial specimen was WNL)

IRT Results by Specimen

BABY_SYSKEY	GUTHRIE	TEST_NAME	TEST_V	SPECIMEN_DATETIME
<input type="text"/>	<input type="text"/>	CF_CONCENT	29.6	10/19/2002 5:00:00 AM
<input type="text"/>	<input type="text"/>	CF_PERCENT	70.0	10/19/2002 5:00:00 AM
<input type="text"/>	<input type="text"/>	CF_CONCENT	145	11/2/2002
<input type="text"/>	<input type="text"/>	CF_PERCENT	99.8	11/2/2002
<input type="text"/>	<input type="text"/>	CF_CONCENT	50.7	12/1/2002 4:00:00 AM
<input type="text"/>	<input type="text"/>	CF_PERCENT	93.1	12/1/2002 4:00:00 AM

Record: of 6

Diagnostic Testing

CF Ctr Sweat at
 Initial Sweat Cl Last Sweat Cl
 Dx by Genotype only
 NENSP Dx
 Is child a borderline?
 Other Clinical Conditions/Diseases

Extended DNA Results

Lab
 CFTR1:
 CFTR2:
 CFTR PolyT:
 CFTR Poly:

CF Center Tracking

Current CF Center
 Ctr MR#
 Has child transferred centers (if ever)
 Transfer Ctr comment
 Was there a delay in diagnosis?

Cystic Fibrosis Database: Growth Measures

LAST FIRST SEX DOB
 MOTHER'S LAST MOTHER'S FIRST BABY_SYSKEY
 MOTHER'S ADDRESS STREET STATE
 MOTHER'S CITY
 MOTHER'S STATE MOTHER'S ZIP

Guthrie Numbers of all Specimens

GUTHRIE_NUMBER
<input type="text"/>

Enter CF in Babylist subform (in candidate disorder) so culture results can be entered!

NBS, Diagnostic Results | Family History | Complications, Outcomes | Growth | Culture Results | Comments from Core | Borderline | Medical Home | Other Comments/Info

On pancreatic enzymes NOT on pancreatic enzymes Pancreatic enzyme status unknown
 classified with nutritional failure (if ever)

fk baby:

Growth Measure 1 | Growth Measure 2

Measurement Date	<input type="text" value="12/6/2005"/>	Age (months)	<input type="text" value="44"/>
Ht (cm)	<input type="text" value="101"/>	Wt (kg)	<input type="text" value="17"/>
BMI	<input type="text" value="16.7"/>	Ht Z	<input type="text" value="0.196392574025"/>
Wt Z	<input type="text" value="0.686826251905"/>	BMI	<input type="text" value="0.770292154802"/>
Ht %	<input type="text" value="57.9"/>	Wt %	<input type="text" value="75.5"/>
BMI %	<input type="text" value="77.9"/>	% IBW	<input type="text" value="106.2"/>

If child is < 24 months old, BMI results are actually Weight-Height results and not actual BMI

Cystic Fibrosis Database: Culture Results

NBS, Diagnostic Results	Family History	Complications, Outcomes	Growth	Culture Results	Comments from Core	Borderline	Medical Home	Other Comments/Info
103bMedicalVisitFormJH								
Date of Culture		<input type="text" value="7/26/2002"/>	Comment					
Culture Type		<input type="text" value="unknown"/>						
<input checked="" type="checkbox"/> Are results from PORT CF Query?								
Organism Results from this culture								
CultureResults								
<input checked="" type="checkbox"/> Pseudomonas								
<input type="checkbox"/> Pseudomonas (mucoid)								
<input checked="" type="checkbox"/> Pseudomonas (non-mucoid)								
<input type="checkbox"/> Pseudomonas (mucoid unk)								
<input type="checkbox"/> Staph aureus								
<input type="checkbox"/> Cepacia								
<input type="checkbox"/> H flu								
<input type="checkbox"/> E coli								
Other Organisms		<input type="text"/>						
Culture Comment		<input type="text"/>						
Record: 1 of 13								

If culture results are from PORT CF Query then there could be other organisms present besides PA and Cepacia. Port CF Query was designed to query only for PA and/or Cepacia.

Hemoglobin Database

LAST FIRST SEX DOB:
 MOTHER'S LAST MOTHER'S FIRST VICTORIA
 MOTHER'S ADDRESS
 MOTHER'S CITY
 MOTHER'S STATE MA MOTHER'S ZIP

BABY_SYSKEY
 STATE MA

Guthrie Numbers of all Specimens

GUTHRIE_NUMBER
N
E

NBS, Diagnostic Results | **Outcomes** | **Family History** | **Growth** | **Events** | **Comments from Core** | **Medical Home** | **Other Comments/Info** | **Conversion to Me**

HGB NBS results

BABY_SYSKEY	GUTHRIE_	TEST_ \	SPECIM
00000007	E	FS	1:00 AM
00000007	E	FS	
*	(AutoNumber)		

Record: 1 of 2

HGB Confirmatory results

NBS result FS

Heme DX SS

Other

Enter HGB in Babylist subform (in candidate disorder) so events results can be entered!

Hematology Clinic

Has child trans clinic? (if ever)

Initial Clinic: UMass

Date of 1st Clinic Visit 8/22/2007

Current Clinic: UMass

date most recent clinic visit 1/27/2009

102dBabylist subform

Baby Syskey <input type="text"/>	Candidate Disorder <input type="text"/> HGB
----------------------------------	---------------------------------------------

fk baby: 00000007

Hemoglobin Database: Variables & Outcomes

LAST FIRST SEX DOB
 MOTHER'S LAST MOTHER'S FIRST BABY_SYSKEY
 MOTHER'S ADDRESS
 MOTHER'S CITY
 MOTHER'S STATE MOTHER'S ZIP
 STATE

Guthrie Numbers of all Specimens

GUTHRIE_NUMBER
N 1012007UC00
E 1001001UC01

[NBS, Diagnostic Results](#) | [Outcomes](#) | [Family History](#) | [Growth](#) | [Events](#) | [Comments from Core](#) | [Medical Home](#) | [Other Comments/Info](#) | [Conversion to Metric](#)

103bMedicalVisitFormJH

Event: Comment
 Clinic of
 Event D
 Event D
 Clinic Visit
 Radiological Stroke
 Clinic Visit
 Bacturemia
 Acute Chest

Normal Abnormal no comment
 Start Date:
 End Date:
 Comment:

Status of LTFU in New England States



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Lessons Learned from CF LTFU in Massachusetts

- In 2006 NENSP collected outcomes on 125 CF affected infants (born 1999-2004) identified by NBS

- Height & Weights (at 1 year of age & most recent)

- Culture Results (at 1 year of age & most recent) & later by organism cultured

- * Not all CF infants are in PORT CF (the CFF registry)

- * Center transfers offer a substantial challenge – local value

- ~ 20% of MA CF infants have transferred between the 5 MA CFF Centers

- * Value of raw data: quality standards in collection, data entry and data translation. (various tools used to calculate height-weight, BMI and % Ideal Body Weight)

- * Infection tracking confounded by successful treatments.

- Focus on a particular organism such as *B. cepacia* or *Pseudomonas* and collect all dates that child had positive culture for that particular organism