Adaptive Turnaround Documents, Newborn Screening and the Medical Home

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Children's Health Services Research
Regenstrief Institute

Overview Two Related Studies

- Regional Health Information Networks to Improve Newborn Screening Follow-up
- Data Standards to Share Newborn Screening Results

Supported by a grant from the Genetics Section, Maternal & Child Health Bureau, HRSA, through the Region 4 Genetic Collaborative

Expanded Newborn Screening

- Tandem Mass Spectrometry
- Over 50 Conditions
- Prevent Morbidity and Mortality
- Save Costs

Challenges

- Most Conditions Rare
  - MDs Unfamiliar
- Families Need Guidance
- Diagnosis & Treatment Must Be Timely
  - Who has the baby?
- No Mechanisms for LTFU

Solution

- Regional health information network
- Two way communication with providers

Project Objectives

- Adaptive turnaround document technology to facilitate communication: NBS program, subspecialists, medical home
  - Provide “just in time” information to the primary care clinician & family
  - Reduce the risk of “missed opportunities” to screen
  - Facilitate tracking of children with detected conditions

What ***is*** an Adaptive Turnaround Document (ATD)?

- Computer Generated
- Paper
- Delivers tailored information
- Scannable
- Captures structured data
  - Computer, scanner, or fax

Jennifer D. Patient
DOB: 14 May 2007
Newborn Screening Alert: Elevated C8 with Lesser Elevations of C6 and C10 Acylcarnitine

Suggestive of Medium-chain Acyl-CoA Dehydrogenase (MCAD) Deficiency

Condition Description: MCAD deficiency is a fatty acid oxidation (FAO) disorder. FAO occurs during prolonged fasting and/or periods of increased energy demands (fever, stress) when energy production relies increasingly on fat metabolism. In an FAO disorder, fatty acids and potentially toxic derivatives accumulate because of a deficiency in one of the mitochondrial FAO

Please check ALL of the following that apply:

[ ] Family contacted
[X] Newborn clinical status assessed
- **[ ] Problems (poor feeding, vomiting, lethargy, hypotonia, hepatomegaly)**
  - **→ [ ] Treated with IV glucose**
- **[X] Infant stable**

[X] Family provided attached educational materials

Diagnostic Evaluation

- **[X] Plasma acylcarnitine sent**
- **[X] Referral made to metabolic center**

[ ] Family could not be contacted
[ ] This is not my patient

[ ] Family provided attached educational materials

Diagnostic Evaluation
- **[ ] Plasma acylcarnitine sent**
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Generating the ATD

Knowledge Base

RULES:

IF lurelicubwliuc
THEN isjfafkjdf;lksdf
IF lurelicubwliuc
THEN isjfafkjdf;lksdf
IF lurelicubwliuc
THEN isjfafkjdf;lksdf
IF lurelicubwliuc
THEN isjfafkjdf;lksdf
IF lurelicubwliuc
THEN isjfafkjdf;lksdf
IF lurelicubwliuc
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MEDICAL EMERGENCY - TAKE THE FOLLOWING IMMEDIATE ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn (poor feeding, lethargy, hypotonia, hepatomegaly). If signs are present or infant is ill, initiate emergency treatment with IV glucose. Transport to hospital for further treatment in consultation with metabolic specialist. If infant is normal initiate timely confirmatory/diagnostic testing, as recommended by specialist.
- Educate family about need for infant to avoid fasting. Even if mildly ill, immediate treatment with IV glucose is needed.
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma acylcarnitine analysis will show elevated **octanoylcarnitine (C8)**. Urine acylglycine will show elevated **hexanoylglycine**. Diagnosis is confirmed by mutation analysis of the **MCAD gene**.

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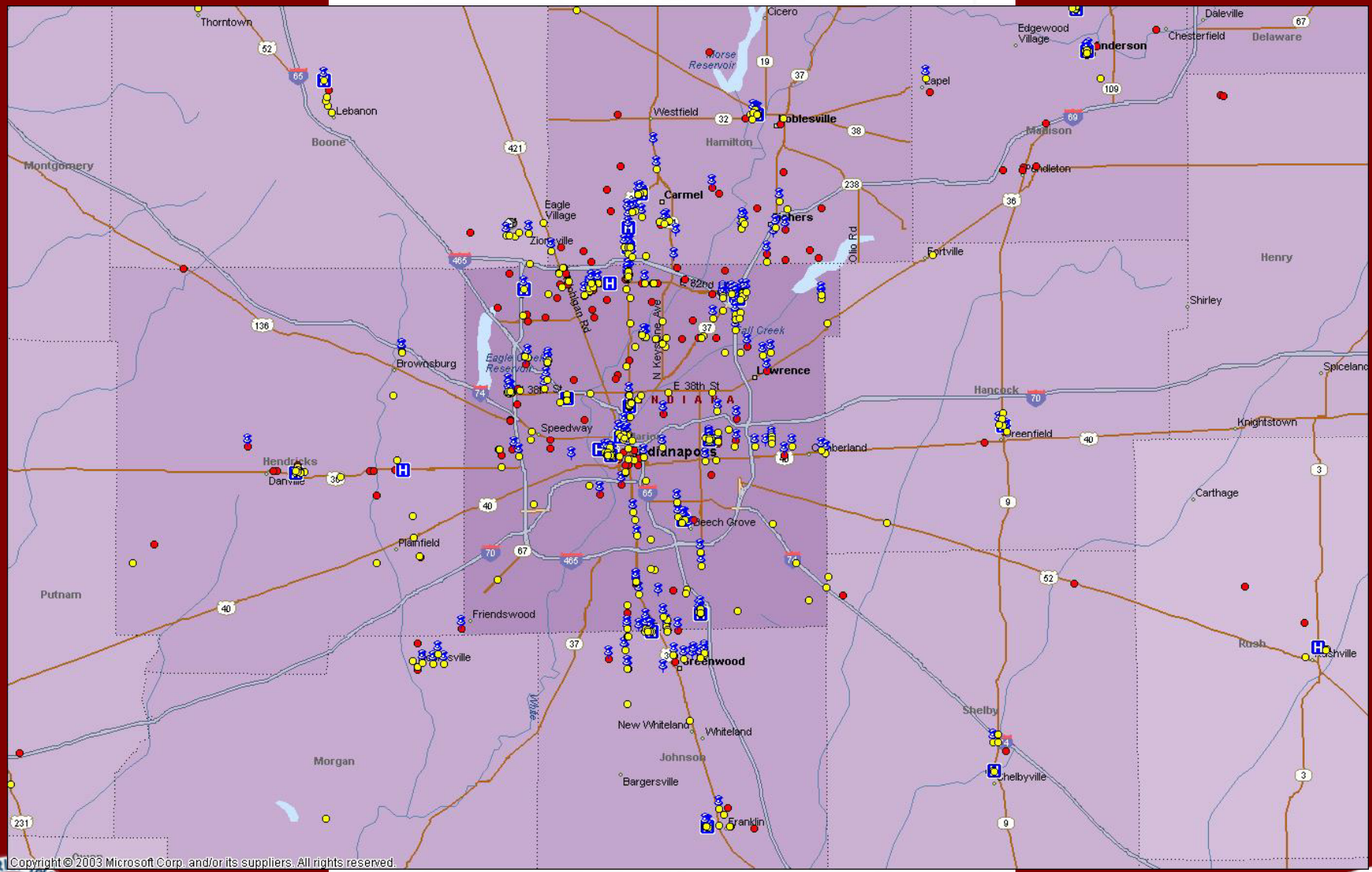
Applying ATD Technology on a Regional Basis

- Indiana Network for Patient Care (INPC)
- 5 hospital systems, 15 hospitals
- County and State Health Departments
- RxHub
- Medicaid administrative data
- 660 million results


Thorntown
Lebanon
Boone
Montgomery
Cicero
Morse Reservoir
Westfield
Cobblesville
Hamilton
Carmel
Eagle Village
Zionsville
Fishers
Edgewood Village
Anderson
Chesterfield
Daleville
Delaware
Lapel
Madison
Pendleton
Fortville
Henry
Shirley
Spiceland
Hancock
Knightstown
Greenfield
Carthage
Rush
Shelby
Shelbyville
Browns burg
Lawrence
E 38th St
INDIANA
Speedway
Indianapolis
Beech Grove
Greenwood
Hendricks
Danville
Plainfield
Putnam
Friendswood
New Whiteland
Whiteland
Johnson
Bargersville
Franklin
Morgan
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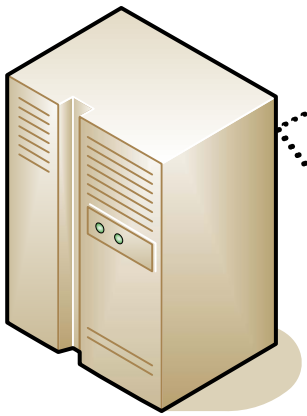
How it was accomplished (technical)

- Federated repository
- Data from difference sources are stored in separate physical files
- Global patient index
- Same data structure and data dictionary
- Data arrive by HL7 (also DICOM, NCPDP)
  - Parsed
  - Translated

Consolidating the Silos

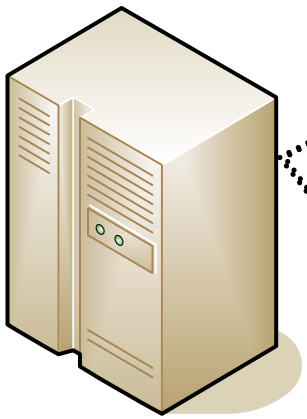


Immunization Registry

Jane Doe's Immunizations:

3/1/04	DipTetaPur	30936-9
3/1/04	HemInfB	30938-5
3/1/04	PolioVir	33555-4
3/1/04	HepaB	30937-7

DTaP Dose Count:	30936-9
HIB Dose Count:	30938-5
IPV Dose Count:	33555-4
VZV Dose Count:	30943-5
MMR Dose Count:	30940-1
HepB Dose Count:	30937-7



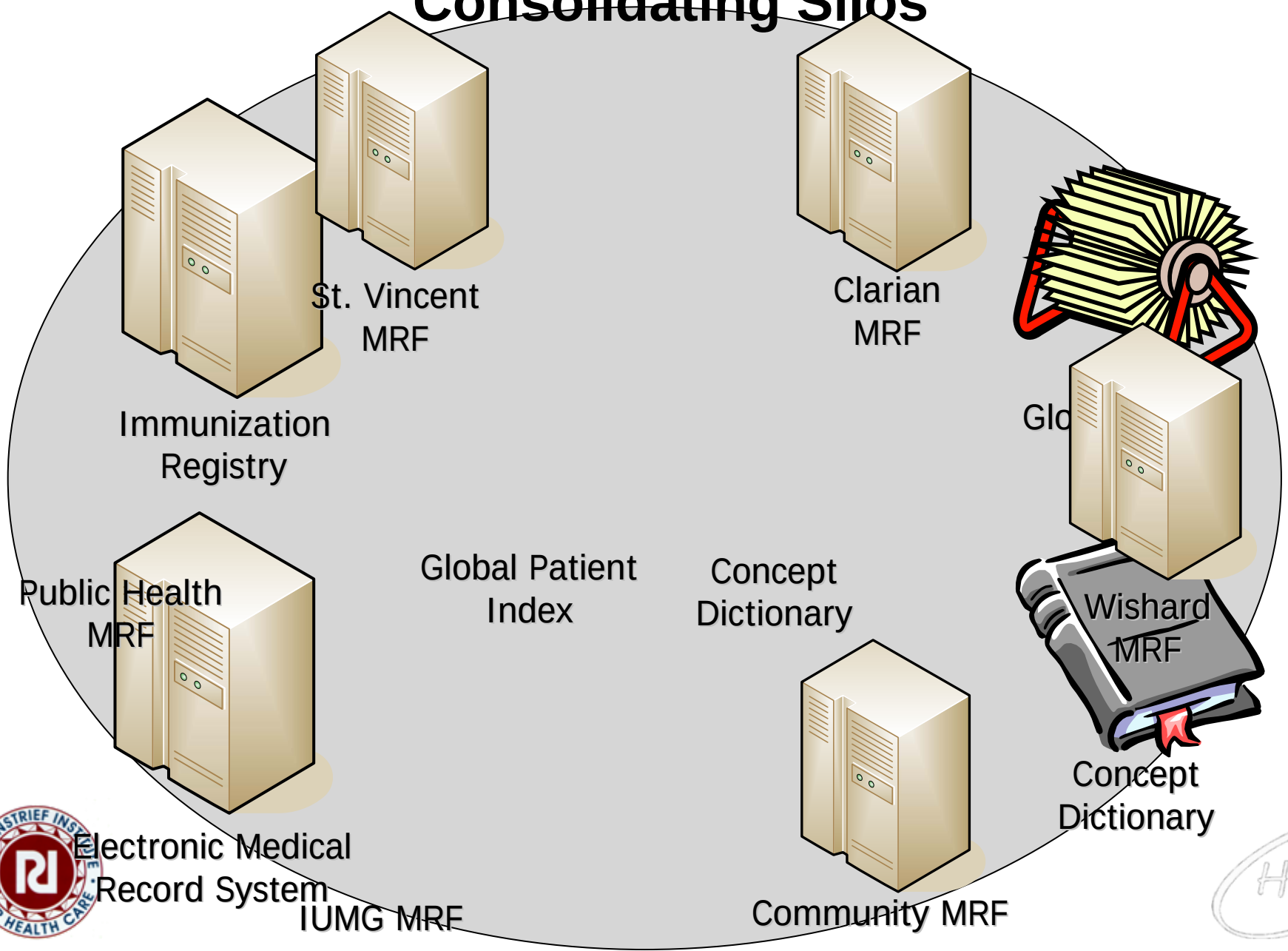
Electronic Medical Record System

Jane Ellen Doe's Shots:

5/1/04	DTaP Imm	30936-9
5/1/04	HIB Imm	30938-5
5/1/04	IPV Imm	33555-4
7/9/04	DTaP Imm	30936-9
7/9/04	IPV Imm	33555-4

Global ID:	45[illegible]8
Name:	Jane Ellen Doe
Lots of Demographics..	
MRF1 ID:	OU81247
MRF2 ID:	4564356
PH MRF ID:	123LMNOP
MRF3 ID:	6789XYZ

The Indian Network for Patient Care
Consolidating Silos



Docs4Docs - Links to clinical providers

- Results delivery service
- All hospitals send reports through same mechanism (HL7)
- HL7 results messages flow through INPC and are routed to MD in-boxes or faxes
- Eliminates mail costs

An HL7 Message

OBX = Observation

493.90^493.90 ASTH W/O STAT ASTHM NOS^I9

Code number

Short Name

Vocab (ICD-9)

DCT|1|074.3^074.3 HAND, FOOT
opathy^DCT~9985^conjunctivitis
493.90^493.90
NOS^ ~493.91^493.91 ASTH A W STATUS
AS 2^reactive e^DCT~
OT S^I9~510 T~2297
nos RI^DCT~8 ~466.19
BR TH ORG TRACH S
DIS asthma^D .03 VO
ALONE^I9~780.39^780.39 CONVULSIONS
NEC^I9~2257^constipation^DCT~6694^croup^DCT~486^486
PNEUMONIA, ORGANISM NOS^I9~2202^otitis
media^DCT~9936^Tinea capitis^DCT||||||F|||200411031020

Three Ways ATDs Can Enhance the NBS Program

- Just-in-time information to the medical home (physicians and families)
- Prevent missed opportunities to screen
- Long term tracking of children with identified conditions

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Indiana State Department of Health

October 2, 2006

Jennifer Johnson
100 Toddler Rd
Lebanon, IN 46078

Re: **Newborn screening test of your baby born September 29, 2006**

Just-in-Time Information for the Family

Dear Ms. Jonnson:

Your baby had a blood test shortly after birth to look for a number of inherited medical conditions which require early treatment. The purpose of this newborn screening test is to detect disorders before they can cause problems for your child. Your baby had a positive test for one these disorders: it is called congenital adrenal hyperplasia, or CAH. This booklet is provided to tell you about this disorder.

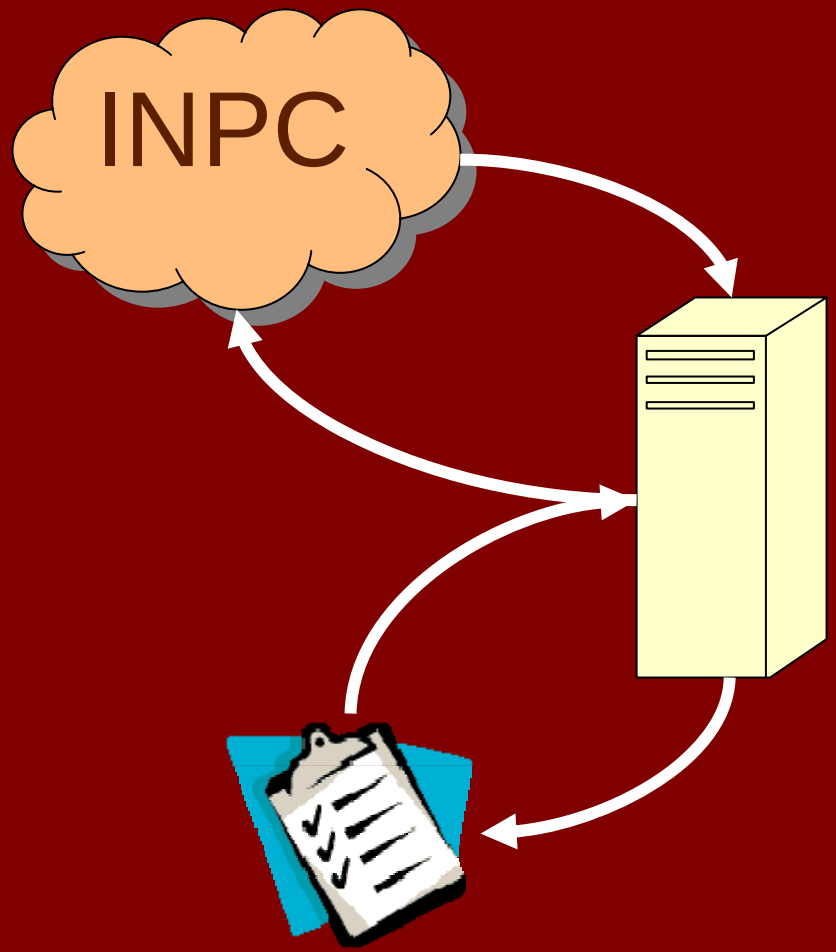
First, an explanation of the screening test. The screening test does not prove that your baby has CAH. More tests will be needed to prove its presence. These blood tests can be performed by a children's hormone specialist (a pediatric endocrinologist) experienced in the diagnosis and care of children with CAH. **It is important to have your child tested as soon as possible.** Untreated CAH can result in serious illness and, in some cases, death. In many cases, the retest will show that CAH is not present. Some reasons why babies without CAH may test positive on the newborn screening test include improper timing of the screening test, unusual health conditions of the baby in the first days of life, and/or improper specimen collection and processing.

It is important for you to know that CAH is a completely treatable medical condition. Other than having to take daily medication, the child with CAH can have a completely normal life.

Erica Eugster, MD
Riley Hospital for Children

Avoiding Missed Opportunities

- Capture HL7 message for neonates
- Check against newborn screening reports
- Alert physician if missing or positive



HL7 Message

MSH|^~\&|RLY_NB|RILEY|NEWBORN|REGEN|200512020702 51||ORU^R01|||2.3
PID|||72360601||Doe^John||20050908155100|M|Jones|White||||||||||Non Hispanic
NK1||Doe^Jane|Mother|123 MAIN
STREET^^GREENWOOD^IN^46143^^^^JOHNSON|3175350809|||||||||19791116
PV1|||||||^SMITH^LYNETTE

Infant LName = Doe
Infant FName = John
Infant DOB = 9/8/2005
Infant Sex = Male
Infant Race = White
Infant Ethnicity = Non Hispanic
Mother LName = Doe
Mother FName = Jane
Mother Street = 123 Main Street
Mother City = Greenwood
Mother State = IN
Mother Zip = 46143
Mother County = Johnson
Mother Phone = 317-535-0809

Infant LName = Doe
Infant FName = Baby
Infant DOB = 9/8/2005
Infant Sex = Male
Infant Race = White
Infant Ethnicity =
Mother LName = Doe
Mother FName = Jane
Mother Street = 123 Main Street
Mother City = Greenwood
Mother State = IN
Mother Zip = 46143
Mother County =
Mother Phone = 317-555-1828

Probabilistic Scoring Example AGREEMENT RATES

- 2 files, 10 Records each
- Form all possible pairs: 10 x 10 = 100 pairs
- Human review of all 100 record pairs shows that 10 are true-links, 90 are non-links.

Probabilistic Scoring Example AGREEMENT RATES

- Among the 10 true-links, the last names agreed in 9/10 pairs (e.g. one of the last names was misspelled)
- This represents a 90% AGREEMENT RATE for last name among TRUE LINKS.
- Similarly, among the 90 non-links, last names agreed (by random chance) in 2/90 pairs
- This represents a 2% AGREEMENT RATE for last name among NON-LINKS.

Probabilistic Scoring Example AGREEMENT RATES

—— = 45

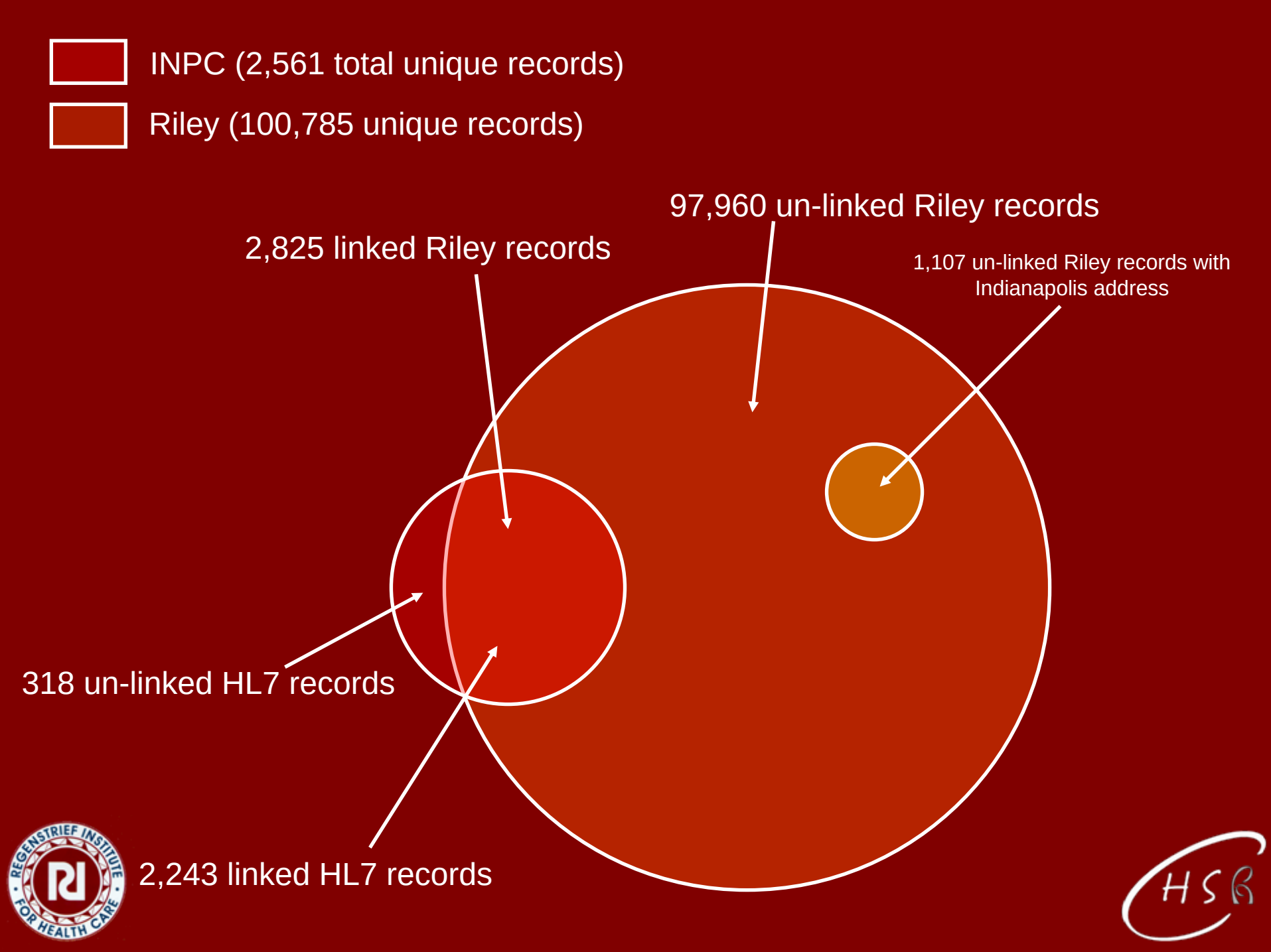
- Records that agree on last name are 45 times more likely to be a true-link than a non-link

- Weights for each field are combined to form a composite record pair score.

90%

- Field disagreement contributes a negative weight, and reduces the overall record pair score.

2%


INPC (2,561 total unique records)
Riley (100,785 unique records)
97,960 un-linked Riley records
2,825 linked Riley records
1,107 un-linked Riley records with Indianapolis address
318 un-linked HL7 records
2,243 linked HL7 records

Is Jennifer still in your care? [_]YES [_] NO

If NO, please provide any contact information you may have

Street address [_|_|_|_|_|_|_|_|_|_|_|_|_|_|_|_|_|_|_]
[_|_|_|_|_|_|_|_|_|_|_|_|_|_|_|_|_|_|_]

City, state, zip [_|_|_|_|_|_|_|_|_|_|_|_|_|_|_|_|_|_|_], [_|_] [_|_|_|_|_|]

Phone ([_|_|_]) [_|_|_]-[_|_|_|_]

Data last seen (mm/dd/yy): [_|_]/[_|_]/[_|_]

Development:	Normal	Concerning
Gross motor	[_]	[_]
Fine motor	[_]	[_]
Personal-Social	[_]	[_]
Language	[_]	[_]

Growth:

Height	[_|_|_].[_]	[_] inches	[_] centimeters
Weight	[_|_|_].[_]	[_] pounds	[_] kilograms

Has Jennifer been hospitalized in the last year? [_]YES [_] NO

If yes, how many times? [_|_|_]

Weight [_|_|_].[_] [_] pounds [_] kilograms

Has Jennifer been hospitalized in the last year? [_]YES [_] NO
If yes, how many times? [_|_|_]

LOINC: Logical Observation Identifiers Names and Codes

- Most clinical labs use HL7, but idiosyncratic codes for tests
- LOINC codes are universal identifiers for laboratory and other clinical observations
- Facilitate the exchange and pooling of results
- Interdigitates with SNOMED

Current Coding

- Idiosyncratic
  - Each lab has its own system
- Condition based
  - Original data “lost”
  - Interpretation dependent (non-standard)

Current Coding

C10:2	Confirmation	C10:2-C-01-003	2,3 Dienoyl-CoA Reductase Deficiency	The above results are suggestive of 2,3 Dienoyl-CoA Reductase Deficiency. An immediate recollection is necessary to further evaluate this infant.	Increased	< 0.40 UMOL /L
C10:2	Confirmation	PrePos 2,3DCA	Presumptive Positive 2,3DCA	The above results are suggestive of 2,4-Dienoyl-CoA Reductase Deficiency. An immediate recollection is necessary to further evaluate this infant.	Increased	< 0.40 UMOL /L
C12	Confirmation	C12-C-01-003	Increased	The above results indicate an abnormal acylcarnitine screen. A recollection is necessary to further evaluate this infant.	Increased	< 0.81 UMOL /L
C12:1	Confirmation	C12:1-C-01-003	Increased	The above results indicate an abnormal acylcarnitine screen. A recollection is necessary to further evaluate this infant.	Increased	< 0.60 UMOL /L
	Confirmation	C14-C-01-003	Increased	The above results indicate an abnormal acylcarnitine screen. A recollection is necessary to further evaluate this infant.	Increased	< 0.90 UMOL /L

LOINC Codes

45198-9	C10:1	Bld dot	x	decenoylcarnitine		Micro mole/L
	C10:1DC	Bld dot	x	dehydrosebacylcarnitine	dicarboxydecenoyl carnitine	Micro mole/L
	C10:2	Bld dot	x	decadienoylcarnitine		Micro mole/L
	C10:3	Bld dot	x	decatrienoylcarnitine		Micro mole/L
	C10DC	Bld dot	x	sebacylcarnitine	Dicarboxydecanoyl carnitine	Micro mole/L

Advantages of LOINC

- Accepted coding standard by ONCHIT
  - Necessary to share data nationally
- Data supporting interpretation are shared
- National cohorts based on analytes
  - Pooled data for optimizing cut-offs
  - Recruitment for trials based on analytes
  - Natural history studies with consistent case definitions

stmdowns@iupui.edu

www.ichsr.org