



Newborn Screening for SCID: New England Update

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July 30th-Aug 1st, 2015

APHL/NewSTEPS National SCID In-Person Meeting

Bethesda, MD



The Massachusetts SCID NBS Workgroup

Representatives from Newborn Screening, Immunology,
Infectious Disease, Public Health and Transplantation

Baystate  Children's Hospital



Dr. Alicia Johnston

Dr. Ellen Rae Cooper

Dr. Alfred DeMaria

Dr. Tony Bonilla

Dr. Luigi Notarangelo

Dr. Sung-Yun Pai

Dr. Cody Meissner

Dr. Paul Hesterberg

Dr. Mark Pasternak

Dr. Jolan Walter

Dr. Beverly Hay

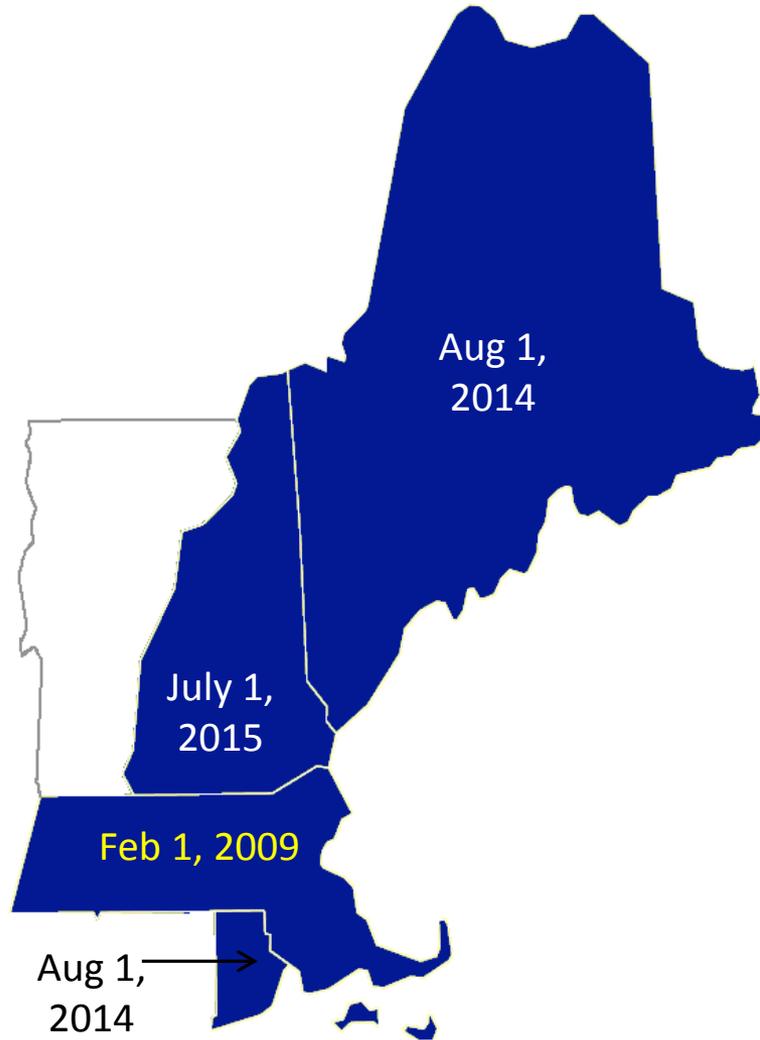
Dr. John Sullivan

Dr. Anne Marie Comeau

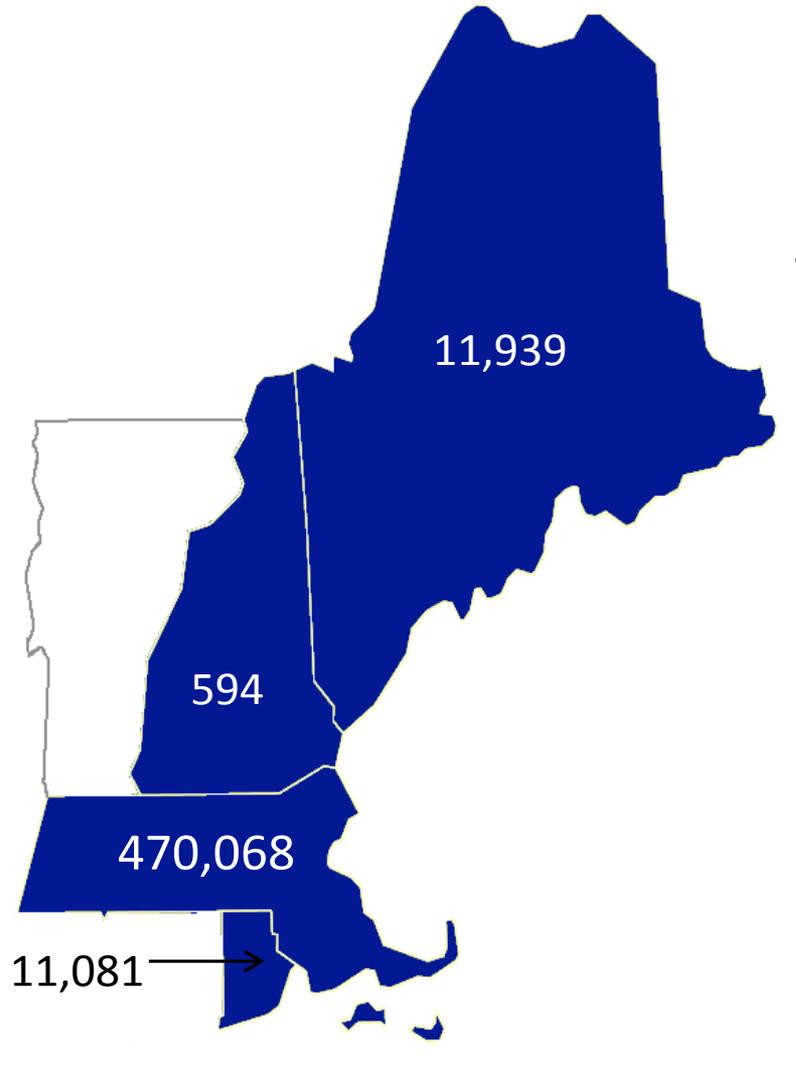
Dr. Roger Eaton

Dr. Inderneel Sahai

SCID NBS Start Dates in NENSP Partner States



Number of Babies Screened for SCID in NENSP Partner States



Through
7/17/2015

New England Newborn Screening Program

Four State Data and Experience

493,682 infants screened for SCID

4 SCID

~1:123,420

(Plus 1 Leaky SCID)

~1:98,736

Through
7/17/2015

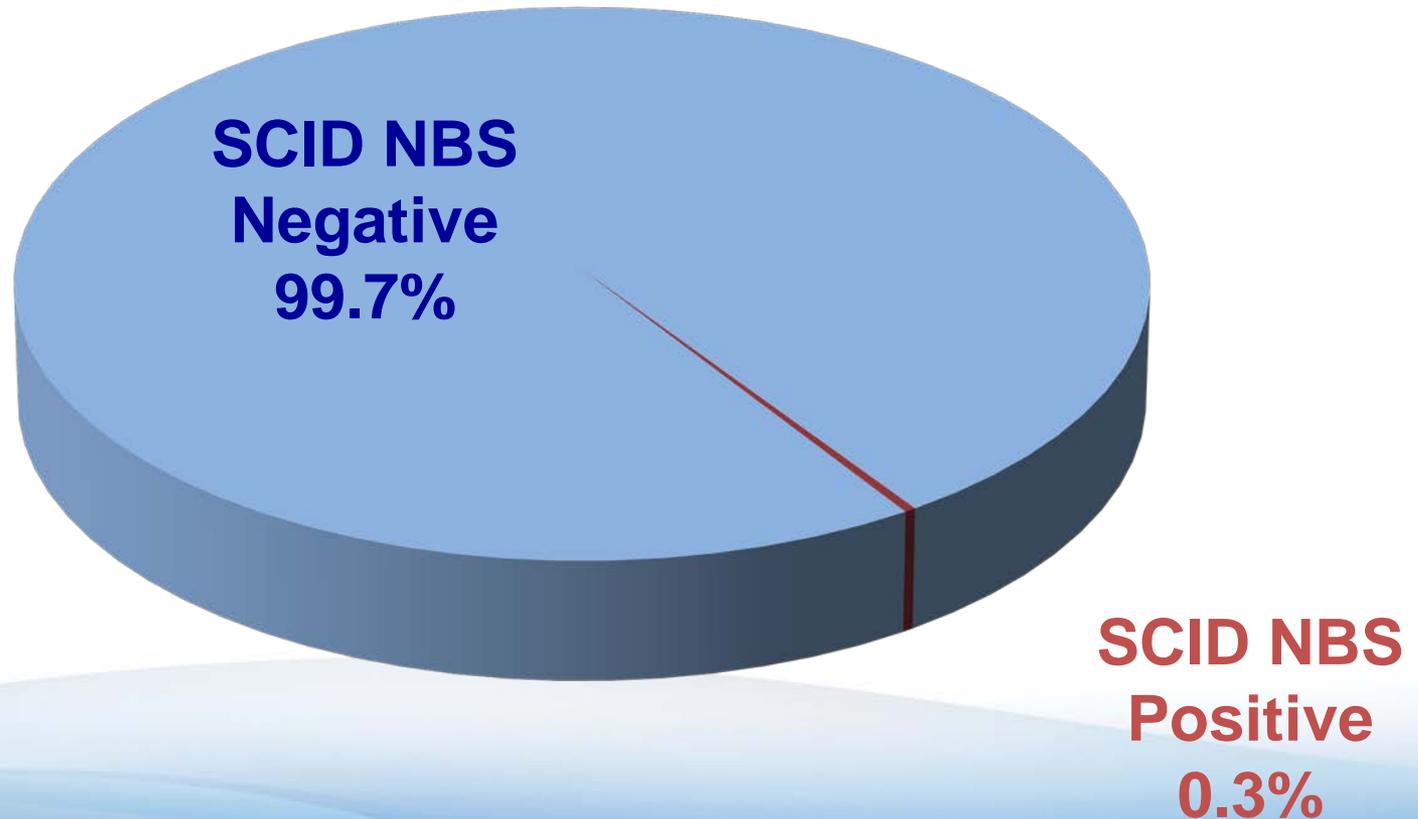
New England Newborn Screening Program

Massachusetts

New England Newborn Screening Program

470,068 infants screened for SCID (MA)

- 1,450 infants with positive SCID NBS result on any specimen



Through
7/17/2015

New England Newborn Screening Program

470,068 infants screened for SCID (MA)

- 1,450 infants with positive SCID NBS result on **any** specimen
 - 147 infants referred to Flow Cytometry* (~1/3,200)

4 SCID

~1:118,000

(1 additional baby with leaky SCID ~1:94,000)

1 additional baby with complete DiGeorge Syndrome

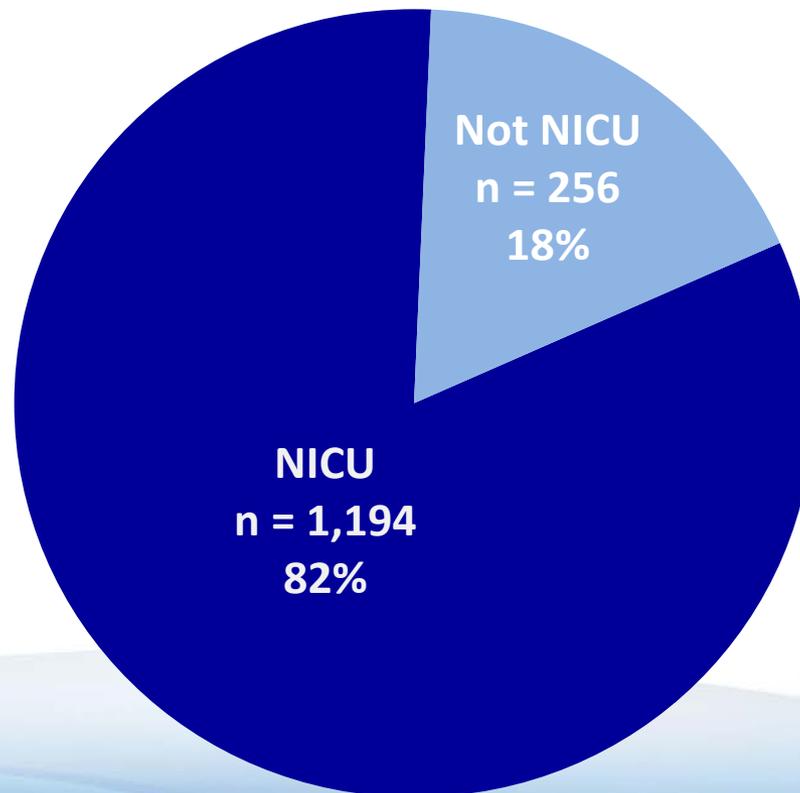
*by current algorithm

Through
7/17/2015

New England Newborn Screening Program

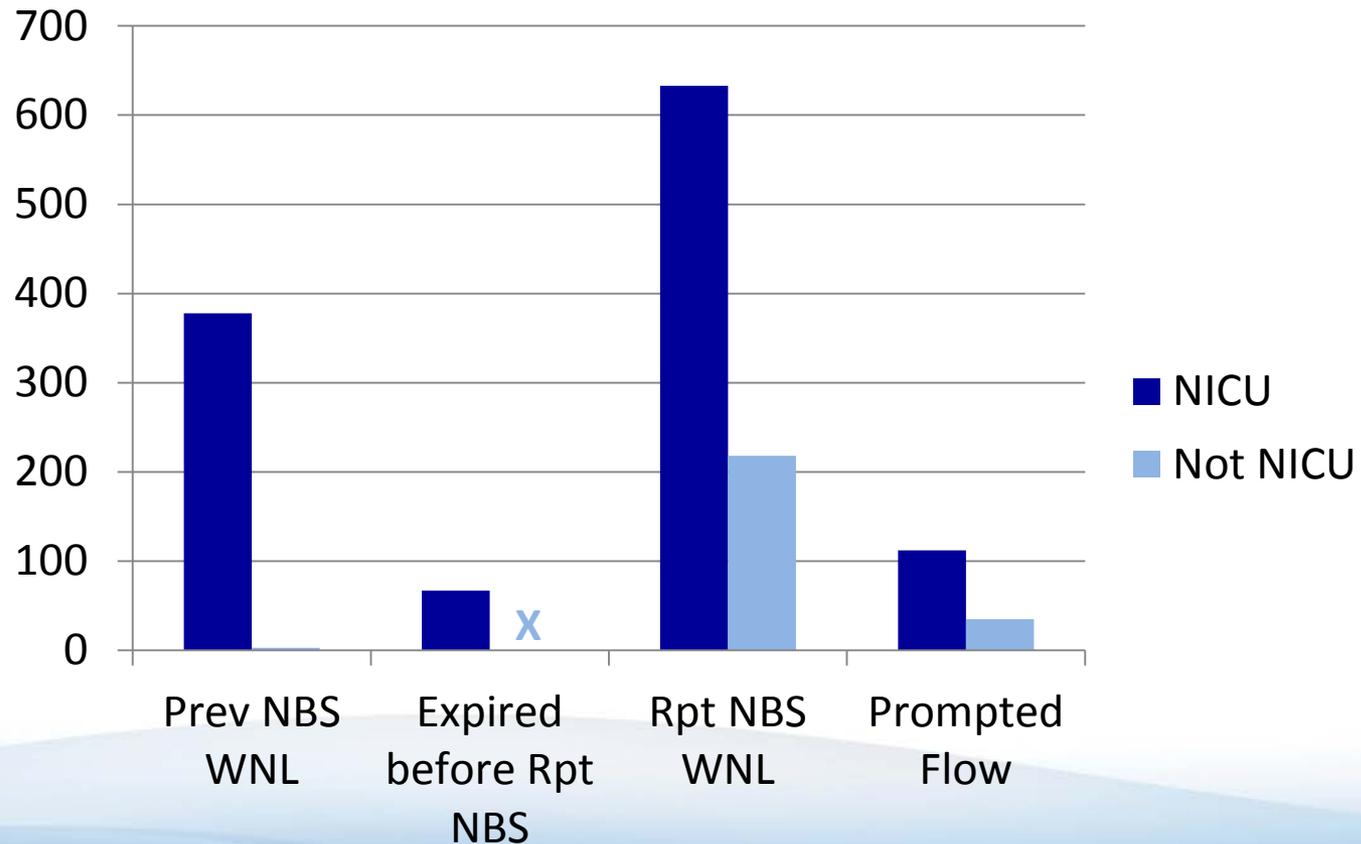
NICU vs. non-NICU status

in 1,450 infants with a positive SCID NBS
on any NBS specimen

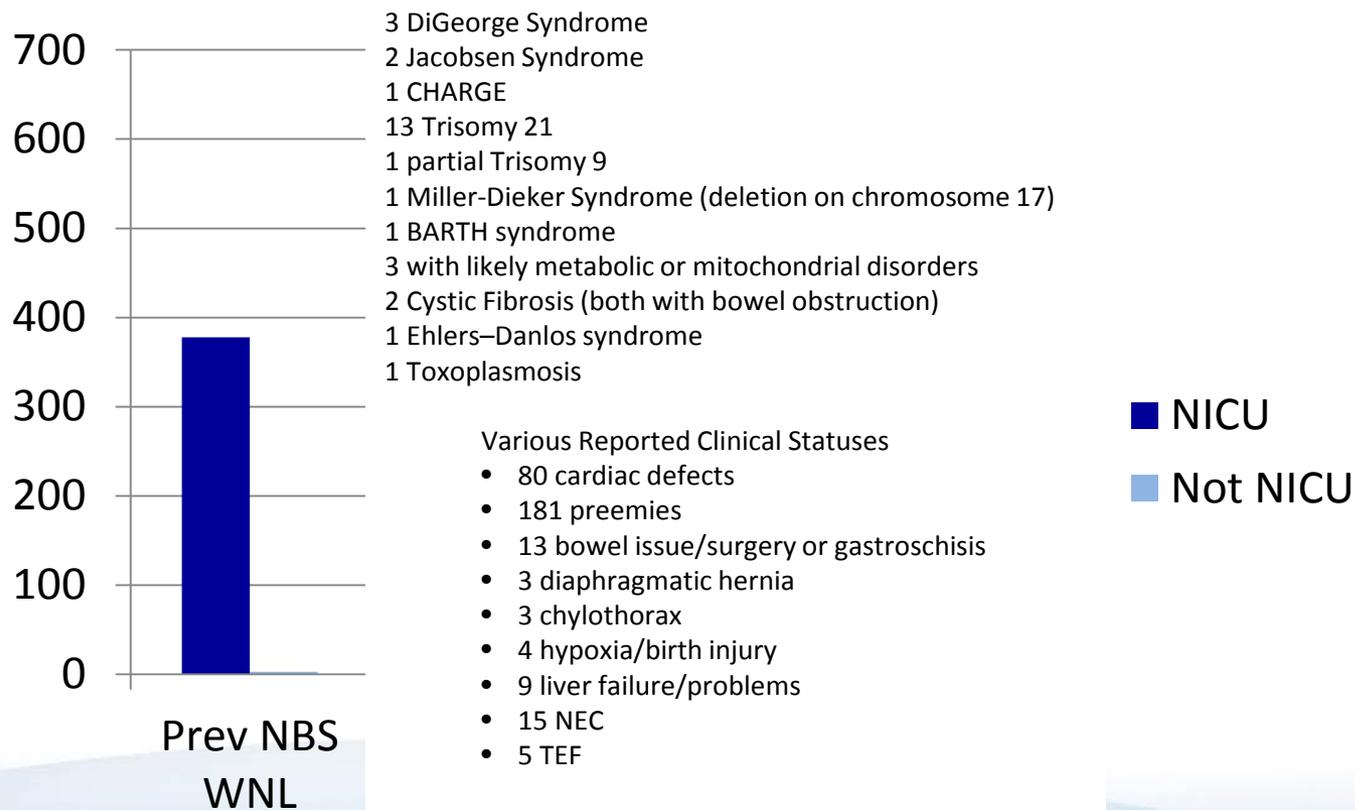


Through
7/17/2015

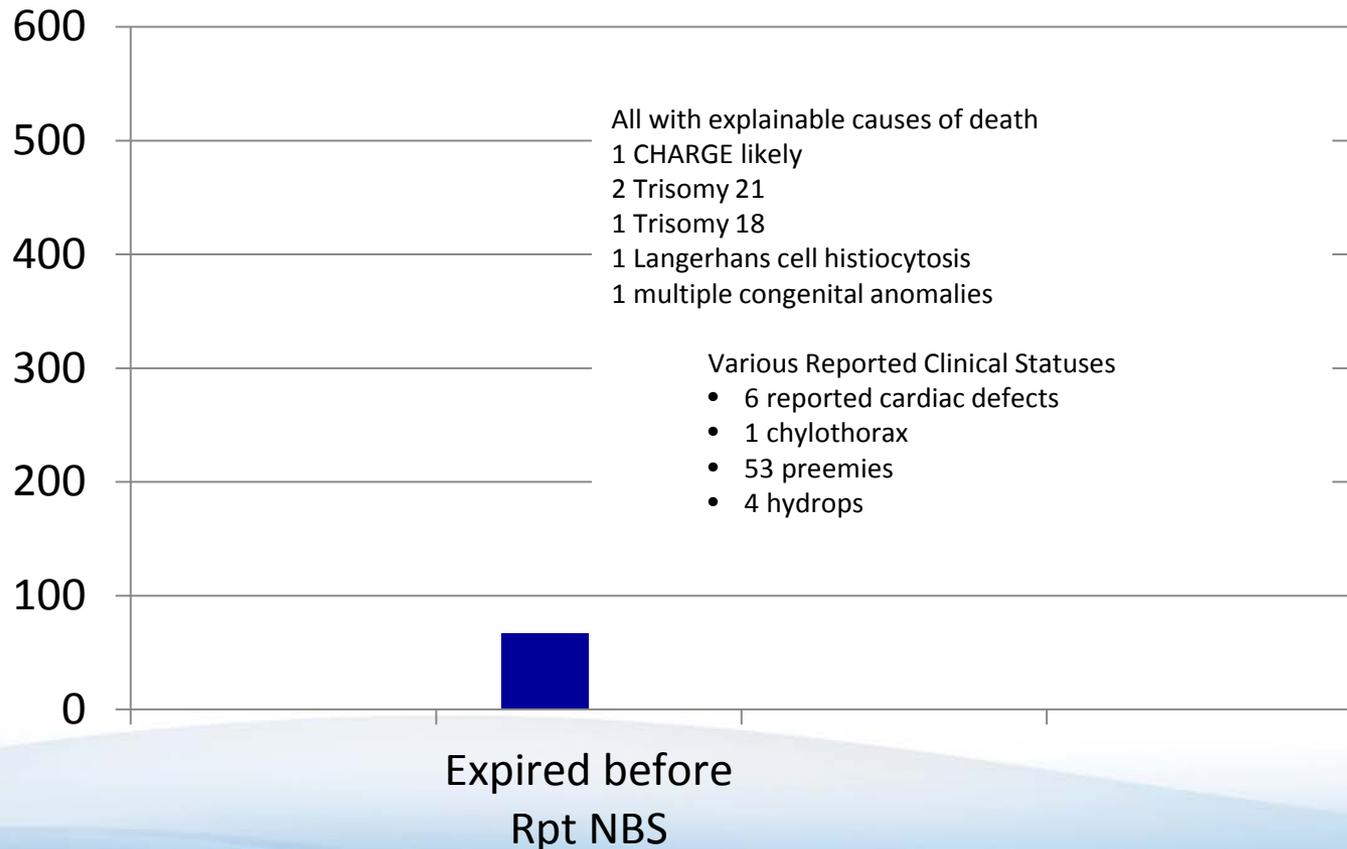
Final Screening Outcomes of 1,450 infants with any positive SCID NBS result



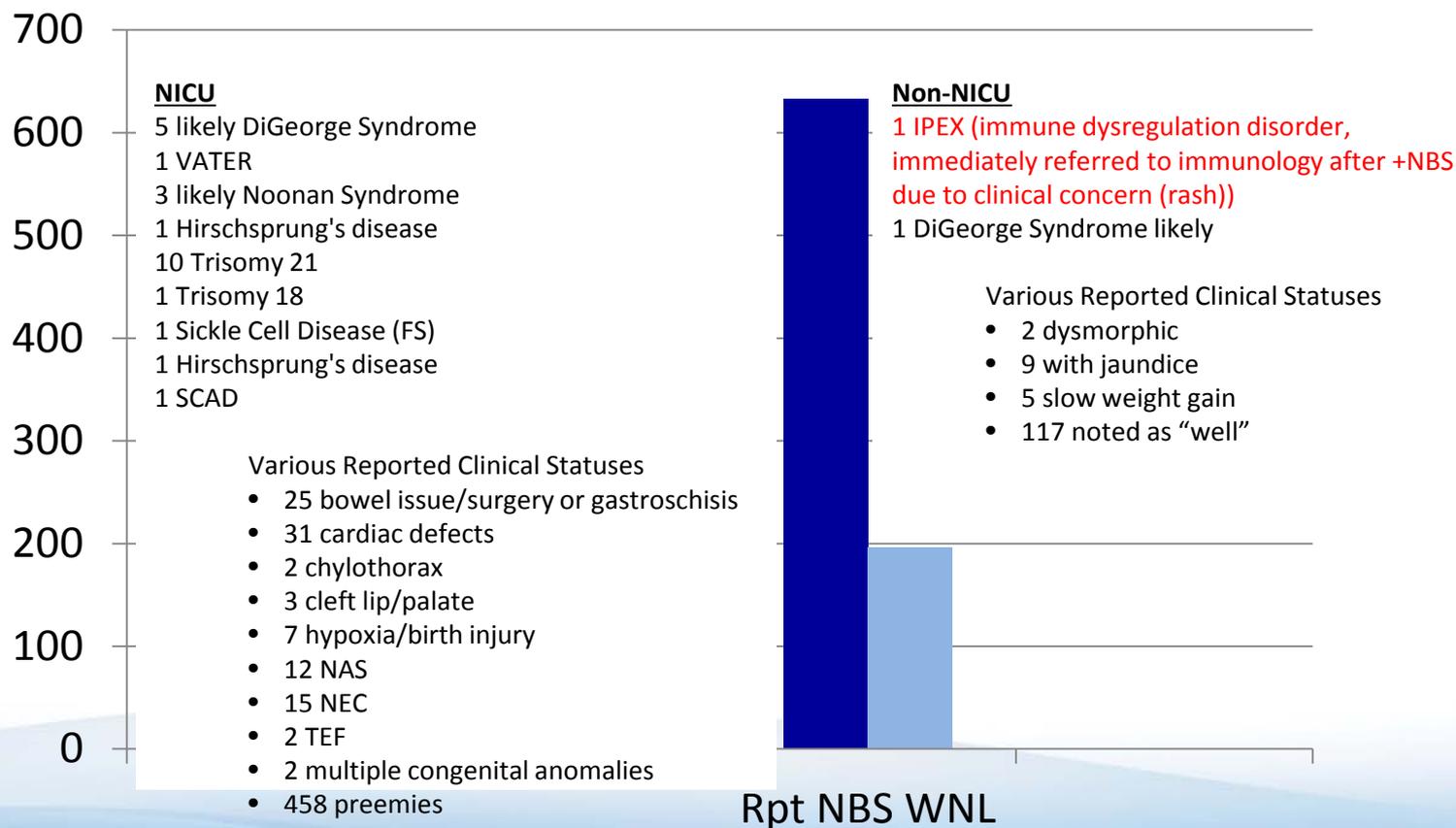
Final Screening Outcomes of 1,450 infants with any positive SCID NBS result



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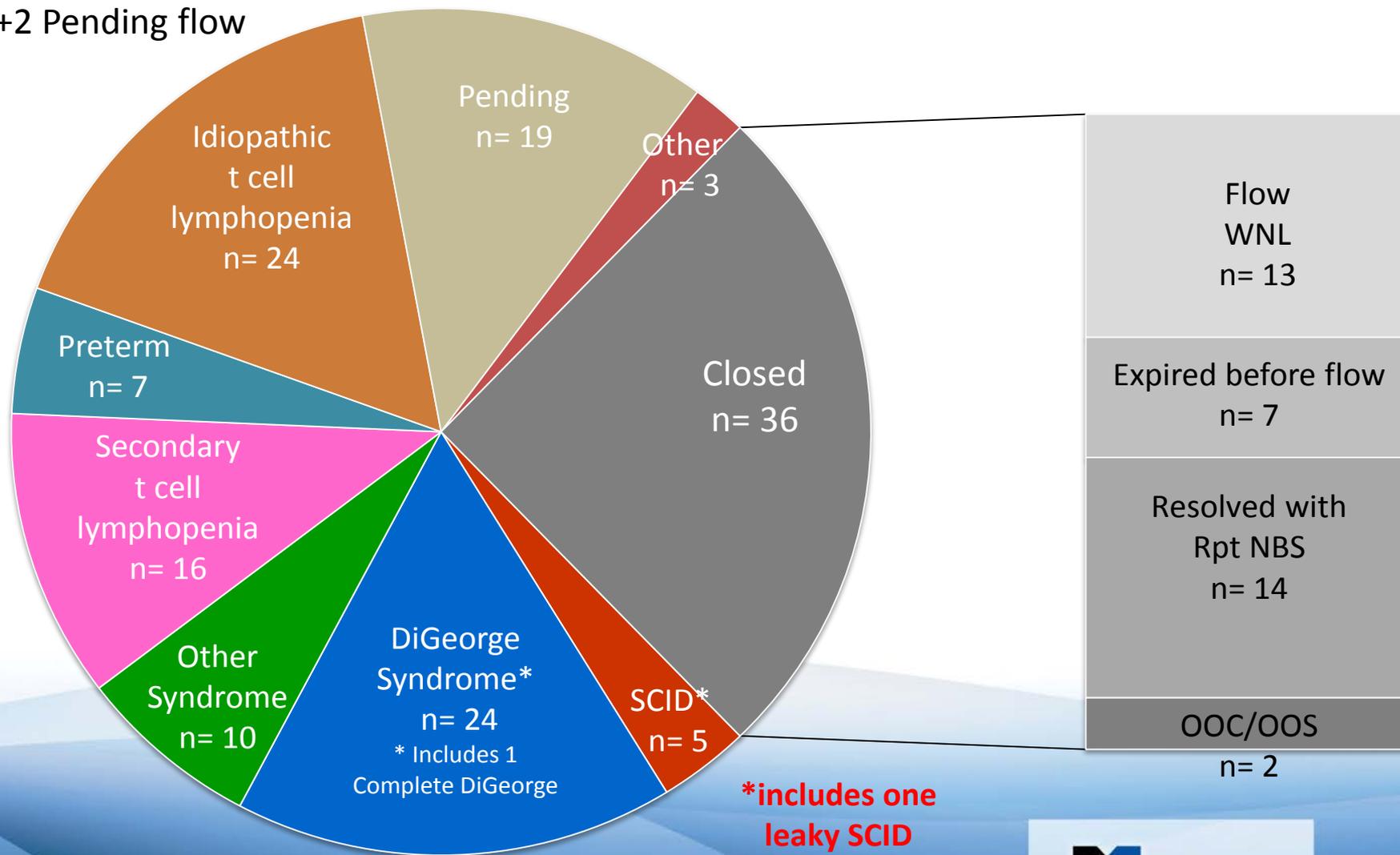


Final Screening Outcomes of 1,450 infants with any positive SCID NBS result



Status of 147 Infants Prompting Flow Cytometry

+2 Pending flow



TREC Results of Infants to Flow

Prompting flow
(either undetectable or 2 OOR TRECs)

n= 147

~1 in 37 risk for SCID

Undetectable TRECs

n= 27

	SCID	Not SCID
NICU	1	18
Not NICU	2	6

Never Normal TRECs

n = 120

	SCID	Not SCID
NICU	1	93
Not NICU	0	26

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~1 in 9 risk if undetectable

~1 in 4 risk if not NICU & undetectable

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n = 120

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Positive Predictive Values of SCID NBS

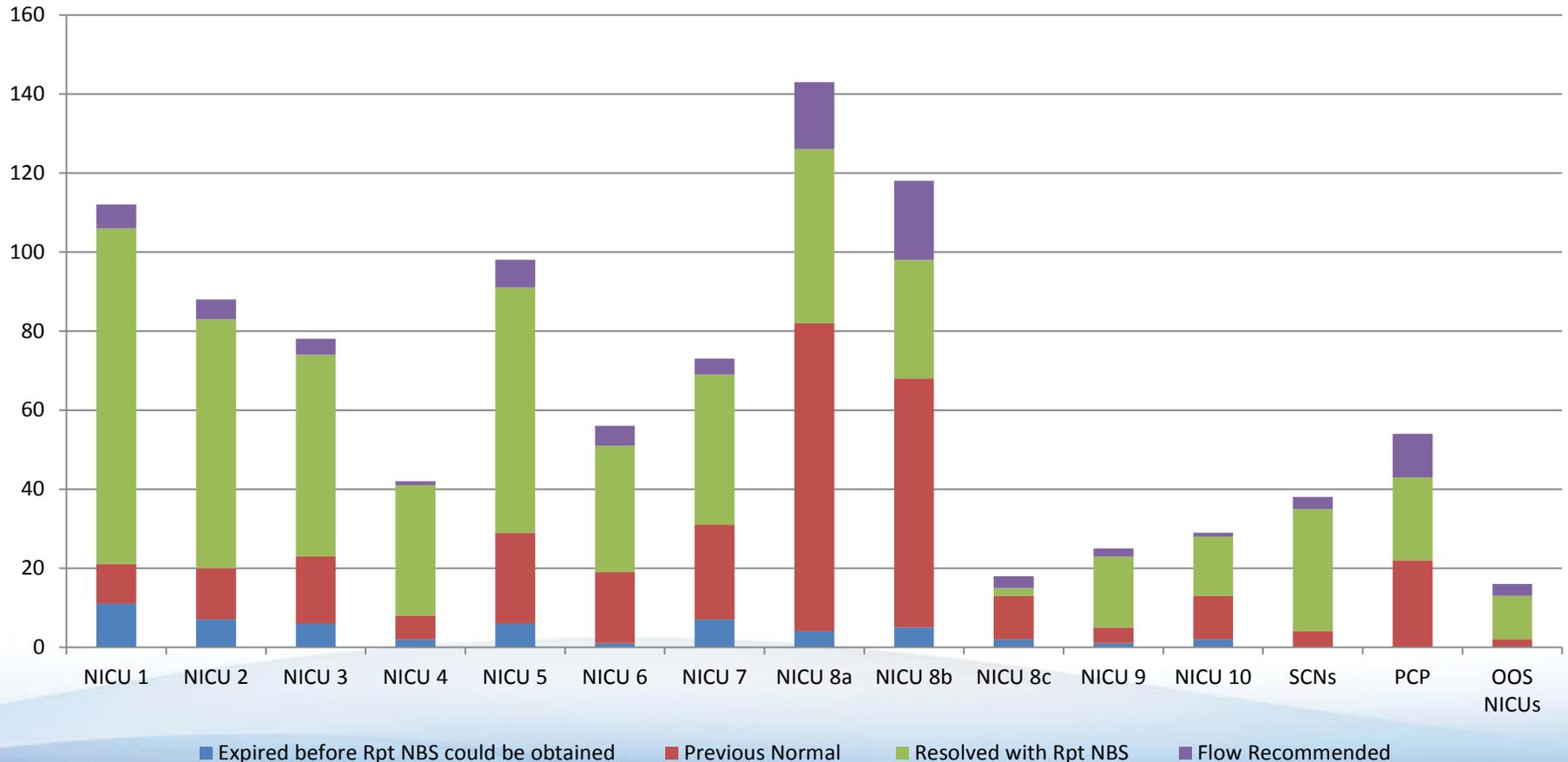
147 infants referred to Flow Cytometry*

PPV for SCID/Leaky SCID:	4/147	3%
PPV for TCL:	105/120	88%

*by current algorithm

NICU contacts (babies)

by NICU that took report of initial OOR result



TRECs and Gestational Age (GA)

Review of ~14,300 infants

TREC results linked to GA at time of specimen collection

	TRECs (copies/ul)
Overall Median	1,504
Gest Age <37 weeks	1,090
Gest Age \geq 37 weeks	1,527

98% of babies with a gestational age <37 weeks had WNL TREC results

99.9% of babies with a gestational age \geq 37 weeks had WNL TREC results

Acknowledgements



Jaime Hale
Jacalyn Thompson
Lan Ji
Jolanta Kordowska
Samantha Barton
Claire Hughes
Jane Griffin

