

California Newborn Screening Report:
Update on Quality Improvement
And Current Program Expansion

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HARBOR-UCLA PEDIATRICS

Timeliness Issues in NBS

- Newborn Screening (NBS) collection and transit timeliness came under scrutiny as a result of public comment in 2013;
- Disparities in NBS practices affecting screening timeliness became apparent;
- The ACHDNC recommended a series of timeframes for NBS- from collection, to transit, to testing and reporting.

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Newborn Screening in California

Over 80 disorders are screened from 6 circles of blood

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Screening in California

Year	Disorders Screened
2018	MPS I & Pompe Disease
2016	Adrenoleukodystrophy (ALD)
2012	Severe Combined Immunodeficiency (SCID)
2007	Cystic Fibrosis & Biotinidase Deficiency
2005	CAH & Metabolic disorders detectable by MS/MS Screening
1999	Hb H & Hb- H Constant Spring
1990	Sickle Cell & Other Hb
1980	PCH & Galactosemia
1980	PKU

SMA approved projected start date July 1, 2020

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Preventive Medical Services Subchapter 9. Heritable Diseases

Title 17
(Registar 86, No. 47-11-22-86)
6505 Collection of Specimen

(a) For each newborn born in a perinatal licensed facility, staff shall:

- Collect the screen after 12 hours of age and no later than 48 hours of age, prior to discharge or transfer;
- Any specimen collected prior to 12 hours of age is an early specimen and another specimen shall be collected after 12 hours of age;
- Collect prior to red blood cell transfusion when stable, even if under 12 hours of age;

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New 32 Million Series Test Request Form (TRF)

TO NAPS

Split specimen allows for rapid screening of all conditions

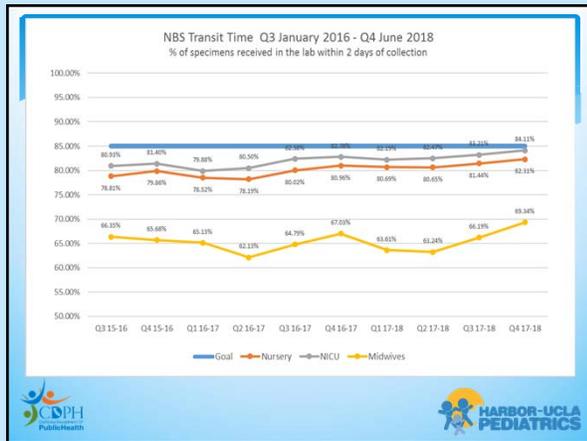
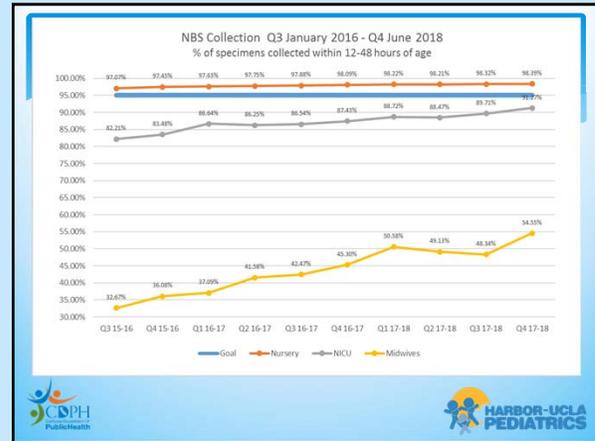
TO GDL

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California Timeliness Goals

- 95% of initial screens be collected between 12 and 48 hours of age.
- 85% of the initial specimens be received in the lab within 48 hours of collection.



Importance of Timeliness

The early identification of time critical disorders leads to

- Pre-Symptomatic Intervention
- Including Implementation of Therapy




Most Common Disorders on NBS

- Congenital Hypothyroidism occurs 1:1,900
- Sickle Cell Disease occurs 1:4,400 births

In contrast:

- PKU (All forms) 1: 16,813
- **SCID 1:65,000**




SCID

38 week 3230g male infant born in a hospital in Los Angeles.
NBS collected at 24 hours:

SCID Positive

Type	Result	Cutoff
TREC	1	> 18 copies/µL
ACTIN	1670	

• Also positive for HB C Beta+ thalassemia

145-54-150/21-2018-32




SCID

- Screen was received on Day 3
- Positive was called to the PCP at 8 days
- Referred to Specialist: Dr. Joseph A. Church of CHLA at 8 days
- Flow Cytometry collected at 14 days
- Admitted to CHLA at 16 days of age

145-54-150/21-2018-32



Joseph A. Church, MD

