A New Best Start:

The Licensed Midwife's crucial role in Newborn Screenings for Out-of-Hospital Births

Medical Board of California

Licensed Midwives Advisory Committee Meeting

12/09/2021

CDPH California Department of PublicHealth

Overview

- NBS Screening Introduction
- State Regulations and Policy
- NBS Screening process
- Summary

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What is Screening?

Screening, in general, is a public health measure designed to identify individuals in a population who may be at an increased risk of a certain disorder

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Goal of Newborn Screening

Newborn Screening (NBS) can identify babies with certain serious disorders so that treatment can be started right away. Early treatment can prevent delayed developmental growth, neurological or physical disabilities, and/or life-threatening illness.

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Brief History of NBS

- 1966 – California NBS program began with PKU
- Newborn Screening has expanded dramatically as testing methodologies improved and treatments have been developed
- The California NBS Program decides on our testing panel based on the federal Recommended Uniform Screening Panel (RUSP)

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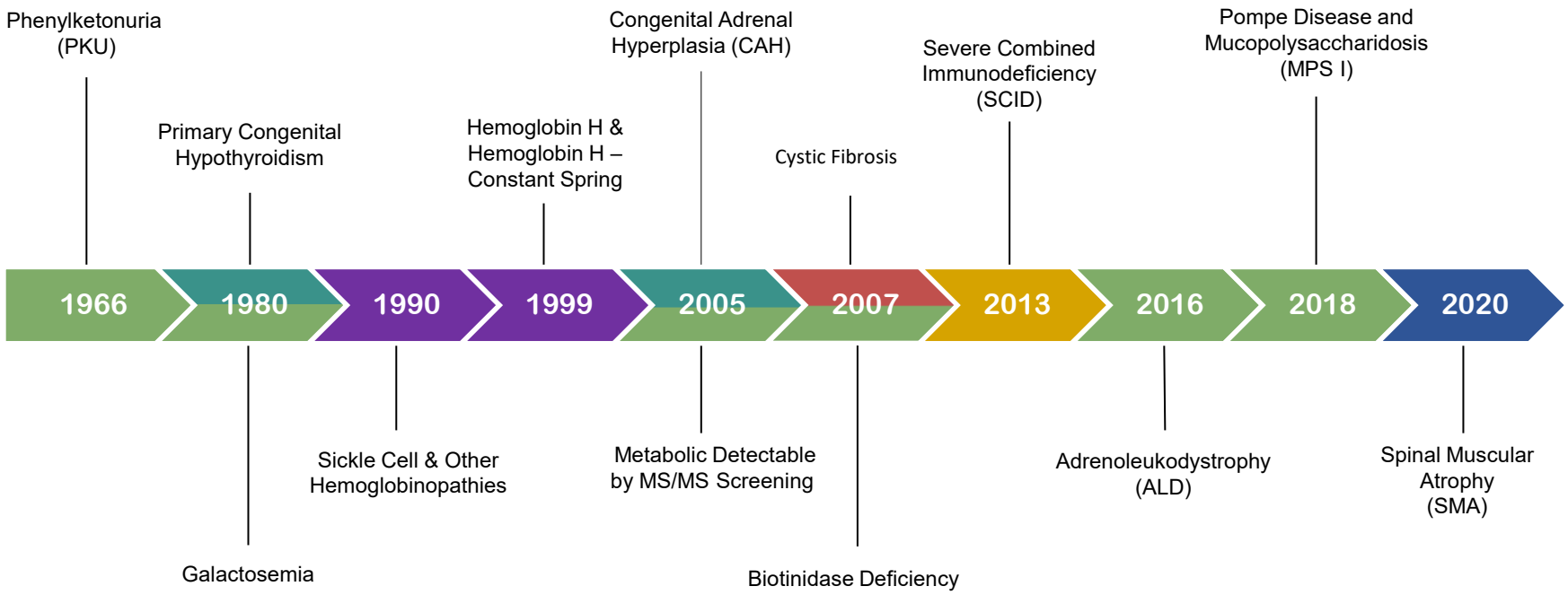
Conditions Screened

Advancements in the CA Newborn Screening Program (1966-2020)



Metabolic and Genetic Disorder
Hemoglobin Disorder
Severe Combined Immunodeficiency
Endocrine Disorder
Cystic Fibrosis
Neuromuscular Disorder

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Newborn Screening

- The California NBS program tests for a large panel of congenital and genetic disorders that includes endocrine, metabolic, neuromuscular, hemoglobin and immunologic ones
- The disorders are rare, but together nearly 1 in 500-600 infants are identified with one by NBS
- Treatments vary widely but include hormone replacement for hypothyroidism, dietary restriction for metabolic conditions and even gene therapy for SMA

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Newborn Screening is Law

Because NBS is important and so successful, it is mandated in the California Code of Regulations:

Title 17. Public Health Division 1. State Department of Health Services Chapter 4. Preventive Medical Service Subchapter 9. Testing for Heritable Disorders Group 3. Newborn Screening Program

- Families can only refuse on religious grounds
- Regulations cover the fee for the CA NBS panel (currently $176.25) and all program work is covered by these fees
- Sets responsibilities for stakeholders

California State Law & Regulations - Section §6504

Parent Education -

- All birth attendants … shall provide pregnant women, prior to the estimated date of delivery, with a copy of the informational material, entitled "*Important Information for Parents*," provided by the Department

CALIFORNIA NEWBORN SCREENING PROGRAM

Screening Your Newborn

Important Information for Parents
About a Newborn Screening Test

Congratulations!

The arrival of a new baby is an exciting time for a family. The California Newborn Screening Program tests every newborn for a group of rare disorders. This newborn screening can prevent serious health problems and save your baby's life.

California Department of Public Health
Newborn Screening Program
www.cdph.ca.gov/NBS

What Is Newborn Screening?
Newborn screening is a blood test that checks for certain rare disorders that can cause serious health problems, including brain damage or death, if not treated early. The test identifies most, but not all, of the babies who have these disorders.

Why Does My Baby Need This Test?
Babies with one of these disorders can look healthy at birth but still have a serious disease. Making a diagnosis early means treatment can be started before health problems occur.

How Is the Test Done?
A few drops of blood are taken from the baby's heel and put on special filter paper on the bloodspot collection card. The bloodspot card is then sent to a program approved laboratory for testing. This is a safe and simple test.

Who Gets the Test?
Hospitals and midwives must collect a newborn screen on every baby born in the state, by California law. You may refuse the test for religious reasons if you choose. The test is done 12–48 hours after the baby is born. Babies born in the hospital must have the test done before going home.

Which Disorders Are Tested for?
Over 80 disorders are screened for, including the following:
- Metabolic Disorders
- Endocrine Disorders
- Sickle Cell Disease/Hemoglobin Disorders
- Cystic Fibrosis
- Immunological Disorders
- Neurological Disorders

How Can I Get Results?
You will receive the pink and blue copy of a newborn screening form after the blood test. Please check that your contact information and health care provider are correct on the form.

Your baby's newborn screening results are sent to the doctor or clinic listed on the form. Save the pink copy of the form and bring it to your baby's first check-up.

The form number listed on the newborn screening form can be used to get the test result from the program. Your health care provider will tell you if more testing is needed.

What Happens to My Baby's Blood Spots After the Newborn Screening Test is Done?
California, like many other states, stores newborn screening bloodspot cards. The bloodspot cards may be used for tests to improve the screening program or to develop tests for new disorders.

The bloodspots may also be used for studies about diseases in women and children. The stored bloodspot cards do not have information, such as names or addresses, that can be used to identify you or your baby. The program follows all federal and state privacy and research laws.

If you want the bloodspot card destroyed after the newborn screening test is done, that is your right.

To learn more about the storage and use of leftover blood spots, or to find out how to get your baby's bloodspot card destroyed, visit: **https://www.cdph.ca.gov/Programs/CFH/DGDS/Pages/NBS/IIP.aspx.**

For more information about newborn screening, ask your health care provider or visit www.cdph.ca.gov/NBS.

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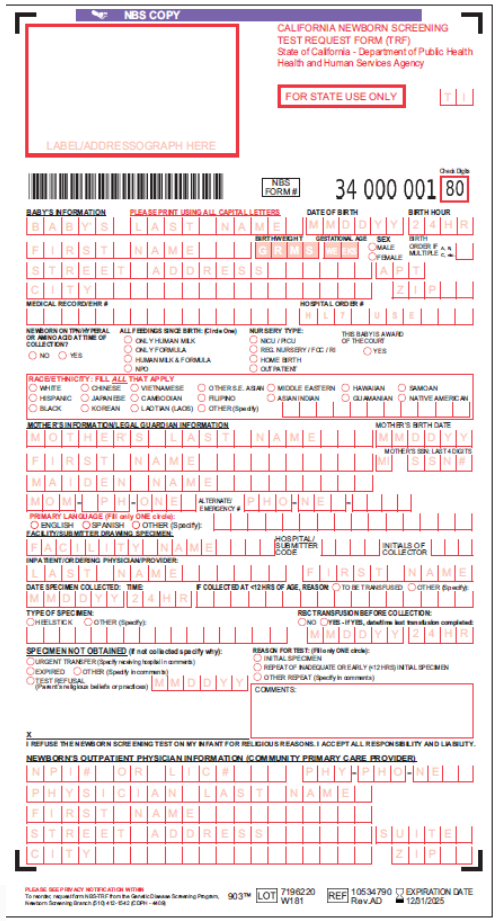
Section §6504 – For Out-of-Hospital Births

(1) Collect a newborn screening specimen between 12 and 48 hours of birth using the Instructions for Collecting Adequate Blood Specimens on the CALIFORNIA NEWBORN SCREENING TEST REQUEST FORM (NBS-TRF) (CDPH-4409) pursuant to §6501.5(a) and §6504.4(b) unless a religious refusal pursuant to §6501.2 is executed, or a newborn screening result is found in the infant's medical record.

(2) Ensure that specimens are given, on the same or next business day of the designated carrier, to a carrier contracted with the Department or contracted laboratory or to another same-day or overnight delivery service for transport

NBS Test Request Form and Collection Card


NBS COPY

CALIFORNIA NEWBORN SCREENING TEST REQUEST FORM (TRF)
State of California - Department of Public Health
Health and Human Services Agency

FOR STATE USE ONLY

LABEL/ADDRESSOGRAPH HERE

NBS FORM # 34 000 001 80

BABY'S INFORMATION — PLEASE PRINT USING ALL CAPITAL LETTERS

BABY'S LAST NAME | FIRST NAME | DATE OF BIRTH | BIRTH HOUR
BIRTHWEIGHT | GESTATIONAL AGE | SEX ○ MALE ○ FEMALE | BIRTH ORDER IF A, B, C, ... MULTIPLE
STREET ADDRESS | APT
CITY | ZIP
MEDICAL RECORD/EHR # | HOSPITAL ORDER #

NEWBORN ON TPN/IV/PERAL OR AMINO ACID AT TIME OF COLLECTION? ○ NO ○ YES
ALL FEEDINGS SINCE BIRTH: (Circle One) ○ ONLY HUMAN MILK ○ ONLY FORMULA ○ HUMAN MILK & FORMULA ○ NPO
NURSERY TYPE: ○ NICU / PICU ○ REG. NURSERY / FCC / RI ○ HOME BIRTH ○ OUT PATIENT
THIS BABY IS AWARD OF THE COURT ○ YES

RACE/ETHNICITY: FILL ALL THAT APPLY
○ WHITE ○ CHINESE ○ VIETNAMESE ○ OTHER S.E. ASIAN ○ MIDDLE EASTERN ○ HAWAIIAN ○ SAMOAN
○ HISPANIC ○ JAPANESE ○ CAMBODIAN ○ FILIPINO ○ ASIAN INDIAN ○ GUAMANIAN ○ NATIVE AMERICAN
○ BLACK ○ KOREAN ○ LAOTIAN (LAOS) ○ OTHER (Specify)

MOTHER'S (INFORMATION) LEGAL GUARDIAN INFORMATION
MOTHER'S LAST NAME | FIRST NAME | MOTHER'S BIRTH DATE
MAIDEN NAME | MOTHER'S SSN: LAST 4 DIGITS
MOM'S PHONE | ALTERNATE / EMERGENCY #
PRIMARY LANGUAGE (Fill only ONE circle) ○ ENGLISH ○ SPANISH ○ OTHER (Specify):

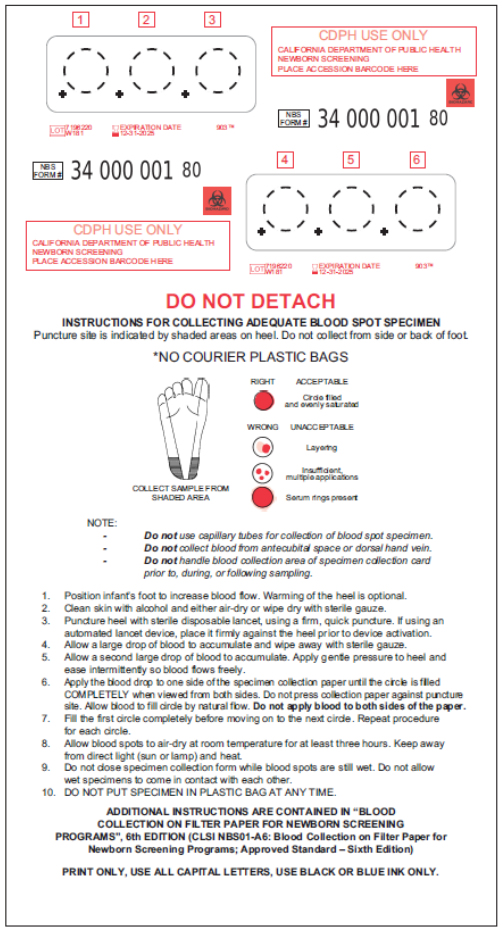
FACILITY/SUBMITTER DRAWING SPECIMEN:
FACILITY NAME | HOSPITAL/SUBMITTER CODE | INITIALS OF COLLECTOR
INPATIENT/ORDERING PHYSICIAN/PROVIDER:
LAST NAME | FIRST NAME
DATE SPECIMEN COLLECTED: | TIME: | IF COLLECTED AT <12 HRS OF AGE, REASON ○ TO BE TRANSFUSED ○ OTHER (Specify)
TYPE OF SPECIMEN: ○ HEELSTICK ○ OTHER (Specify):
RBC TRANSFUSION BEFORE COLLECTION: ○ NO ○ YES - If YES, date/time last transfusion completed:

SPECIMEN NOT OBTAINED (If not collected specify why):
○ URGENT TRANSFER (Specify receiving hospital in comments)
○ EXPIRED ○ OTHER (Specify in comments)
○ TEST REFUSAL (Parent's religious beliefs or practices)
REASON FOR TEST: (Fill only ONE circle):
○ INITIAL SPECIMEN
○ REPEAT OF INADEQUATE OR EARLY (<12 HRS) INITIAL SPECIMEN
○ OTHER REPEAT (Specify in comments)
COMMENTS:

X
I REFUSE THE NEWBORN SCREENING TEST ON MY INFANT FOR RELIGIOUS REASONS. I ACCEPT ALL RESPONSIBILITY AND LIABILITY.

NEWBORN'S OUTPATIENT PHYSICIAN INFORMATION (COMMUNITY PRIMARY CARE PROVIDER)
NPI # | OR LIC # | PHYSICIAN PHONE
PHYSICIAN LAST NAME | FIRST NAME
STREET ADDRESS | SUITE
CITY | ZIP

PLEASE SEE PRIVACY NOTIFICATION WITHIN
To reorder, request form NBS-TRF from the Genetic Disease Screening Program, Newborn Screening Branch (510) 450-7642 (CDPH - 4416)
903™ LOT 7196220 W181 REF 10534790 Rev.AD EXPIRATION DATE 12/31/2025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CDPH USE ONLY
CALIFORNIA DEPARTMENT OF PUBLIC HEALTH
NEWBORN SCREENING
PLACE ACCESSION BARCODE HERE

NBS FORM # 34 000 001 80

LOT 7196220 W181 EXPIRATION DATE 12-31-2025 903™

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CDPH USE ONLY
CALIFORNIA DEPARTMENT OF PUBLIC HEALTH
NEWBORN SCREENING
PLACE ACCESSION BARCODE HERE

LOT 7196220 W181 EXPIRATION DATE 12-31-2025 903™

DO NOT DETACH

INSTRUCTIONS FOR COLLECTING ADEQUATE BLOOD SPOT SPECIMEN
Puncture site is indicated by shaded areas on heel. Do not collect from side or back of foot.

*NO COURIER PLASTIC BAGS

COLLECT SAMPLE FROM SHADED AREA

RIGHT ACCEPTABLE
Circle filled and evenly saturated

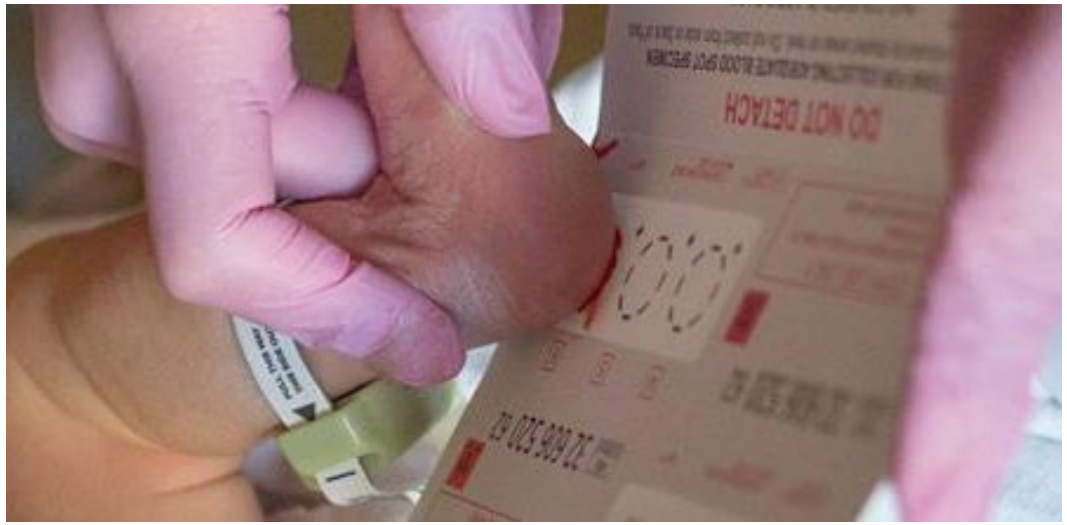
WRONG UNACCEPTABLE
Layering
Insufficient, multiple applications
Serum rings present

NOTE:
- ***Do not*** *use capillary tubes for collection of blood spot specimen.*
- ***Do not*** *collect blood from antecubital space or dorsal hand vein.*
- ***Do not*** *handle blood collection area of specimen collection card prior to, during, or following sampling.*

1. Position infant's foot to increase blood flow. Warming of the heel is optional.
2. Clean skin with alcohol and either air-dry or wipe dry with sterile gauze.
3. Puncture heel with sterile disposable lancet, using a firm, quick puncture. If using an automated lancet device, place it firmly against the heel prior to device activation.
4. Allow a large drop of blood to accumulate and wipe away with sterile gauze.
5. Allow a second large drop of blood to accumulate. Apply gentle pressure to heel and ease intermittently so blood flows freely.
6. Apply the blood drop to one side of the specimen collection paper until the circle is filled COMPLETELY when viewed from both sides. Do not press collection paper against puncture site. Allow blood to fill circle by natural flow. **Do not apply blood to both sides of the paper.**
7. Fill the first circle completely before moving on to the next circle. Repeat procedure for each circle.
8. Allow blood spots to air-dry at room temperature for at least three hours. Keep away from direct light (sun or lamp) and heat.
9. Do not close specimen collection form while blood spots are still wet. Do not allow wet specimens to come in contact with each other.
10. DO NOT PUT SPECIMEN IN PLASTIC BAG AT ANY TIME.

ADDITIONAL INSTRUCTIONS ARE CONTAINED IN "BLOOD COLLECTION ON FILTER PAPER FOR NEWBORN SCREENING PROGRAMS", 6th EDITION (CLSI NBS01-A6: Blood Collection on Filter Paper for Newborn Screening Programs; Approved Standard – Sixth Edition)

PRINT ONLY, USE ALL CAPITAL LETTERS, USE BLACK OR BLUE INK ONLY.



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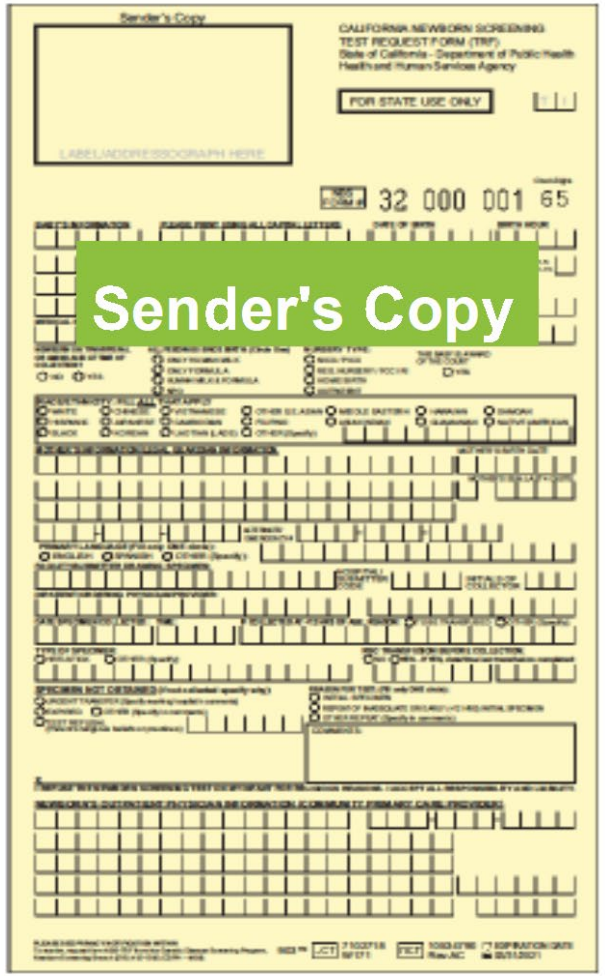
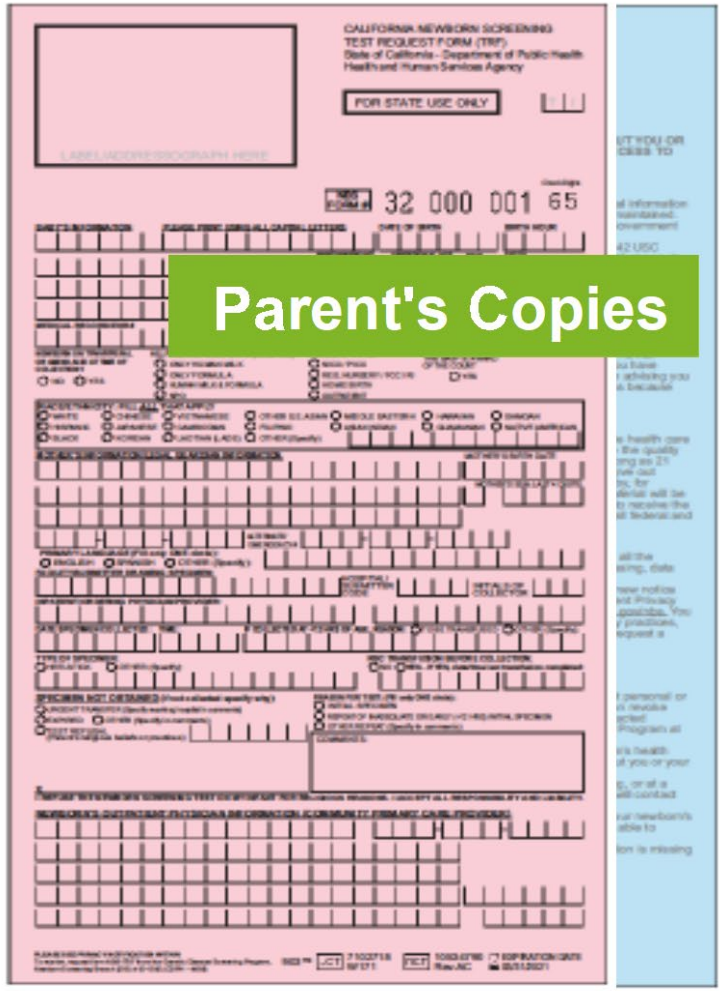
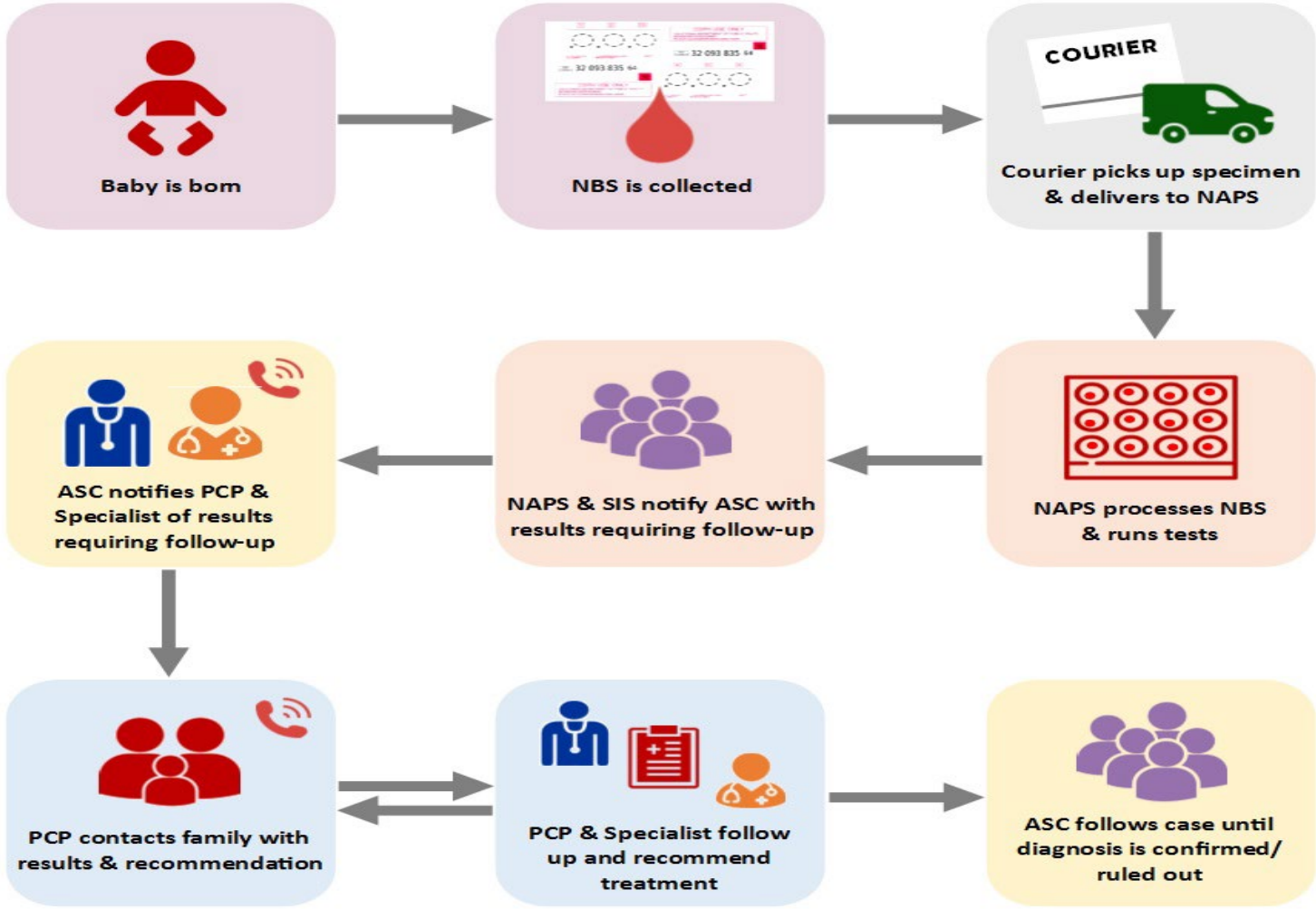
Test Request Form – In Detail





NBS Flow from Birth to Diagnosis



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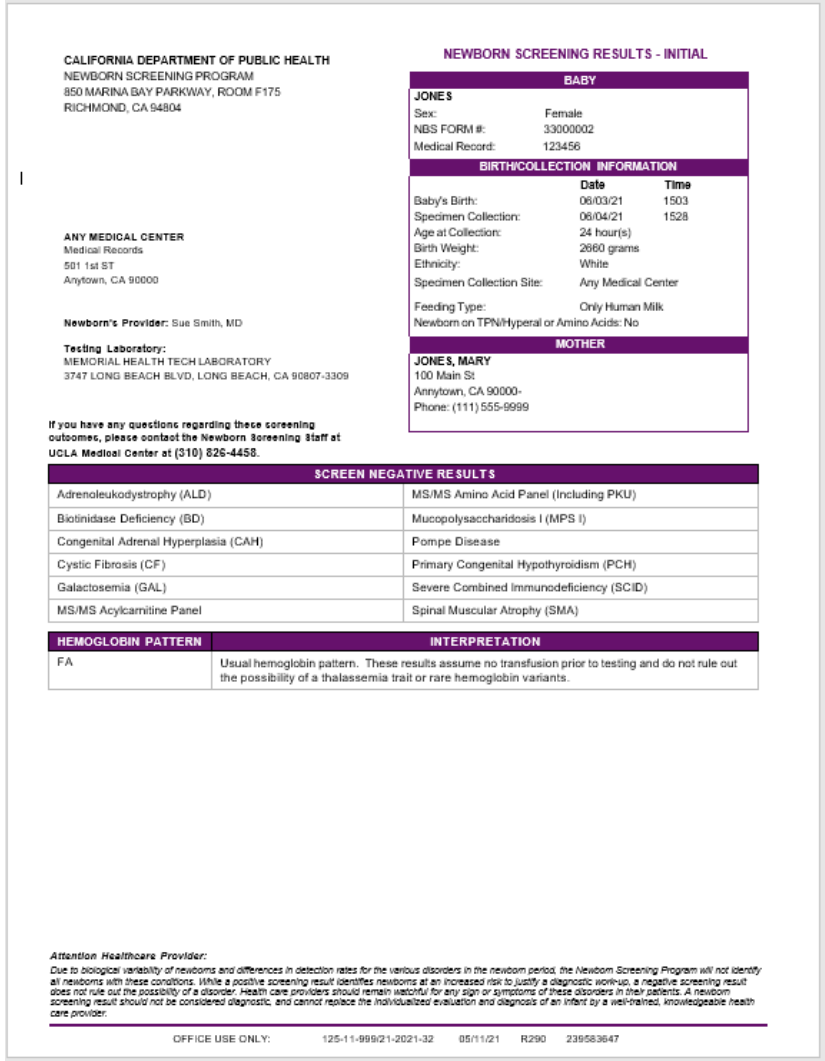
NBS Mailer



CALIFORNIA DEPARTMENT OF PUBLIC HEALTH
NEWBORN SCREENING PROGRAM
850 MARINA BAY PARKWAY, ROOM F175
RICHMOND, CA 94804

ANY MEDICAL CENTER
Medical Records
501 1st ST
Anytown, CA 90000

Newborn's Provider: Sue Smith, MD

Testing Laboratory:
MEMORIAL HEALTH TECH LABORATORY
3747 LONG BEACH BLVD, LONG BEACH, CA 90807-3309

If you have any questions regarding these screening outcomes, please contact the Newborn Screening Staff at UCLA Medical Center at (310) 826-4458.

NEWBORN SCREENING RESULTS - INITIAL

BABY	
JONES	
Sex:	Female
NBS FORM #:	33000002
Medical Record:	123456

BIRTH/COLLECTION INFORMATION	Date	Time
Baby's Birth:	06/03/21	1503
Specimen Collection:	06/04/21	1528
Age at Collection:	24 hour(s)	
Birth Weight:	2660 grams	
Ethnicity:	White	
Specimen Collection Site:	Any Medical Center	
Feeding Type:	Only Human Milk	
Newborn on TPN/Hyperal or Amino Acids:	No	

MOTHER
JONES, MARY
100 Main St
Annytown, CA 90000-
Phone: (111) 555-9999

SCREEN NEGATIVE RESULTS	
Adrenoleukodystrophy (ALD)	MS/MS Amino Acid Panel (Including PKU)
Biotinidase Deficiency (BD)	Mucopolysaccharidosis I (MPS I)
Congenital Adrenal Hyperplasia (CAH)	Pompe Disease
Cystic Fibrosis (CF)	Primary Congenital Hypothyroidism (PCH)
Galactosemia (GAL)	Severe Combined Immunodeficiency (SCID)
MS/MS Acylcarnitine Panel	Spinal Muscular Atrophy (SMA)

HEMOGLOBIN PATTERN	INTERPRETATION
FA	Usual hemoglobin pattern. These results assume no transfusion prior to testing and do not rule out the possibility of a thalassemia trait or rare hemoglobin variants.

Attention Healthcare Provider:
Due to biological variability of newborns and differences in detection rates for the various disorders in the newborn period, the Newborn Screening Program will not identify all newborns with these conditions. While a positive screening result identifies newborns at an increased risk to justify a diagnostic work-up, a negative screening result does not rule out the possibility of a disorder. Health care providers should remain watchful for any sign or symptoms of these disorders in their patients. A newborn screening result should not be considered diagnostic, and cannot replace the individualized evaluation and diagnosis of an infant by a well-trained, knowledgeable health care provider.

OFFICE USE ONLY: 125-11-999/21-2021-32 05/11/21 R290 239583647

Jones, Female — Date of Birth: 05/03/21 — Medical Record #: 123456 — 125-11-999/21-2021-32

Acylcarnitine	Expected Range	Value	Flag
PC	> 8 to < 125 µmol/L	15.6 µmol/L	
PC / (C16 + C18:1) Ratio	< 75	3.17	
C-2	> 11 to < 80 µmol/L	19.8 µmol/L	
C-3	< 6.3 µmol/L	2.0 µmol/L	
C03 / C02 Ratio	< 0.3	0.10	
C-3DC	< 0.4 µmol/L	0.060 µmol/L	
C03DC / C10 Ratio	< 5.2	1.50000	
C03DC / C03DC Ratio	> 0.5	1.33333	
C-4	< 1.7 µmol/L	0.160 µmol/L	
C-5	< 1 µmol/L	0.080 µmol/L	
C05 / CO3 Ratio	< 0.45	0.04000	
C-5:1	< 0.5 µmol/L	0.010 µmol/L	
C-5OH	< 0.85 µmol/L	0.190 µmol/L	
C-5DC	< 0.5 µmol/L	0.120 µmol/L	
C-6	< 0.95 µmol/L	0.040 µmol/L	
C-8	< 0.8 µmol/L	0.040 µmol/L	
C08 / C10 Ratio		0.67	
C-8:1	< 0.65 µmol/L	0.020 µmol/L	
C-10	< 0.65 µmol/L	0.060 µmol/L	
C-10:1	< 0.45 µmol/L	0.080 µmol/L	
C-12	< 2 µmol/L	0.130 µmol/L	
C-12:1		0.07 µmol/L	
C-14	< 1.2 µmol/L	0.270 µmol/L	
C14:1	< 0.8 µmol/L	0.120 µmol/L	
C14:1 / C12:1 Ratio		1.71	
C14:2		0.02 µmol/L	
C-14OH	< 0.2 µmol/L	0.020 µmol/L	
C-16	< 10 µmol/L	3.590 µmol/L	
C-16:1	< 1.4 µmol/L	0.190 µmol/L	
C-16OH	< 0.1 µmol/L	0.030 µmol/L	
C16OH / C16 Ratio	< 0.07	0.00836	
C-18	< 3.5 µmol/L	1.180 µmol/L	
C-18:1	< 7 µmol/L	1.330 µmol/L	
C-18:2		0.22 µmol/L	
C-18OH	< 0.1 µmol/L	0.010 µmol/L	
C-18:1OH	< 0.1 µmol/L	0.020 µmol/L	

Amino Acid	Expected Range	Value	Flag
Glycine		405 µmol/L	
Alanine	< 1000 µmol/L	265 µmol/L	
Valine		98.4 µmol/L	
Valine / Phenylalanine Ratio	< 3.5	1.43239	
Leucine/Isoleucine	< 250 µmol/L	81.3 µmol/L	
Leucine/Alanine Ratio	< 1.1	0.31	
Phenylalanine	< 165 µmol/L	67.3 µmol/L	
Phenylalanine/Tyrosine Ratio	< 2.4	0.82	
Tyrosine	< 850 µmol/L	107.9 µmol/L	
Succinylacetone	< 4.5 µmol/L	0.38 µmol/L	
Methionine	> 8 to < 100 µmol/L	19 µmol/L	
Citrulline	> 5 to < 60 µmol/L	23 µmol/L	
Citrulline/Arginine Ratio	< 6	2.32	
Ornithine	< 800 µmol/L	80 µmol/L	
Ornithine/Citrulline Ratio		3.42	
Arginine	< 50 µmol/L	10 µmol/L	
Arginine/Ornithine Ratio	< 1.4	0.13	
Proline	< 1500 µmol/L	137 µmol/L	
5-Oxoproline		24 µmol/L	

Disorder: Analyte	Expected Range	Value	Flag
CF: Immunoreactive Trypsinogen	< 69 ng/mL	24.14 ng/mL	
BD: Biotinidase	> 10 BRU	39.77 BRU	
GAL: Galactose 1 Unidyl transferase (GALT)	> 90 enz.units	320.60 enz.units	
PCH: Thyroid Stimulating Hormone (TSH)	< 29 mIU/L	4.06 mIU/L	
SCID: T-cell Receptor Excision Circle (TREC)	> 18 copies/µL	48 copies/µL	
CAH: 17-Hydroxyprogesterone (17-OHP)	< 85 nmol/L	0011.10 nmol/L	

Very Long Chain Fatty Acid Tier 1(ALD)*			
Disorder: Analyte	Expected Range	Value	Flag
ALD: C26	<0.42 µmol/L	0.280 µmol/L	

Lysosomal Storage Diseases*				
Disorder: Analyte	Expected Range	Value	Flag	Interpretation Comments
Pompe: Acid alpha-glucosidase (GAA)	>=1.917 µmol/L/h	10.58 µmol/L/h		The acid alpha-glucosidase (GAA) enzyme activity is above the cut-off of the daily patient median - suggestive of screen negative for Pompe disease.
MPS I: Alpha-L-iduronidase (IDUA)	>=1.1484 µmol/L/h	7.35 µmol/L/h		The alpha-L-iduronidase (IDUA) enzyme activity is above the cut-off of the daily patient median - suggestive of screen negative for Mucopolysaccharidosis I (MPS I) disorder.

Neuromuscular Disorders*				
Disorder: Analyte	Expected Range	Value	Flag	Interpretation Comments
SMA: SMN1	Exon 7 Present	Exon 7 Present		

***Genetic Disease Laboratory:** GDL 850 MARINA BAY PKWY, # G265, RICHMOND, CA 94804-6403
Director: Genetic Disease Laboratory, (510) 231-1790

Methods and Limitations:
Assays for ALD Tier-1, ALD Tier-2, CAH Tier-2, Pompe Tier-1, MPS I Tier-1 and SMA were developed and/or optimized by the California Department of Public Health Genetic Disease Laboratory Branch (GDLB). Performance characteristics of these assays are determined by GDLB. The SMA assay is designed to identify 95% of SMA patients who have homozygous deletion/mutation of SMN1 gene in 5q chromosome. These assays have not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. The assays are used for clinical purposes. They should not be regarded as investigational or for research. GDLB is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA-88) to perform high complexity genetic disease screening.

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Newborn Screening Program

Newborn Screening is more than a blood test. It is an interconnected process that involves many stakeholders and components such as education, testing, follow-up and evaluation of the program.

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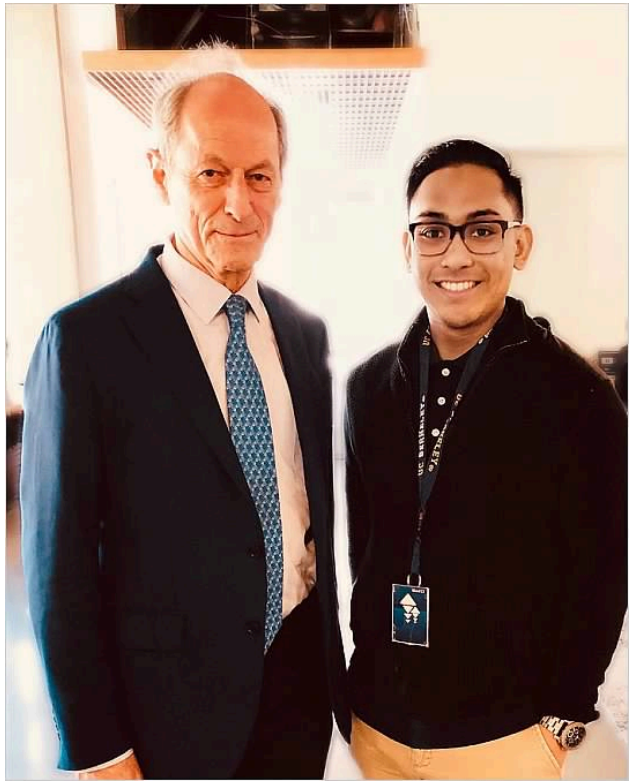
Critical Role in NBS for Licensed Midwives

- NBS is life-changing
- NBS is a multifaceted process with many stakeholders
- NBS advances a health equity framework
- NBS is mandatory

Summary



Rt Hon., Sir Michael Marmot, *2018*

“Give *every child* the best start in life.”

Thank you!

Visit our website:

cdph.ca.gov/NBS

Questions? Email us!

nbsoh@cdph.ca.gov

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