

**Spring
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Newborn Screening 411

RCHSD

Newborn Screening Area Service Center

George Knapp, RN, BSN, MS
Project Director
gknapp@rchsd.org

Barbara Galvin, RN, MSN
Nurse Coordinator
bgalvin@rchsd.org

Gail Maher, RN, BSN, PHN
Nurse Coordinator
gmaher@rchsd.org

Kimberly Kopita, MS, CGC
Genetic Counselor
kkopita@rchsd.org

Charlotte Pannell-Taylor, BA
Program Specialist
cpannelltaylor@rchsd.org

Michelle Gutierrez, BSW
Community Liaison
mjgutierrez@rchsd.org

Kristin Kersey
Administrative Assistant
kkersey@rchsd.org



Ensuring Testing of All Infants

The Newborn Screening (NBS) Program has several mechanisms in place to ensure testing of all babies born in California. State NBS Regulations specify reporting requirements for both licensed perinatal health facilities and county registrars to ensure testing. All newborns must be tested; **the only legal ground for refusal is a conflict with religious beliefs**. The following procedures and forms are utilized to ensure testing.

Perinatal facilities must review each newborn's medical record within 14 days from the date of discharge to determine that the NBS results are filed in it, or that a parent's or legal guardian's signed refusal is present. If it has been determined that a newborn was not tested, the facility must notify the infant's physician and the NBS Program. If a specimen was collected (as indicated by the presence of the yellow copy of the specimen collection form) but there is no NBS Results Mailer in the chart, the facility must complete a Missing Result Form (see below) and submit it to the State within five days.

Newborn Screening Missing Result Form (NBS-MR):

Used by licensed perinatal health facilities when a newborn screening specimen was collected but no results have been received.

Hospital Report of Newborn Screening Specimen Not Obtained (NBS-NO):

Used by licensed perinatal health facilities to report infants discharged without a newborn screening test, including those transferred to another hospital before a specimen is collected and those who have expired.

County birth registrars are required to inform persons registering the birth of a baby born outside of a licensed perinatal health facility of newborn screening. The registrars are also required to notify the NBS Program of these births via the NBS-OH form.

Notification of Registration of Birth Which Occurred Outside of a Licensed Health Facility (NBS-OH):

Used by county birth registrars to report babies born outside of a licensed health facility.

Important Information for Parents about the Newborn Screening Test (IIP):

Licensed perinatal health facilities are required to give this pamphlet to the parent before collection of the newborn screening specimen. Birth registrars are also required to give this pamphlet to the person registering the birth of a baby born outside of a licensed perinatal health facility.

It is essential that the NBS-NO and NBS-OH forms be mailed promptly to the State NBS Program. The state follows-up on each of these forms to make sure the baby has been tested. Unless there is a record of parent refusal on file, the State refers all untested babies to the Newborn Screening Area Service Centers for assistance in obtaining the test. If there is a delay in sending in the forms, we are delayed in getting the babies tested. This could delay treatment if a baby has one of the screened-for disorders, resulting in irreversible damage or even death.

RCHSD

Newborn Screening Area Service Center

LOCATED AT:

7920 Frost Street, Suite 302
San Diego, CA 92123

MAILING ADDRESS:

3020 Children's Way
Mail Code 5116
San Diego, CA 92123

PHONE:

(858) 966-8708

FAX:

(858) 966-8709

For more information visit:

www.dhs.ca.gov/NBS/



Always kiss your children goodnight - even if they're already asleep.

--H. Jackson Brown, Jr.

Post-Transfusion Follow-Up

If a newborn screening specimen is not collected before a packed red blood cell (RBC) transfusion, or if the pre-transfusion specimen is deemed inadequate and cannot be processed, the Newborn and Prenatal Screening (NAPS) lab will be unable to provide valid results for hemoglobinopathies and galactosemia. In post-RBC transfusion specimens, the donor's hemoglobin type can mask the newborn's. The donor blood can also supply the enzyme that the galactosemia screen measures; this could result in normal-appearing enzyme levels in a newborn that actually has low levels. If a baby does not have an adequate pre-transfusion specimen, providers should remain vigilant and evaluate the infant for any signs or symptoms of galactosemia (hypoglycemia, jaundice, vomiting, failure to thrive, lethargy, poor sucking, sepsis). Also, additional testing (on DNA from the white blood cells) may be done to evaluate for hemoglobin disorders. If you would like to request DNA testing for hemoglobinopathies, please contact your Area Service Center at 1-800-793-1313.

Question of the Quarter

Question: When a parent refuses to have the newborn screening test done, why must we complete both the test refusal (NBS-TR) and the not obtained (NBS-NO) forms?

Answer: The two forms serve different purposes. The test refusal form is the only document the parents sign; it describes their right to refuse testing (only if it conflicts with their religious beliefs and practices) and explains the possible consequences of declining screening. A test refusal signed by the parent releases the birth facility and the Newborn Screening Program from any responsibility for the fact that the screening was not done. The parents receive a copy of the test refusal form. The not obtained form is needed in order to alert the pediatrician that a newborn screen was not done. This allows the pediatrician to be particularly diligent in monitoring the baby for any signs or symptoms of a screened-for disorder. The pediatrician does not receive a copy of the test refusal form and the parent does not receive a copy of the not obtained form.

Diagnosed Cases in Riverside County (2009)

FACILITY	TSH	CAH	MS/MS	Hb	CF	GAL	BD
Corona Regional Medical Center	1		hyperphe				
Desert Regional Medical Center	1		2 SCADD	Alpha-thal major	suspect CF	DG	
Hemet Valley Medical Center							
Inland Valley Medical Center	1		MCADD				
JFK Memorial Hospital	1						
Moreno Valley Community Hospital					suspect CF		
Parkview Community Hosp Med Ctr		1	VLCADD				
Rancho Springs Medical Center					2 suspect CF		
Riverside Community Hospital	1			Hb S-Beta-thal	2		
Riverside County Regional Med Ctr				2 Hb SS			
San Gorgonio Memorial Hospital							
Totals	5	1	5	4	6	1	0
ASC totals (Riverside, San Diego, Orange, and Imperial Counties)	45	3	22	22	13	4	1

Key to Diagnoses:

TSH – Primary Congenital Hypothyroidism
CAH – Congenital Adrenal Hyperplasia
CF – Cystic Fibrosis
BD – Biodinidase Deficiency

GAL – Galactosemia (GG) & Duarte-Galactosemia (DG)
Hb – Hemoglobinopathies
MS/MS – Amino Acidopathies & Acylcarnitine Disorders