

CALIFORNIA DEPARTMENT OF PUBLIC HEALTH
 NEWBORN SCREENING PROGRAM
 850 MARINA BAY PARKWAY, ROOM F175
 RICHMOND, CA 94804
 (510) 412-1502

Severe Combined Immune Deficiency (SCID)

Supplemental NEWBORN SCREENING RESULTS

INITIAL Specimen

INITIAL Specimen Accession Number:

HOSPITAL
 TORRANCE, CA 90505

Newborn's Physician:

Telephone #: Ext:

Testing Laboratory:

PerkinElmer Genetics
 850 Marina Bay Parkway
 Richmond, CA 94804

Director:

John E. Sherwin, PhD
 Director of Laboratory Operations
 NTD Labs and PKI Genetics
 631.768.3011; 412.220.2300 x 113
 Email John.Sherwin@PerkinElmer.com

BABY		
F		
Gender:		M
Medical Record #:		
BIRTH/COLLECTION INFORMATION		
	Date	Time
Baby's Birth:		1327
Specimen Collection:		
Age at Collection (in hours):	6	
Birth Weight:		
Specimen Collection Site:	HOSPITAL	
Feeding Type:		
Newborn on TPN/Hyperal or Amino Acids:	No	
Transfusion:	No	
MOTHER		

Test interpretations are based on the Birth/Collection Information provided above and subject to disclaimer below.

TEST	Analyte:	Value	Reference Range
Severe Combined Immune Deficiency (SCID)	TREC	9	TREC#: Greater than or equal to 25
	Actin	1530	ACTIN#: Greater than or equal to 10,000

Interpretation: DNA AMPLIFICATION FAILURE FOR SCID TEST. The screen for SCID was incomplete.

Action Required: A repeat dried blood spot specimen is requested. The repeat blood spot specimen should be sent directly to the Genetic Disease Laboratory in Richmond.

DNA analysis was performed for the determination of copy number of one of the T-cell Receptor Excision Circles (TRECs) by real time quantitative PCR. TRECs are generated during the differentiation of T cells, and are used as surrogate markers for the number of naïve T cells. FOR THIS SAMPLE, THE COPY NUMBERS OF THE TREC WERE DETERMINED TO BE BELOW THE NORMAL LIMIT BUT THERE WAS A FAILURE TO AMPLIFY THE DNA AS INDICATED BY THE ACTIN NUMBER BEING LESS THAN 10,000. Result should be interpreted in the context of clinical presentation.

***DNA testing was developed and its performance characteristics determined by PerkinElmer Genetics, Inc. It has 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. Genetic testing is performed by PCR and allele specific hybridization. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA-88) as qualified to perform high complexity testing.*

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 the newborns at an increased risk to justify a diagnostic work-up, a negative screening 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.

If you have questions regarding these results, please contact Genetic Disease Screening Program Staff Doris Bryant at (510) 412-1453.