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:
365-17-160/21-2010-11

Newborn's Physician:
Telephone #: (650) 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		
Gender:	Female	
Medical Record #:		
BIRTH/COLLECTION INFORMATION		
	Date	**Time**
Baby's Birth:		
Specimen Collection:		
Age at Collection (in hours):	34	
Birth Weight:	3288	
Specimen Collection Site:		
Feeding Type: Human Milk & Formula		
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	20	TREC#: Greater than 25
	Actin	15400	ACTIN#: Greater than or equal to 10,000

Interpretation: **POSITIVE SCID TEST RESULT**

Action Required: **A liquid blood specimen for CBC differential and flow cytometry sent to Quest Diagnostics is required for confirmation of the screening result.**

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 SIGNIFICANTLY BELOW NORMAL LIMIT. ALTHOUGH THE ASSAY IS INTENDED FOR THE IDENTIFICATION OF SCID, IT CAN ALSO DETECT OTHER FORMS OF PROFOUND T CELL LYMPHOPENIA. 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 no 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 replae 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.