ClinGenLab
Clinical Testing Laboratory, Inc.

Clinical Testing Lab of Washington
2150 Pennsylvania Avenue NW
Washington, DC 20037
Phone: 202-555-1212

Specimen Number	Specimen Type	Control Number	Account Number	Account Phone Number	Route
	Peripheral Blood				

Patient Last Name	Patient Barcode

Patient First Name	Patient Middle Name
David	

Patient SS#	Patient Phone	Total Volume

Age (Y/M/D)	Date of Birth	Sex	Fasting
6 y.o.		Male	

Patient Address	Indication: Severe Combined Immunodeficiency Family History: No family history Ethnicity: Western European Caucasian

Date and Time Collected	Date Entered	Date and Time Reported	Physician Name	NPI	Physician ID
			Jane Ferreiro, MD		

Tests Ordered
Severe Combined Immunodeficiency (SCID): Three-gene Profile (IL2RG, ADA, IL7R)

General Comments
Please send a copy of the final report to the Molecular Science/M1 Training office via Fax at (202) 555-1212

Clinical test results for Severe Combined Immunodeficiency (SCID)

GENE	TEST RESULTS	EXPLANATION
IL2RG (Xq13.1)	Cys115Arg	**This result confirms the diagnosis of X-linked Severe Combined Immunodeficiency (SCID)**. This result should be interpreted in the context of clinical presentation and results of other laboratory tests. A PCR/sequencing study has confirmed one copy of the Cys115Arg (IL2RG: g.5939T>C, c.343T>C or p.Cys115Arg) variation. The Cys115Arg mutation is caused by a T to C change at nucleotide position 5939 in the IL2RG gene. This encodes an amino acid at position 115 (arginine) that is different from the reference (cysteine) and may have implications on structure and or function of the resulting protein. This individual's result has important implications for other family members. Clinical and laboratory evaluations should be considered for at risk individuals. Genetic counseling is recommended for at risk individuals.
ADA (20q13.12) IL7R (5p13.2)	Negative	No known pathogenic variant detected in these genes

INDICATIONS FOR TESTING
Individuals with a diagnosis of Severe Combined Immunodeficiency (SCID) with genetic counseling, are candidates for testing.

METHODOLOGY
Gene sequencing: All coding exons and associated intron junctions are analyzed by direct DNA sequence analysis using an automated fluorescent sequencing machine. When a mutation is detected, confirmation is carried out on an independent amplification of PCR using a second prep (B-prep) by sequencing in the opposite direction. If no mutation is found, sequence analysis is performed in both directions.

PERFORMANCE
Gene sequencing: From previous experience, we have been able to detect ADA, IL2RG, or IL7R mutations in about 99% of individuals with the diagnosis of Severe Combined Immunodeficiency (SCID) with specificity of mutation detection in probands detection is also estimated to be greater than 99%.

LIMITATIONS
The sequence analysis will not detect mutations located in regions of ADA, IL2RG, or IL7R that are not analyzed (non-coding exon regions, intron regions other than the splice junctions, and upstream and downstream regions). The sequencing method also will not detect gross genetic alterations including most duplications, inversions, or deletions (in females). Some sequence alterations that may be detected (such as those causing missense or synonymous changes) will be of unknown clinical significance.

CLINICAL DESCRIPTION
Group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. It is inherited as an X-linked or autosomal recessive defect. Mutations occurring in many different genes cause human Severe Combined Immunodeficiency (SCID).

-from MeSH