



Division of Human Genetics

X-linked Severe Combined Immunodeficiency (*IL2RG*)

Molecular Genetics Laboratory

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Additional information and test requisitions are available at:

www.cincinnatichildrens.org/molecular-genetics



Helping you fit the pieces together

Shipping Instructions

Please enclose test requisition with sample. All information must be completed before sample can be processed.

Place samples in Styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday

Ship to:

Cytogenetics and Molecular Genetics Laboratories

3333 Burnet Avenue NRB 1042

Cincinnati, OH 45229

513-636-4474

X-linked Severe Combined Immunodeficiency (X-SCID), typically presents within a few months after birth with persistent viral, bacterial, and fungal infections, lymphocytopenia, growth failure, and thymic hypoplasia. Patients have a low number of T cells, variable number of B cells, low immunoglobulins, and no specific antibodies. X-SCID is caused by a mutation in the common gamma chain gene. This gene, located on the X chromosome at Xq13.1, is composed of eight exons and seven introns and spans approximately 4.2 kilobases. Pathologic mutations have been described in all exons.

INDICATIONS

- Confirmation of diagnosis in an at-risk or symptomatic individual.
- Carrier identification in females with a family history.
- Prenatal diagnosis of an at-risk fetus, after identification of a mutation in a proband (by previous arrangement only).

METHODOLOGY

PCR-based sequencing of entire coding region of the interleukin 2 receptor gamma chain (*IL2RG*) gene and intron/exon boundaries.

IL2RG sequencing is also available as part of our **Severe Combined Immunodeficiencies Panel** which detects mutations in *IL2RG*, as well as in other genes which cause Severe combined immunodeficiency and other T-cell disorder. This SCID panel is expected to identify > 85% of the genetic causes of SCID. Please see our website for details.

SPECIMEN:

Sequencing detects about 99% of mutations in the *IL2RG* gene in males. In females, test sensitivity is somewhat lower as PCR-based sequencing does not detect large deletions, insertions or rearrangements within a gene.

SENSITIVITY:

At least 3mLs whole blood in lavender top (EDTA) tube. Label tube with patient's name, birth date, and date of collection. Cytobrushes are required for analysis in patients who have undergone transplantation and may facilitate DNA isolation in patients undergoing chemotherapy or in individuals with leukopenia. Please call for a free cytobrush collection kit.

TURN-AROUND TIME:

30 days

COST:

Please call 1-866-450-4198 for institutional pricing or with any billing questions

CPT CODES:

| | |
|------------------------------------|-------|
| Full gene mutation analysis | 81405 |
| Family specific mutation detection | 81403 |

RESULTS:

Results will be reported to the referring physician or health care provider as specified on the requisition form.