Cincinnati Children's

Division of Human Genetics

X-linked hyper IgM immunodeficiency syndrome (*CD40LG*)

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 hyper IgM syndrome typically presents as recurrent bacterial infections, such as otitis media, sinusitis, and pneumonias by age one year. Males with this condition often develop autoimmune hematologic disorders including neutropenia, thrombocytopenia, and hemolytic anemia. Other medical complications may include lymphomas and other malignancies, serious gastrointestinal complications, and neurologic deterioration. Elevated IgM, in the absence of other immunoglobulins is suggestive of this condition. X-linked hyper IgM syndrome is caused by a mutation in the *CD40LG* gene. The *CD40LG* maps to Xq26 and contains five exons and four introns. Single base mutations, small deletions, and insertions 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 and intron/exon boundaries of the *CD40LG* .

SPECIMEN:

At least 3mLs whole blood in lavendar top (EDTA) tube. Label tube with patient's name, birth date, and date of collection. Buccal swabs or 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.

SENSITIVITY:

Sequencing detects about 98% of mutations in *CD40LG* in males.
In females, test sensitivity is somewhat lower as PCR-based sequencing does not detect large deletions, insertions or rearrangements within a gene.

TURN-AROUND TIME:

30 days

COST:

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

CPT CODES:

Full gene mutation analysis	81404
Family specific mutation detection	81403

RESULTS:

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

Updated 10/15