Cincinnati Children's

Division of Human Genetics

X-linked immune dysregulation, polyendocrinopathy, enteropathy syndrome (*FOXP3*)

Molecular Genetics Laboratory
CLIA#: 36D0656333
Phone: (513) 636-4474
Fax: (513) 636-4373
Email: moleculargenetics@cchmc.org

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

IPEX typically presents during infancy with systemic autoimmunity often presenting as the classical triad of endocrinopathy (type I diabetes or thyroid disease), severe watery diarrhea and dermatitis.
Older individuals typically develop failure-to-thrive, life-threatening infections and additional autoimmune disorders including various cytopenias, tubular nephrology and alopecia. IPEX is an X-linked recessive disorder and is caused by mutations in the FOXP3 gene, located at Xp11.23. *FOXP3* is composed of 11 exons. Pathologic mutations have been described in most of the 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 the entire coding region and intron/exon boundaries of the *FOXP3* gene.

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 99% of mutations in the coding regions and intron/exon boundaries of the *FOXP3* gene in males. Approximately 60% of males with IPEX have an identifiable mutation in *FOXP3*.

TURN-AROUND TIME:

30 days

COST:

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

CPT CODES:

Full gene sequence analysis	81479
Family specific mutation analysis	81403

RESULTS:

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

Updated 10/2015