Cincinnati Children's

Division of Human Genetics

Wiskott-Aldrich syndrome (WAS)

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

Wiskott-Aldrich syndrome (WAS) typically presents during infancy with thrombocytopenia, eczema, and recurrent infections. Bleeding episodes tend to decrease with age, but older patients have increasing problems with infections, and may develop autoimmune diseases and malignancies. Diagnostically, patients have thrombocytopenia with decreased platelet size, variable immunodeficiency, absent isohemagglutinins, and poor antibody response to polysaccharide antigens.

WAS is caused by a mutation in the WAS gene, located on the X chromosome at Xp11.22-23. The WAS gene is composed of 12 exons. Pathologic mutations have been described in all 12 exons. X-linked thrombocytopenia and X-linked severe congenital neutropenia are also caused by mutations in the WAS gene.

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).

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 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.

METHODOLOGY:

PCR-based sequencing of the entire coding region and intron/exon boundaries of the WAS gene.

SENSITIVITY & SPECIFICITY:

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

TURN-AROUND TIME:

30 days

COST:

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

CPT CODES:

Full gene mutation analysis	81406
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 11/13