

Using Genetic Testing to Diagnose Wiskott-Aldrich Syndrome (WAS)

Indications:

- Thrombocytopenia with small platelets, eczema, and recurrent, severe infections by both pyogenic bacteria and opportunistic pathogens
- Male-limited family history of bleeding disorders and recurrent, severe infections, or family history of Wiskott-Aldrich Syndrome

Benefits:

- By identifying the exact molecular cause, genetic testing can confirm or establish a diagnosis of Wiskott-Aldrich Syndrome and may clarify the prognosis.¹
- Carrier testing can allow early diagnosis in descendants. Early diagnosis is important, since prognosis is improved if treatment is initiated before onset of serious infections or bleeding disorders.²

Background:

- Wiskott-Aldrich Syndrome (WAS) is characterized by thrombocytopenia with very small platelets, eczema, and immune deficiency resulting in recurrent infections, autoimmunity, and malignancies.²⁻⁵
- To date, X-linked recessive loss-of-function mutations in *WAS*, the gene encoding the Wiskott-Aldrich Syndrome protein, are the only documented cause of WAS.⁶
- Mutations in *WAS* can lead to a range of clinical phenotypes, from classic, severe WAS, to X-linked thrombocytopenia (XLT), which is a milder disease phenotype characterized by persistent or intermittent thrombocytopenia with small platelets and minimal or no immunodeficiency, eczema, or malignancy.¹
- Hematopoietic stem cell transplantation (HSCT) offers the prospect of a cure for Wiskott-Aldrich Syndrome. When HSCT is not an option, intravenous immunoglobulin therapy (IVIG) coupled with prophylactic antibiotics are prescribed to control infections, while local or systemic steroids are recommended to treat eczema. Splenectomy may be necessary to reverse severe thrombocytopenia.^{1,7}

References: 1. Ochs HD and Thrasher AJ (2006) *J Allergy Clin Immunol* 117:725-738. 2. Filipovich AH et al (2001) *Blood* 97:1598-1603. 3. Sullivan KE et al (1994) *J Pediatr* 125:876-885. 4. Dupuis-Girod S et al (2003) *Pediatrics* 111:e622-e627. 5. Imai K et al (2004) *Blood* 103:456-464. 6. Derry JM et al (1994) *Cell* 78:635-644. 7. Orange J et al (2004) *Cell Mol Life Sci* 61:2361-85.

Ordering Information: please see other side



Ordering Information for Wiskott-Aldrich Syndrome Testing

Indications for Testing

- Thrombocytopenia with small platelets, eczema, and recurrent, severe infections by both pyogenic bacteria and opportunistic pathogens
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Ordering Information

Gene	Test Code
WAS	100501

Family Testing (single amplicon)

Family Testing is available. Please contact Client Services at 1-866-647-0735 for requirements.

Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

For test information, sample requirements, or to request a sample shipping kit, please contact Client Services at 1-866-647-0735 or visit us on the web at www.correlagen.com.