

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
- Male-limited family history of bleeding disorders and recurrent, severe infections, or family history of Wiskott-Aldrich Syndrome

Ordering Information

Gene	CPT Codes	Test Code
WAS	83891(1) 83892(1) 83898(12) 83904(24) 83909(24) 83912(1)	100501

Family Testing (single amplicon)

WAS	83891(1) 83892(1) 83898(1) 83904(2) 83909(2) 83912(1)	100501
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Test Methodology

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

Sample Requirements

- For blood samples:
 - 2 mL whole blood in EDTA tube (lavender top)
 - Samples can be stored briefly at 4°C, but should be shipped on day of collection .
- For buccal swab samples:
 - Please contact client services at 1-866-647-0735 for instructions.
- All sample types should be shipped overnight at room temperature.
- To request a sample shipping kit, please call 1-866-647-0735.

Turn-around Times

Turn-around times typically range from 7 to 21 days of receipt of sample and all required forms, but may vary depending on test volume and test-specific technical difficulties. Current TATs are posted on our website. Please schedule patient follow-up appointments for discussion of test results conservatively at 6 weeks.

For more information, please contact Correlagen Diagnostics, Inc., at 1-866-647-0735 or visit us on the web at www.correlagen.com.