

## Using Genetic Testing to Diagnose X-Linked Lymphoproliferative Disease (XLP)

### Indications:

- Presence of at least one of the three primary phenotypes of XLP, including:
  - Fulminant infectious mononucleosis (FIM) following EBV infection
  - Lymphoproliferative disorders, usually B-cell non-Hodgkin lymphomas
  - Failure to generate specific antibodies in response to vaccination and/or recurrent, severe infections due to defects in T-cell, NK-cell, and B-cell function (dysgammaglobulinemia)
- Male-limited family history of any of the primary phenotypes of XLP, or a family history of *SH2D1A*-related XLP

### Benefits:

- Identification of a disease-associated mutation in *SH2D1A* can establish a firm diagnosis of XLP, which can be difficult to distinguish from other, clinically similar disorders, such as common variable immunodeficiency (CVID) and hemophagocytic syndromes.<sup>1</sup>
- Early diagnosis of XLP can allow timely initiation of potentially life-saving treatment.

### Background:

- XLP, also known as Duncan's Disease, is a rare primary immunodeficiency, originally characterized by extreme susceptibility to EBV infection.<sup>2</sup>
- XLP is now recognized as a heterogeneous disorder, with three primary clinical phenotypes, including FIM, dysgammaglobulinemia, and lymphoproliferative disorders. These phenotypes can appear either singly or sequentially and usually, but not always, follow EBV infection.<sup>1,3,4</sup>
- XLP is a potentially life-threatening disorder, with 70% of patients dying by the age of 10 and very few patients living beyond the age of 40.<sup>3</sup> However, hematopoietic stem cell transplantation offers the prospect of a cure.<sup>5</sup>

**References:** 1. Nichols KE et al (2005) *Immunol Rev* 203:180-199. 2. Purtilo DT (1975) *Lancet* 1:935-940. 3. Seemayer TA et al (1995) *Pediatr Res* 38:471-478. 4. Sumegi J et al (2002) *Leuk Lymphoma* 43:1189-1201. 5. Gross GT et al (1996) *Bone Marrow Transplant* 17:741-744.

**Ordering Information:** please see other side

## Ordering Information for X-Linked Lymphoproliferative Disease (XLP) Testing

### Indications for Testing

- Presence of at least one of the three primary phenotypes of XLP, including
  - Fulminant infectious mononucleosis (FIM) following EBV infection
  - Lymphoproliferative disorders, usually B-cell non-Hodgkin lymphomas
  - Failure to generate specific antibodies in response to vaccination and/or recurrent, severe infections due to defects in T-cell, NK-cell, and B-cell function (dysgammaglobulinemia)
- Male-limited family history of any of the primary phenotypes of XLP, or a family history of *SH2D1A*-related XLP

### Ordering Information

Gene	CPT Codes	Test Code
<i>SH2D1A</i>	83891(1) 83892(1) 83898(4) 83904(8) 83909(8) 83912(1)	101001

### Family Testing (single amplicon)

<i>SH2D1A</i>	83891(1) 83892(1) 83898(1) 83904(2) 83909(2) 83912(1)	101001
---------------	---	--------

### 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](http://www.correlagen.com).**