

## 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	Test Code
<i>SH2D1A</i>	101001

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