

### Diagnostic Criteria

Which of the following clinical phenotypes led to initial testing and diagnosis of XIAP deficiency?

- HLH
- Inflammatory Bowel disease
- Recurrent arthritis
- Recurrent cytopenias
- Recurrent fevers
- Recurrent Infections
- Splenomegaly
- Hepatitis and/or liver failure
- Other \_\_\_\_\_

### Family History

	History of Disease	Family Member(s)	Other Member
Hemophagocytic Disorders	<input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Unknown	<input type="checkbox"/> Uncle(s) <input type="checkbox"/> Cousin(s) <input type="checkbox"/> Sibling(s) <input type="checkbox"/> Other	_____
X-Linked HLH	<input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Unknown	<input type="checkbox"/> Uncle(s) <input type="checkbox"/> Cousin(s) <input type="checkbox"/> Sibling(s) <input type="checkbox"/> Other	_____
X-Linked Lymphoproliferative Disease	<input type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Unknown	<input type="checkbox"/> Uncle(s) <input type="checkbox"/> Cousin(s) <input type="checkbox"/> Sibling(s) <input type="checkbox"/> Other	_____

### HLH-Associated Features

Approximate # of HLH episodes \_\_\_\_\_

Sites of hemophagocytosis at diagnosis

- Bone Marrow
- CSF
- Liver
- Lymph Nodes
- Spleen
- Other \_\_\_\_\_

Patient experienced neurological abnormalities

- Yes
- No
- Unknown

On cyclosporine at the time?

- Yes
- No
- Unknown

### HLH-Associated Lab Data

Serum Ferritin	_____	mg/dL
Triglycerides	_____	mg/dL
Fibrinogen	_____	mg/dL

### HLH-Associated Testing

	Result
Soluble Interleukin-2 receptor alpha chain (sCD25)	<input type="radio"/> Normal <input type="radio"/> Elevated <input type="radio"/> Unknown or Not Done
IL-18	<input type="radio"/> Normal <input type="radio"/> Elevated <input type="radio"/> Unknown or Not Done
Soluble CD163	<input type="radio"/> Normal <input type="radio"/> Elevated <input type="radio"/> Unknown or Not Done
Cerebrospinal Neopterin	<input type="radio"/> Normal <input type="radio"/> Elevated <input type="radio"/> Unknown or Not Done
Cerebrospinal Protein	<input type="radio"/> Normal <input type="radio"/> Elevated <input type="radio"/> Unknown or Not Done
Cerebrospinal WBC Count	<input type="radio"/> Normal <input type="radio"/> Elevated <input type="radio"/> Unknown or Not Done