

**DISEASE:**  
**Familial hemophagocytic lymphohistiocytosis**

<b>NAME:</b>	Familial hemophagocytic lymphohistiocytosis
<b>DESCRIPTION:</b>	Familial Hemophagocytic lymphohistiocytosis (FHL) is a rare primary immunodeficiency characterized by a macrophage activation syndrome (see this term) with an onset usually occurring within a few months or less common several years after birth.
<b>ORPHACODE:</b>	540
<b>SYNOMYS:</b>	Familial HLH
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">OMIM</a> <a href="#">OMIM</a> <a href="#">OMIM</a> <a href="#">OMIM</a> <a href="#">OMIM</a> <a href="#">MedDRA</a> <a href="#">ICD-10</a> <a href="#">OMIM</a>

<b>ANALYTE(S):</b>	<u>PRF1</u> <u>STX11</u> <u>UNC13D</u> <u>STXBP2</u>
<b>CREATED:</b>	13 May 2019 - 01:02
<b>CHANGED:</b>	22 Jun 2023 - 16:14

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## RELATED CONTENT

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### Related Genetic Tests

- Primary immune deficiencies (gene panel)
- Trombosis - Hemostasis (gene panel)

### Related Laboratories

- Centrum Menselijke Erfelijheid - KUL

### Related Analytes

- perforin 1
- syntaxin 11
- syntaxin binding protein 2
- unc-13 homolog D

### Related Gene Panels

- Primary immune deficiencies (444 genes) - KUL
- Trombosis - Hemostasis (107 genes) - KUL

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