

**DISEASE:**  
**Bernard-Soulier syndrome**

<b>NAME:</b>	Bernard-Soulier syndrome
<b>DESCRIPTION:</b>	A rare, inherited platelet disorder characterized by mild to severe bleeding tendency , macrothrombocytopenia and absent ristocetin-induced platelet agglutination.
<b>ORPHACODE:</b>	274
<b>SYNONYMS:</b>	Giant platelet syndrome Hemorrhagiparous thrombocytic dystrophy
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">OMIM</a> <a href="#">OMIM</a> <a href="#">MeSH</a> <a href="#">MedDRA</a> <a href="#">ICD-10</a>
<b>ANALYTE(S):</b>	<a href="#">GP1BA</a> <a href="#">GP1BB</a> <a href="#">GP9</a>
<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

- Trombosis - Hemostasis (gene panel)

### Related Laboratories

- Centrum Menselijke Erfelijheid - KUL

### Related Analytes

- glycoprotein Ib platelet subunit alpha
- glycoprotein Ib platelet subunit beta
- glycoprotein IX platelet

### Related Gene Panels

- Trombosis - Hemostasis (107 genes) - KUL

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