

**DISEASE:**  
**Diamond-Blackfan anemia**

<b>NAME:</b>	Diamond-Blackfan anemia
<b>DESCRIPTION:</b>	Blackfan-Diamond anemia (DBA) is a congenital aregenerative and often macrocytic anemia with erythroblastopenia.
<b>ORPHACODE:</b>	124
<b>SYNONYMS:</b>	Congenital PRCA Congenital pure red cell aplasia Diamond-Blackfan anemia syndrome

**XREF(S):**

Orphanet

OMIM

ICD-10

MeSH

MedDRA

OMIM

OMIM

OMIM

OMIM

OMIM

<p style="text-align: center;"><b>ANALYTE(S):</b></p>	<p> <u>RPL18</u>  <u>RPS27</u>  <u>RPL27</u>  <u>RPS15A</u>  <u>TSR2</u>  <u>RPL35</u>  <u>RPL8</u>  <u>HEATR3</u>  <u>RPL31</u>  <u>RPL9</u>  <u>ADA2</u>  <u>RPS20</u>  <u>RPS19</u>  <u>RPS24</u>  <u>GATA1</u>  <u>RPS17</u>  <u>RPS7</u>  <u>RPL5</u>  <u>RPL11</u>  <u>RPL35A</u>  <u>RPS10</u>  <u>RPS26</u>  <u>RPL26</u>  <u>RPL15</u>  <u>RPS29</u>  <u>RPS28</u> </p>
<p><b>CREATED:</b></p>	<p>13 May 2019 - 01:02</p>
<p><b>CHANGED:</b></p>	<p>22 Jun 2023 - 16:14</p>

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- [Periodic Fever \(88 genes\)](#)
- [Polyarteritis nodosa, childhood-onset / Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome](#)
- [Trombosis - Hemostasis \(gene panel\)](#)
- [cleft lip with/without cleft palate \(virtual gene panel\)](#)

### Related Laboratories

- [Centre de Génétique Humaine - Erasme ULB](#)
- [Centre de Génétique Médicale UCL](#)
- [Centrum Menselijke Erfelijkheid - KUL](#)

### Related Analytes

- [adenosine deaminase 2](#)
- [GATA binding protein 1](#)
- [HEAT repeat containing 3](#)
- [ribosomal protein L11](#)
- [ribosomal protein L15](#)
- [ribosomal protein L18](#)
- [ribosomal protein L26](#)
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- [ribosomal protein L31](#)
- [ribosomal protein L35](#)

- [ribosomal protein L35a](#)
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- [ribosomal protein S10](#)
- [ribosomal protein S15a](#)
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- [ribosomal protein S28](#)
- [ribosomal protein S29](#)
- [ribosomal protein S7](#)
- [TSR2 ribosome maturation factor](#)

## Related Gene Panels

- [Cleft lip and palate / dysmorphic facial features / craniofacial anomalies \(255 genes\) - UCL](#)
- [Immunogenetics \(21 genes\)](#)
- [Periodic Fever \(88 genes\) - ULB](#)
- [Trombosis - Hemostasis \(107 genes\) - KUL](#)

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