

**DISEASE:**  
**Blau syndrome**

|                     |                                                                                                                                                                                                                                                                                                                                                                                                  |
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| <b>NAME:</b>        | Blau syndrome                                                                                                                                                                                                                                                                                                                                                                                    |
| <b>DESCRIPTION:</b> | Blau syndrome (BS) is a rare systemic inflammatory disease characterized by early onset granulomatous arthritis, uveitis and skin rash. BS now refers to both the familial and sporadic (formerly early-onset sarcoidosis) form of the same disease. The proposed term pediatric granulomatous arthritis is currently questioned since it fails to represent the systemic nature of the disease. |
| <b>ORPHACODE:</b>   | 90340                                                                                                                                                                                                                                                                                                                                                                                            |
| <b>XREF(S):</b>     | <u>Orphanet</u><br><u>ICD-10</u><br><u>MedDRA</u><br><u>OMIM</u>                                                                                                                                                                                                                                                                                                                                 |
| <b>ANALYTE(S):</b>  | <u>NOD2</u>                                                                                                                                                                                                                                                                                                                                                                                      |
| <b>CREATED:</b>     | 13 May 2019 - 01:02                                                                                                                                                                                                                                                                                                                                                                              |
| <b>CHANGED:</b>     | 22 Jun 2023 - 16:14                                                                                                                                                                                                                                                                                                                                                                              |

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