

**DISEASE:**  
**Alpha delta granule deficiency**

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|---------------------|---|
| <b>NAME:</b>        | Alpha delta granule deficiency  |
| <b>DESCRIPTION:</b> | A rare hemorrhagic disorder due to a constitutional platelet anomaly characterized by moderate to severe deficiency in both platelet alpha-granules and dense bodies, resulting in impaired platelet function and decreased aggregation responses. Patients present increased bleeding tendency with symptoms like easy bruising, or menorrhagia. |
| <b>ORPHACODE:</b>   | 734   |
| <b>SYNONYMS:</b>    | Alpha dense granule deficiency<br>Combined alpha-delta platelet storage pool deficiency   |
| <b>XREF(S):</b>     | <u>Orphanet</u><br><u>OMIM</u><br><u>ICD-10</u>   |
| <b>ANALYTE(S):</b>  | <u>GFI1B</u>  |
| <b>CREATED:</b>     | 13 May 2019 - 01:02   |
| <b>CHANGED:</b>     | 22 Jun 2023 - 16:14   |

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- [Centrum Menselijke Erfelijkheid - KUL](#)

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