

**DISEASE:****DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome**

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| <b>NAME:</b>        | DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome                                                                                                                                                                                              |
| <b>DESCRIPTION:</b> | A rare genetic disease characterized by progressive and severe sensorineural hearing loss with onset in the first decade of life, associated with mild thrombocytopenia, often with enlarged platelets. Most patients do not show significant bleeding tendency. |
| <b>ORPHACODE:</b>   | 494444                                                                                                                                                                                                                                                           |
| <b>SYNONYMS:</b>    | DIAPH1-related sensorineural deafness-thrombocytopenia syndrome                                                                                                                                                                                                  |
| <b>XREF(S):</b>     | <u>Orphanet</u><br><u>OMIM</u><br><u>ICD-10</u>                                                                                                                                                                                                                  |
| <b>ANALYTE(S):</b>  | <u>DIAPH1</u>                                                                                                                                                                                                                                                    |
| <b>CREATED:</b>     | 13 May 2019 - 01:02                                                                                                                                                                                                                                              |
| <b>CHANGED:</b>     | 22 Jun 2023 - 16:14                                                                                                                                                                                                                                              |

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