

**DISEASE:**  
**Hartsfield syndrome**

|                     |                                                                                                                                                                                                                                                                                                                       |
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| <b>NAME:</b>        | Hartsfield syndrome                                                                                                                                                                                                                                                                                                   |
| <b>DESCRIPTION:</b> | A rare, genetic, multiple congenital anomalies syndrome characterized by variable expression of the holoprosencephaly (HPE) spectrum in association with ectrodactyly, cleft lip/palate and/or other ectodermal anomalies. Developmental delay of variable severity and endocrine abnormalities are often associated. |
| <b>ORPHACODE:</b>   | 2117                                                                                                                                                                                                                                                                                                                  |
| <b>SYNONYMS:</b>    | Holoprosencephaly-ectrodactyly-cleft lip/palate syndrome                                                                                                                                                                                                                                                              |
| <b>XREF(S):</b>     | <u>Orphanet</u><br><u>OMIM</u><br><u>ICD-10</u>                                                                                                                                                                                                                                                                       |
| <b>ANALYTE(S):</b>  | <u>FGFR1</u>                                                                                                                                                                                                                                                                                                          |
| <b>CREATED:</b>     | 13 May 2019 - 01:02                                                                                                                                                                                                                                                                                                   |
| <b>CHANGED:</b>     | 22 Jun 2023 - 16:14                                                                                                                                                                                                                                                                                                   |

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