

**DISEASE:****Spondyloepimetaphyseal dysplasia congenita, Strudwick type**

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| <b>NAME:</b>        | Spondyloepimetaphyseal dysplasia congenita, Strudwick type                                                                                                                                                                                                                                                                 |
| <b>DESCRIPTION:</b> | Spondyloepimetaphyseal dysplasia congenita, Strudwick type is characterized by disproportionate short stature from birth (with a very short trunk and shortened limbs) and skeletal abnormalities (lordosis, scoliosis, flattened vertebrae, pectus carinatum, coxa vara, clubfoot, and abnormal epiphyses or metaphyses). |
| <b>ORPHACODE:</b>   | 93346                                                                                                                                                                                                                                                                                                                      |
| <b>XREF(S):</b>     | <u>Orphanet</u><br><u>OMIM</u><br><u>ICD-10</u>                                                                                                                                                                                                                                                                            |
| <b>ANALYTE(S):</b>  | <u>COL2A1</u>                                                                                                                                                                                                                                                                                                              |
| <b>CREATED:</b>     | 13 May 2019 - 01:02                                                                                                                                                                                                                                                                                                        |
| <b>CHANGED:</b>     | 22 Jun 2023 - 16:14                                                                                                                                                                                                                                                                                                        |

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