

**DISEASE:****Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome**

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| <b>NAME:</b>        | Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome                                                                                                                                                                                                                                                                                                                                                                                                                                                                                            |
| <b>DESCRIPTION:</b> | Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome is a rare, genetic, primary bone dysplasia disorder characterized by severe pre- and post-natal short stature, facial dysmorphism (incl.dolicocephaly, long triangular face, tall forehead, down-slanting palpebral fissures, prominent nose, long philtrum, small ears), early-onset or postpubertal sparse, short hair and hypoplastic fingernails. Small hands with tapering fingers, brachydactyly and fifth-finger clinodactyly, as well as a high-pitched voice are also associated. |
| <b>ORPHACODE:</b>   | 314394                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             |
| <b>SYNONYMS:</b>    | SOFT syndrome                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      |
| <b>XREF(S):</b>     | <a href="#">Orphanet</a><br><a href="#">ICD-10</a><br><a href="#">OMIM</a>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         |
| <b>ANALYTE(S):</b>  | <a href="#">POC1A</a>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              |
| <b>CREATED:</b>     | 13 May 2019 - 01:02                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                |
| <b>CHANGED:</b>     | 22 Jun 2023 - 16:14                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                |

## RELATED CONTENT

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### Related Genetic Tests

- [Short stature/ Growth retardation/ \(gene panel\)](#)

### Related Laboratories

- [Centrum Medische Genetica - UZ Antwerpen](#)

### Related Analytes

- [POC1 centriolar protein A](#)

### Related Gene Panels

- [Growth retardation/short stature \(genepanel\) - UZA](#)

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