# DISEASE:
# Saethre-Chotzen syndrome

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| **NAME:** | Saethre-Chotzen syndrome |
| **DESCRIPTION:** | A syndrome characterized by unilateral or bilateral coronal synostosis, facial asymmetry, ptosis, strabismus and small ears with prominent superior and/or inferior crus, among other less common manifestations. |
| **ORPHACODE:** | 794 |
| **SYNONYMS:** | ACS3<br>Acrocephalosyndactyly type 3<br>SCS |
| **XREF(S):** | Orphanet<br>OMIM<br>ICD-10<br>OMIM |
| **ANALYTE(S):** | FGFR3<br>FGFR2<br>TWIST1 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

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# RELATED CONTENT

## Related Genetic Tests

- Craniosynostosis (gene panel)
- Saethre-Chotzen syndrome
- Short Stature (gene panel)
- Short stature/ Growth retardation/ (gene panel)

## Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

## Related Analytes

- fibroblast growth factor receptor 2
- fibroblast growth factor receptor 3
- twist family bHLH transcription factor 1

## Related Gene Panels

- Craniosynostosis (32 genes) - KUL
- Growth retardation/short stature (genepanel) - UZA

- Short Stature (46 genes) - IPG

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