

DISEASE:
Apert syndrome

NAME:	Apert syndrome
DESCRIPTION:	A frequent form of acrocephalosyndactyly, a group of inherited congenital malformation disorders, characterized by craniosynostosis, midface hypoplasia, and finger and toe anomalies and/or syndactyly.
ORPHACODE:	87
SYNONYMS:	ACS1 Acrocephalosyndactyly type 1
XREF(S):	<u>Orphanet</u> <u>MedDRA</u> <u>MeSH</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>FGFR2</u>
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RELATED CONTENT

Related Genetic Tests

- Craniosynostosis / Apert syndrome (hot spot mutations - exon 7)
- Craniosynostosis syndromes (Apert, Crouzon)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- fibroblast growth factor receptor 2

Source URL: <http://gentest.healthdata.be/disease/196>