DISEASE:
Carpenter syndrome

NAME:	Carpenter syndrome
DESCRIPTION:	A rare syndromic craniosynostosis with variable phenotypic expression characterized by craniosynostosis, intellectual disability, distinctive facies, abnormalities of the fingers and toes (brachydactyly, polydactyly and syndactyly), short stature, congenital heart disease, skeletal defects, obesity, genital abnormalities and umbilical hernia.
ORPHACODE:	65759
SYNONYMS:	ACPS2 Acrocephalopolysyndactyly type 2
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	MEGF8 RAB23
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RELATED CONTENT

Related Genetic Tests

- Craniosynostosis (gene panel)
- Early-onset severe obesity

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- multiple EGF like domains 8
- RAB23, member RAS oncogene family

Related Gene Panels

- Craniosynostosis (32 genes) - KUL
- Early-onset severe obesity (44 genes) - ULG