DISEASE:
Cowden syndrome

NAME:	Cowden syndrome
DESCRIPTION:	A genodermatosis characterized by the presence of multiple hamartomas in various tissues and an increased risk for malignancies of the breast, thyroid, endometrium, kidney and colorectum. When CS is accompanied by germline PTEN mutations, it belongs to the PTEN hamartoma tumor syndrome (PHTS) group.
ORPHACODE:	201
SYNONYMS:	Cowden disease Multiple hamartoma syndrome
XREF(S):	Orphanet OMIM MeSH MedDRA ICD-10 OMIM OMIM OMIM OMIM

ANALYTE(S):	PTEN USF3 PIK3CA SDHB SDHD SDHC SEC23B AKT1 KLLN
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RELATED CONTENT

Related Genetic Tests

- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Cowden disease (3 genes)
- Cowden disease (PTEN gene)
- Cowden disease / PTEN hamartoma tumor syndrome
- Epidermal nevus syndrome (gene panel)
- Overgrowth (gene panel)
- Renal cell carcinoma (kidney cancer) (gene panel)
- Venous malformation (3 genes)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- AKT serine/threonine kinase 1
- killin, p53 regulated DNA replication inhibitor
- phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit alpha
- phosphatase and tensin homolog

- succinate dehydrogenase complex iron sulfur subunit B
- succinate dehydrogenase complex subunit C
- succinate dehydrogenase complex subunit D
- SEC23 homolog B, COPII coat complex component
- upstream transcription factor family member 3

Related Gene Panels

- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG
- Cowden (3 genes) - KUL
- Cowden disease (3 genes) -KUL
- Overgrowth (24 genes) - IPG
- Renal cell carcinoma - UGent
- epidermal nevus syndrome (65 genes) - KUL
