DISEASE:
Proteus syndrome

NAME:	Proteus syndrome
DESCRIPTION:	Proteus syndrome (PS) is a very rare and complex hamartomatous overgrowth disorder characterized by progressive overgrowth of the skeleton, skin, adipose, and central nervous systems.
ORPHACODE:	744
SYNONYMS:	Partial gigantism-nevi-hemihypertrophy-macrocephaly syndrome
XREF(S):	Orphanet ICD-10 OMIM MeSH
ANALYTE(S):	PTEN AKT1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Cowden disease (3 genes)
- Cowden disease / PTEN hamartoma tumor syndrome
- Epidermal nevus syndrome (gene panel)
- Epilepsy (gene panel)
- Overgrowth & vascular anomalies / Proteus syndrome (c.49G>A (p.Glu17Lys) mutation)
- Overgrowth (gene panel)
- Vascular malformations (somatic)
- Venous malformation (3 genes)

Related Laboratories

- Centre de Génétique Médicale UCL
- 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

- AKT serine/threonine kinase 1
- phosphatase and tensin homolog

Related Gene Panels

- Cowden (3 genes) - KUL
- Overgrowth (24 genes) - IPG
- Rare epilepsy with developmental delay (> 240 genes) - UZA
- epidermal nevus syndrome (65 genes) - KUL
