DISEASE:
Sturge-Weber syndrome

NAME:	Sturge-Weber syndrome
DESCRIPTION:	A rare congenital neurocutaneous syndrome defined by a facial capillary malformation or port-wine birthmark (PWB) associated with cerebral and ocular ipsilateral vascular malformations in most of the cases resulting in variable ocular and neurological complications.
ORPHACODE:	3205
SYNONYMS:	Encephalofacial angiomatosis Encephalotrigeminal angiomatosis SWS Sturge-Weber-Dimitri syndrome Sturge-Weber-Krabbe angiomatosis Sturge-Weber-Krabbe syndrome
XREF(S):	Orphanet MedDRA MedDRA OMIM MeSH ICD-10
ANALYTE(S):	GNAQ

CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/361

RELATED CONTENT

Related Genetic Tests

- Congenital hemangioma (2 genes)
- Sturge-Weber syndrome (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- G protein subunit alpha q

Source URL: http://gentest.healthdata.be/disease/361