

DISEASE:
Klippel-Trénaunay syndrome

NAME:	Klippel-Trénaunay syndrome
DESCRIPTION:	A rare congenital complex vascular malformation syndrome characterized by capillary malformations manifesting as port-wine stains and venous varicosities typically prominent along the lateral aspect of the lower extremities, associated with overgrowth of a limb (most commonly a leg), more rarely other regions of the body, involving bone and/or soft tissue. The diagnosis is usually made when at least 2 of these 3 features exist. Lymphatic malformations are also observed, while arteriovenous fistulas are absent. Patients present recurrent painful thrombophlebitis, venous thrombosis, and sudden venous hemorrhage.
ORPHACODE:	90308
XREF(S):	<u>Orphanet</u> <u>MedDRA</u> <u>OMIM</u> <u>ICD-10</u>
ANALYTE(S):	<u>AGGF1</u>
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