

DISEASE:
Congenital prekallikrein deficiency

NAME:	Congenital prekallikrein deficiency
DESCRIPTION:	A rare genetic coagulation disorder characterized by the usually incidental laboratory finding of a prolonged activated partial thromboplastin time (aPTT) but normal prothrombin time, due to a deficiency of normal prekallikrein or the presence of nonfunctional prekallikrein. Most patients remain clinically asymptomatic, although an association with cardiovascular conditions (hypertension, myocardial infarction, other coronary artery diseases, and ischemic strokes) and venous thrombosis, as well as rare cases with increased bleeding tendency have been reported.
ORPHACODE:	749
XREF(S):	<u>Orphanet</u> <u>OMIM</u> <u>ICD-10</u>
ANALYTE(S):	<u>KLKB1</u>
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