DISEASE:
Vasculitis due to ADA2 deficiency

NAME:	Vasculitis due to ADA2 deficiency
DESCRIPTION:	Vasculitis due to ADA2 deficiency is a rare, genetic, systemic and rheumatologic disease due to adenosine deaminase-2 inactivating mutations, combining variable features of autoinflammation, vasculitis, and a mild immunodeficiency. Variable clinical presentation includes chronic or recurrent systemic inflammation with fever, livedo reticularis or racemosa, early-onset ischemic or hemorrhagic strokes, peripheral neuropathy, abdominal pain, hepatosplenomegaly, portal hypertension, cutaneous polyarteritis nodosa, variable cytopenia and immunoglobulin deficiency.
ORPHACODE:	404553
SYNONYMS:	Vasculitis due to DADA2
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	ADA2
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Periodic Fever (88 genes)
- Polyarteritis nodosa, childhood-onset / Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome
- Primary immune deficiencies (gene panel)

Related Laboratories

- Centre de Génétique Humaine - Erasme ULB
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- adenosine deaminase 2

Related Gene Panels

- Periodic Fever (88 genes) - ULB
- Primary immune deficiencies (444 genes) - KUL