DISEASE:
Activated PI3K-delta syndrome

NAME:	Activated PI3K-delta syndrome
DESCRIPTION:	A rare, genetic, primary immunodeficiency disease characterized by increased susceptibility to recurrent and/or severe bacterial and viral infections (in particular, sinopulmonary bacterial and herpesvirus infections), chronic benign lymphoproliferation (manifesting as lymphadenopathy, hepatosplenomegaly and focal nodular lymphoid hyperplasia), and/or autoimmune disease (including immune cytopenias, juvenile arthritis, glomerulonephritis and sclerosing cholangitis). Immunophenotypically, variable degrees of agammaglobulinemia with increased IgM levels, increased circulating transitional B cells, decreased naïve CD4 and CD8 T-cells with increased CD8 effector/memory T cells are observed.
ORPHACODE:	397596
SYNONYMS:	APDS Senescent T-cells-lymphadenopathy-immunodeficiency syndrome due to p110delta-activating mutation
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	PTEN PIK3R1 PIK3CD

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RELATED CONTENT

Related Genetic Tests

- Immunodeficiency - Activated PI3K-delta syndrome
- Primary immune deficiencies (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit delta
- phosphoinositide-3-kinase regulatory subunit 1
- phosphatase and tensin homolog

Related Gene Panels

- Immunogenetics (21 genes)
- Primary immune deficiencies (444 genes) - KUL