DISEASE:
Autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation

NAME:	Autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation
DESCRIPTION:	A rare, mixed autoinflammatory and autoimmune syndrome disorder characterized by recurrent neutrophilic blistering skin lesions, arthralgia, ocular inflammation, inflammatory bowel disease, absence of autoantibodies, and mild immunodeficiency manifested by recurrent sinopulmonary infections and deficiency of circulating antibodies. Inflammatory phenotype is not provoked by cold temperatures.
ORPHACODE:	324530
SYNONYMS:	APLAID
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PLCG2
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