

DISEASE:**PLCG2-associated antibody deficiency and immune dysregulation**

NAME:	PLCG2-associated antibody deficiency and immune dysregulation
DESCRIPTION:	PLCG2-associated antibody deficiency and immune dysregulation is a rare, hereditary, immune deficiency with skin involvement characterized by early-onset cold urticaria after generalized exposure to cold air or evaporative cooling and not after contact with cold objects. Additional immunologic abnormalities are often present - antibody deficiency, recurrent infections, autoimmune disease and symptomatic allergic disease.
ORPHACODE:	300359
SYNONYMS:	FACU Familial atypical cold urticaria Familial cold urticaria with common variable immunodeficiency PLAID
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	PLCG2
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