

DISEASE:**Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis**

NAME:	Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis
DESCRIPTION:	A rare, genetic, mixed autoinflammatory and autoimmune syndrome characterized by chronic systemic autoinflammation (presenting as recurrent fever in the neonatal or infantile period) and combined immunodeficiency (manifesting as recurrent viral and invasive bacterial infections). Muscular amylopectinosis may be subclinical or be complicated by myopathy/cardiomyopathy.
ORPHACODE:	329173
XREF(S):	<u>Orphanet</u> <u>OMIM</u> <u>ICD-10</u>
ANALYTE(S):	<u>RBCK1</u> <u>RNF31</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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