DISEASE:
Periodic fever-infantile enterocolitis-autoinflammatory syndrome

NAME:	Periodic fever-infantile enterocolitis-autoinflammatory syndrome
DESCRIPTION:	A rare genetic systemic or rheumatologic disease characterized by neonatal or infantile onset of enterocolitis (which resolves with age), periodic fever, and episodes of severe systemic inflammation, which may be precipitated by infections, stress, or fatigue. Signs and symptoms include splenomegaly, urticaria-like rashes, arthralgia, and myalgia. Associated laboratory findings are elevated inflammatory markers (such as ferritin, C-reactive protein), pancytopenia, and elevated transaminases. If left untreated, flares can progress to coagulopathy, organ failure, and death.
ORPHACODE:	436166
SYNONYMS:	NLRC4-related MAS NLRC4-related autoinflammatory syndrome with MAS NLRC4-related autoinflammatory syndrome with macrophage activation syndrome NLRC4-related infantile enterocolitis-autoinflammatory syndrome NLRC4-related macrophage activation syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	NLRC4
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