DISEASE:
Neonatal inflammatory skin and bowel disease

NAME:	Neonatal inflammatory skin and bowel disease
DESCRIPTION:	Neonatal inflammatory skin and bowel disease is a rare, life-threatening, autoinflammatory syndrome with immune deficiency disorder characterized by early-onset, life-long inflammation, affecting the skin and bowel, associated with recurrent infections. Patients present perioral and perianal psoriasiform erythema and papular eruption with pustules, failure to thrive associated with chronic malabsorptive diarrhea, intercurrent gastrointestinal infections and feeding troubles, as well as absent, short or broken hair and trichomegaly. Recurrent cutaneous and pulmonary infections lead to recurrent blepharitis, otitis externa and bronchiolitis.
ORPHACODE:	294023
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	EGFR ADAM17
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