

DISEASE:
Hyperzincemia and hypercalprotectinemia

NAME:	Hyperzincemia and hypercalprotectinemia
DESCRIPTION:	A rare inborn error of zinc metabolism characterized by recurrent infections, hepatosplenomegaly, anemia (unresponsive to iron supplementation) and chronic systemic inflammation in the presence of high plasma concentrations of zinc and calprotectin. Patients typically present dermal ulcers or other cutaneous manifestations (e.g. inflammation) and arthralgia. Severe epistaxis and spontaneous hematomas have also been reported.
ORPHACODE:	251523
SYNONYMS:	Hz/Hc PAMI syndrome PSTPIP1-associated myeloid-related proteinemia inflammatory syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	PSTPIP1
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