

DISEASE:
Farber disease

NAME:	Farber disease
DESCRIPTION:	A subcutaneous tissue disease characterized by a spectrum of clinical signs ranging from the classical triad of painful and progressively deformed joints, subcutaneous nodules, and progressive hoarseness (due to laryngeal involvement) that presents in infancy, to varying phenotypes with respiratory and neurologic involvement.
ORPHACODE:	333
SYNONYMS:	Acid ceramidase deficiency Farber lipogranulomatosis
XREF(S):	Orphanet ICD-10 MeSH MeSH OMIM
ANALYTE(S):	ASAH1
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