DISEASE:
Ferro-cerebro-cutaneous syndrome

NAME:	Ferro-cerebro-cutaneous syndrome
DESCRIPTION:	Ferro-cerebro-cutaneous syndrome is a rare, genetic, metabolic liver disease characterized by progressive neurodegeneration, cutaneous abnormalities, including varying degrees of ichthyosis or seborrheic dermatitis, and systemic iron overload. Patients manifest with infantile-onset seizures, encephalopathy, abnormal eye movements, axial hypotonia with peripheral hypertonia, brisk reflexes, cortical blindness and deafness, myoclonus and hepato/splenomegaly, as well as oral manifestations, including microdontia, widely spaced and pointed teeth with delayed eruption, and gingival overgrowth.
ORPHACODE:	397922
SYNONYMS:	Cerebro-cutaneous syndrome with iron overload
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	PIGA
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