DISEASE:
Marshall-Smith syndrome

NAME:	Marshall-Smith syndrome
DESCRIPTION:	A rare genetic multiple congenital anomalies syndrome characterized by abnormal bone maturation with skeletal anomalies, airway obstructions, failure to thrive, developmental delay, moderate to severe intellectual disability and characteristic facial features with macrocephaly, prominent forehead, shallow orbits, proptosis and blue sclerae.
ORPHACODE:	561
SYNONYMS:	Accelerated skeletal maturation-facial dysmorphism-failure to thrive syndrome
XREF(S):	Orphanet MeSH OMIM ICD-10
ANALYTE(S):	NFIX
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