DISEASE:
Bohring-Opitz syndrome

NAME:	Bohring-Opitz syndrome
DESCRIPTION:	A rare multiple congenital anomalies syndrome characterized by intrauterine growth retardation (IUGR), postnatal failure to thrive, severe feeding difficulties, microcephaly/trigonocephaly, facial dysmorphism, a recognizable upper limb posture and severe developmental delay. The upper limb posture consists of internal rotation of the shoulders, flexion of the elbows, ulnar deviation of wrists and/or metacarpophalangeal joints.
ORPHACODE:	97297
SYNONYMS:	BOS syndrome Bohring syndrome C-like syndrome Oberklaid-Danks syndrome Opitz trigonocephaly-like syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	ASXL1
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- Centre de Génétique Médicale UCL
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