DISEASE:
Infantile-onset X-linked spinal muscular atrophy

NAME:	Infantile-onset X-linked spinal muscular atrophy
DESCRIPTION:	A rare form of spinal muscular atrophy characterized by the neonatal onset of severe hypotonia, areflexia, profound weakness, multiple congenital contractures, facial dysmorphic features (myopathic face with open, tent-shaped mouth), cryptorchidism, and mild skeletal abnormalities (i.e. kyphosis, scoliosis), that is often preceded by polyhydramnios and reduced fetal movements in utero and followed by bone fractures shortly after birth. Muscle weakness is progressive and chest muscle involvement eventually leads to ventilatory insufficiency and respiratory failure.
ORPHACODE:	1145
SYNONYMS:	SMAX2 Spinal muscular atrophy with arthrogryposis X-linked distal arthrogryposis multiplex congenita X-linked spinal muscular atrophy type 2
XREF(S):	Orphanet MeSH ICD-10 OMIM
ANALYTE(S):	UBA1
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