DISEASE:
X-linked spondyloepimetaphyseal dysplasia

NAME:	X-linked spondyloepimetaphyseal dysplasia
DESCRIPTION:	A rare, genetic primary bone dysplasia disorder characterized by disproportionate short stature with mesomelic short limbs, leg bowing, lumbar lordosis, brachydactyly, joint laxity and a waddling gait. Radiographs show platyspondyly with central protrusion of anterior vertebral bodies, kyphotic angulation and very short long bones with dysplastic epiphyses and flarred, irregular, cupped metaphyses.
ORPHACODE:	93349
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	BGN
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