DISEASE:
Blepharophimosis-intellectual disability syndrome, MKB type

NAME:	Blepharophimosis-intellectual disability syndrome, MKB type
DESCRIPTION:	A rare, X-linked, syndromic, intellectual disability disorder affecting only boys and characterized by global development delay with little or no speech, urogenital abnormalities, including scrotal hypoplasia, micro penis, and cryptorchidism, autistic behavior, and facial dysmorphism. Most typical facial features are ptosis, blepharophimosis, a bulbous nasal tip, a long philtrum, and maxillar hypoplasia with full cheeks. Other variable features include microcephaly, hearing loss, dental anomalies, and hyperextensible joints.
ORPHACODE:	293707
SYNONYMS:	BMRS, MKB type BMRS, Maat-Kievit-Brunner type Blepharophimosis-intellectual disability syndrome, Maat-Kievit-Brunner type X-linked Ohdo syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	MED12
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