DISEASE:
2q37 microdeletion syndrome

NAME:	2q37 microdeletion syndrome
DESCRIPTION:	A rare chromosomal anomaly involving deletion of chromosome band 2q37 and characterized by a broad spectrum of clinical findings including mild-moderate developmental delay/intellectual disability, brachymetaphalangy of digits 3-5, short stature, obesity, hypotonia, specific facial dysmorphism, abnormal behavior, autism or autism spectrum disorder, joint hypermobility/dislocation, and scoliosis.
ORPHACODE:	1001
SYNONYMS:	Albright hereditary osteodystrophy type 3 Albright hereditary osteodystrophy-like syndrome Brachydactyly-intellectual disability syndrome Del(2)(q37) Deletion 2q37 Monosomy 2q37qter
XREF(S):	Orphanet MeSH OMIM ICD-10
ANALYTE(S):	HDAC4
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