DISEASE:
22q11.2 duplication syndrome

NAME:	22q11.2 duplication syndrome
DESCRIPTION:	A rare chromosomal anomaly characterized by an extremely variable clinical phenotype and may include heart defects, urogenital abnormalities, velopharyngeal insufficiency with or without cleft palate, and ranging from multiple defects to mild learning difficulties with some individuals being essentially normal.
ORPHACODE:	1727
SYNONYMS:	22q11.2 microduplication syndrome Dup(22)(q11) Duplication 22q11.2 Trisomy 22q11.2
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	TBX1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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