DISEASE:
22q11.2 deletion syndrome

NAME:	22q11.2 deletion syndrome
DESCRIPTION:	A rare chromosomal anomaly which causes a congenital malformation disorder that is typically characterized by cardiac defects, palatal anomalies, facial dysmorphism, developmental delay and immune deficiency.
ORPHACODE:	567
SYNONYMS:	22q11DS CATCH 22 Cayler cardiofacial syndrome Conotruncal anomaly face syndrome DiGeorge sequence DiGeorge syndrome Microdeletion 22q11.2 Monosomy 22q11 Sedlackova syndrome Shprintzen syndrome Takao syndrome Velocardiofacial syndrome

XREF(S):	Orphanet OMIM OMIM OMIM MedDRA MedDRA ICD-10 MeSH
ANALYTE(S):	TBX1 TBX1 ARVCF GP1BB UFD1 HIRA COMT JMJD1C RREB1 SEC24C
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Analytes

- ARVCF delta catenin family member
- catechol-O-methyltransferase
- glycoprotein Ib platelet subunit beta
- histone cell cycle regulator
- jumonji domain containing 1C
- ras responsive element binding protein 1
- SEC24 homolog C, COPII coat complex component
- T-box transcription factor 1
- ubiquitin recognition factor in ER associated degradation 1