DISEASE:
Williams syndrome

NAME:	Williams syndrome
DESCRIPTION:	A rare genetic multisystemic neurodevelopmental disorder characterized by a distinct facial appearance, cardiac anomalies (most frequently supravalvular aortic stenosis), cognitive and developmental abnormalities, and connective tissue abnormalities (e.g., joint laxity). Facial dysmorphism is characterized by a broad forehead, bitemporal narrowing, periorbital fullness, stellate and/or lacy iris pattern, short upturned nose with bulbous tip, long philtrum, wide mouth, full lips and mild micrognathia.
ORPHACODE:	904
SYNONYMS:	Deletion 7q11.23 Monosomy 7q11.23 Williams-Beuren syndrome
XREF(S):	Orphanet ICD-10 MedDRA OMIM MeSH

ANALYTE(S):	STX1A TBL2 GTF2IRD2 VPS37D FKBP6 RFC2 NCF1 TMEM270 MLXIPL EIF4H DNAJC30 BCL7B METTL27 BUD23 BAZ1B CLIP2 ELN GTF2I GTF2IRD1 LIMK1
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RELATED CONTENT

Related Analytes

- bromodomain adjacent to zinc finger domain 1B
- BAF chromatin remodeling complex subunit BCL7B
- BUD23 rRNA methyltransferase and ribosome maturation factor
- CAP-Gly domain containing linker protein 2
- DnaJ heat shock protein family (Hsp40) member C30
- eukaryotic translation initiation factor 4H
- elastin
- FKBP prolyl isomerase family member 6 (inactive)
- general transcription factor IIi
- GTF2I repeat domain containing 1
- GTF2I repeat domain containing 2
- LIM domain kinase 1
- methyltransferase like 27
- MLX interacting protein like
- neutrophil cytosolic factor 1
- replication factor C subunit 2
- syntaxin 1A
- transducin beta like 2
- transmembrane protein 270
- VPS37D subunit of ESCRT-I