DISEASE:
Temple syndrome due to maternal uniparental disomy of chromosome 14

NAME:	Temple syndrome due to maternal uniparental disomy of chromosome 14
DESCRIPTION:	A rare chromosomal anomaly characterized by prenatal and postnatal growth retardation, hypotonia, motor delay, early puberty, obesity, short adult stature, small hands and feet, mild intellectual disability, and mild dysmorphic facial features (frontal bossing, short nose with wide nasal tip, micrognathia, high palate, short philtrum).
ORPHACODE:	96184
SYNONYMS:	UPD(14)mat
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	MEG3 DLK1 RTL1
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RELATED CONTENT

Related Genetic Tests

- Temple syndrome / Kagami-Ogata Syndrome
- Uniparental Disomy (UDP7; UDP11; UDP14; UDP15; UDP16)
- Uniparental Disomy (UDP7; UDP11; UDP14; UDP20)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- delta like non-canonical Notch ligand 1
- maternally expressed 3
- retrotransposon Gag like 1