DISEASE:
Thanatophoric dysplasia type 2

NAME:	Thanatophoric dysplasia type 2
DESCRIPTION:	A form of thanatophoric dysplasia characterized by prenatal onset of micromelia with straight femurs, platyspondyly, narrow thorax, and cloverleaf skull with increased risk of hydrocephalus and neurological complications. Fetal MRI can identify temporal lobe abnormalities and a narrow foramen magnum. Postnatally, distinctive facial features include macrocephaly, frontal bossing, midface hypoplasia, low nasal bridge, large anterior fontanel, and proptosis. Neonates usually die shortly after birth due to respiratory insufficiency and/or spinal cord/brain stem compression.
ORPHACODE:	93274
SYNONYMS:	Cloverleaf skull-micromelic bone dysplasia syndrome TD2 Thanatophoric dwarfism type 2 Thanatophoric dwarfism-cloverleaf skull syndrome
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	FGFR3
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RELATED CONTENT

Related Genetic Tests

- Craniosynostosis (gene panel)
- Short Stature (gene panel)
- Short stature/ Growth retardation/ (gene panel)
- Thanatophoric dysplasia (hot spot mutations - p.Arg248 / p.Gly370 / p.Ser371 / p.Tyr373 / p.Lys650 / p.X806 in FGFR3 gene)
- Thanatophoric dysplasia (hot spot mutations - p.Tyr373Cys, p.Arg248Cys, p.Ser249Cys)
- Thanatophoric dysplasia types 1 and 2 (hot spot mutation - p.Lys650Glu)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- fibroblast growth factor receptor 3

Related Gene Panels

- Craniosynostosis (32 genes) - KUL
- Growth retardation/short stature (genepanel) - UZA

- Short Stature (46 genes) - IPG
