

GENETIC TEST:
Thanatophoric dysplasia types 1 and 2 (hot spot mutation - p.Lys650Glu)

FULL NAME:	Thanatophoric dysplasia types 1 and 2 (hot spot mutation - p.Lys650Glu)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Mutation screening and sequence analysis of selected exons
METHOD TECHNIQUE:	Bi-directional Sanger Sequence analysis
RIZIV CODE:	565456-565460
ACCREDITATION (ISO 15189):	2022-10-07 / 2027-10-06
TURNAROUND TIME (MAXIMUM):	20 - 60 days
CREATED:	08 Aug 2019 - 12:07
CHANGED:	15 Dec 2022 - 16:17

RELATED CONTENT

Related Diseases

- [Thanatophoric dysplasia type 1](#)
- [Thanatophoric dysplasia type 2](#)

Related Laboratories

- [Centre de Génétique-Institut de Pathologie et de Génétique \(IPG\)](#)

Related Analytes

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