

GENETIC TEST:

Thanatophoric dysplasia (hot spot mutations - p.Arg248 / p.Gly370 / p.Ser371 / p.Tyr373 / p.Lys650 / p.X806 in FGFR3 gene)

FULL NAME:	Thanatophoric dysplasia (hot spot mutations - p.Arg248 / p.Gly370 / p.Ser371 / p.Tyr373 / p.Lys650 / p.X806 in FGFR3 gene)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Targeted variant analysis
METHOD TECHNIQUE:	Bi-directional Sanger Sequence analysis
RIZIV CODE:	565456-565460
TURNAROUND TIME (MAXIMUM):	6 weeks
CREATED:	02 Aug 2019 - 08:42
CHANGED:	20 May 2022 - 11:07

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