

Version

| S.NO | Test Name | Gene list | Specimen Type | Disease Category | Methodology | TAT |
|------|--|-----------|--|-----------------------------|----------------------------------|---------|
| 1 | Fibrodysplasia ossificans progressive (ACVR1) gene sequencing | ACVR1 | DIRECT DNA,Peripheral Blood in EDTA (Purple Top) | Connective Tissue Disorders | Next Generation Sequencing (NGS) | 19 days |
| 2 | Ehlers-Danlos syndrome type VI (PLOD1) deletion/duplication analysis | | DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF) | Connective Tissue Disorders | MLPA | 14 days |
| 3 | Marfan syndrome (FBN1) gene analysis | | Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA | Connective Tissue Disorders | Next Generation Sequencing (NGS) | 19 days |
| 4 | Connective tissue disorder gene panel | | Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA | Connective Tissue Disorders | Next Generation Sequencing (NGS) | 19 days |
| 5 | Ehler Danlos syndrome gene panel | | Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top) | Connective Tissue Disorders | Next Generation Sequencing (NGS) | 19 days |
| 6 | Cutis-laxa gene panel | | Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA | Connective Tissue Disorders | Next Generation Sequencing (NGS) | 19 days |
| 7 | EVC1 and EVC2 deletion/duplication analysis by MLPA | | Placenta,Cord Tissue / Umbilical Cord,Peripheral Blood in EDTA (Purple Top),DNA (POC),Chorionic Villus Sample (CVS),Crushed Ground Rock,Amniotic fluid (AF),Product of conception (POC),Fetal DNA,DIRECT DNA | Skeletal dysplasia | MLPA | 14 days |
| 8 | SQSTM1 gene sequencing | SQSTM1 | Peripheral Blood in EDTA (Purple Top),DIRECT DNA | Skeletal dysplasia | Next Generation Sequencing (NGS) | 19 days |
| 9 | RUNX2 gene sequencing | RUNX2 | DIRECT DNA,Peripheral Blood in EDTA (Purple Top) | Skeletal dysplasia | Next Generation Sequencing (NGS) | 19 days |
| 10 | COL1A1 deletion/duplication analysis | | DIRECT DNA | Skeletal dysplasia | MLPA | 14 days |
| 11 | WISP3 gene sequencing | | Peripheral Blood in EDTA (Purple Top),DIRECT DNA | Skeletal dysplasia | Next Generation Sequencing (NGS) | 19 days |
| 12 | Vitamin D-dependent rickets type 1A (CYP27B1) gene sequencing | | Peripheral Blood in EDTA (Purple Top),DIRECT DNA | Skeletal dysplasia | Next Generation Sequencing (NGS) | 19 days |
| 13 | Townes-Brocks syndrome (SALL1) gene sequencing | | DIRECT DNA,Peripheral Blood in EDTA (Purple Top) | Skeletal dysplasia | Next Generation Sequencing (NGS) | 19 days |
| 14 | SHOX gene sequencing | | Peripheral Blood in EDTA (Purple Top),DIRECT DNA | Skeletal dysplasia | Next Generation Sequencing (NGS) | 19 days |
| 15 | Holt-Oram syndrome (TBX5) gene sequencing | | DIRECT DNA,Peripheral Blood in EDTA (Purple Top) | Skeletal dysplasia | Next Generation Sequencing (NGS) | 19 days |
| 16 | SMARCAL1 PCR | | DIRECT DNA | Skeletal dysplasia | Long and Accurate PCR (LA PCR) | 56 days |
| 17 | Hereditary multiple exostoses gene panel | | Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA | Skeletal dysplasia | Next Generation Sequencing (NGS) | 19 days |
| 18 | Skeletal dysplasia gene panel | | Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS) | Skeletal dysplasia | Next Generation Sequencing (NGS) | 19 days |

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| 19 | Pachydermoperiostosis & primary hypertrophic osteoarthropathy gene panel | | Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS) | Skeletal dysplasia | Next Generation Sequencing (NGS) | 19 days |
| 20 | Osteopetrosis gene panel | | Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top) | Skeletal dysplasia | Next Generation Sequencing (NGS) | 19 days |
| 21 | Osteogenesis imperfecta gene panel | | Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA,Peripheral Blood in EDTA (Purple Top) | Skeletal dysplasia | Next Generation Sequencing (NGS) | 19 days |
| 22 | Multiple exostoses (EXT1 & EXT2) deletion duplication analysis | | Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA | Skeletal dysplasia | MLPA | 14 days |
| 23 | Achondroplasia (FGFR3) gene analysis | | Peripheral Blood in EDTA (Purple Top),Chorionic Villus Sample (CVS),Amniotic fluid (AF),DIRECT DNA | Skeletal dysplasia | Next Generation Sequencing (NGS) | 19 days |