**پانل اختلالات اسکلتی**

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| **ردیف** | **نام بیماری** | **تعداد ژن** | **ژن های مورد بررسی** |
| 1 | Metaphyseal dysplasia | 8 | COL10A1, FGFR3, MMP13, MMP9, PTH1R, RMRP, RUNX2, SBDS |
| 2 | Multiple epiphyseal dysplasia and pseudoachondroplasia | 8 | COL2A1, COL9A1, COL9A2, COL9A3, COMP, MATN3, SLC26A2, PTH1R |
| 3 | Spondylometaphyseal dysplasia and Spondylo-epi-(meta)-physeal dysplasia | 27 | ACP5, B3GALT6, CANT1, CHST3, COL11A1, COL11A2, COL2A1, DDR2, DYM, EIF2AK3, HSPG2, IMPAD1, KIF22, MATN3, MMP13, NKX3-2, PAPSS2, PCYT1A, POP1, RAB33B, RMRP, SLC39A13, SMARCAL1, TRAPPC2, TRPV4, WISP3, XYLT1 |
| 4 | Micromelic dysplasia: acromelic, acromesomelic, mesomelic and ahizo-mesomelic dysplasia | 19 | ADAMTSL2, DDR2, FBN1, FGFR3, GDF5, GPC6, GSC, IFT122, IFT140, IFT43, IHH, NPR2, PDE4D, PRKAR1A, ROR2, TRPS1, WDR35, WNT5A, ZSWIM6 |
| 5 | Short- rib dysplasia | 13 | DYNC2H1, EVC, EVC2, IFT122, IFT140, IFT172, IFT80, NEK1, TTC21B, WDR19, WDR34, WDR35, WDR60 |
| 6 | Chondrodysplasia punctate | 7 | AGPS, ARSE, EBP, GNPAT, LBR, NSDHL, PEX7 |
| 7 | Osteogenesis imperfecta and skeletal dysplasia with decreased bone density | 22 | ANO5, ATP6V0A2, B4GALT7, BMP1, COL1A1, COL1A2, CRTAP, FKBP10, GORAB, IFITM5, LRP5, P3H1, PLOD2, PLS3, PPIB, PYCR1, SERPINF1, SERPINH1, SP7, TMEM38B, TNFRSF11B, WNT1 |
| 8 | Skeletal dysplasia with increased bone density | 28 | AMER1, ANKH, CA2, CLCN7, COL1A1, CTSK, DHCR24, DLX3, FAM20C, GJA1, HPGD, LEMD3, LRP4, LRP5, MTAP, OSTM1, PLEKHM1, PTDSS1, PTH1R, SLCO2A1, SNX10, SOST, TBXAS1, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11 |
| 9 | Skeletal dysplasia with abnormal mineralization | 16 | ALPL, ANKH, AP2S1, CASR, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FGF23, GNA11, PHEX, SLC34A1, SLC34A3, SLC9A3R1, VDR |
| 10 | Isolated limb hypoplasia and limb reduction defects: Splithand/foot; Synostosis; isolated Brachydactyly; Polydactyly; Syndactyly | 20 | BHLHA9, BMP2, BMPR1B, FBLN1, FGF16, GDF5, GJA1, GLI3, HOXA11, HOXD13, IHH, LMBR1, LRP4, NOG, PTHLH, ROR2, TP63, TRPV4, WNT10B, WNT7A |
| 11 | Craniosynostosis syndromes | 20 | ALX4, EFNB1, ERF, FGFR1, FGFR2, FGFR3, FREM1, IFT122, IFT43, IL11RA, MEGF8, MSX2, POR, RAB23, RECQL4, SKI, TCF12, TWIST1, WDR19, WDR35 |
| 12 | Potentially lethal skeletal disorders | 44 | AGPS, ALPL, ARSE, BMPER, CANT1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CRTAP, DHCR7, DLL3, DYNC2H1, EBP, FAM111A, FAM20C, FGFR2, FGFR3, FLNA, FLNB, GDF5, GLI3, GNPAT, IFT80, INPPL1, LBR, LIFR, NEK1, NSDHL, OFD1, P3H1, PEX7, PPIB, PTH1R, RNU4ATAC, SLC26A2, SLC35D1, SOX9, TCTN3, TRIP11, TRPV4, WDR34, WNT7A |
| 13 | Selected Genetic Syndromes with skeletal involvement | 42 | ATR, CCDC8, CDC6, CDKN1C, CDT1, CENPJ, CEP152, CEP63, CREBBP, CUL7, DNA2, EP300, ESCO2, FAM111A, FAM58A, FGF10, FGF9, FGFR2, FGFR3, GDF3, GDF6, LARP7, LMX1B, MEOX1, MGP, NIN, OBSL1, ORC1, ORC4, ORC6, PCNT, POC1A, RBBP8, RECQL4, SALL1, SALL4, SF3B4, SH3PXD2B, TBCE, TBX15, TBX3, TBX5 |
| 14 | Complete Panel- Skeletal disorders | 214 | ACP5, ADAMTSL2, AGPS, ALPL, ALX4, AMER1, ANKH, ANO5, AP2S1, ARSE, ATP6V0A2, ATR, B3GALT6, B4GALT7, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, CA2, CANT1, CASR, CCDC8, CDC6, CDKN1C, CDT1, CENPJ, CEP152, CEP63, CHST3, CLCN5, CLCN7, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CREBBP, CRTAP, CTSK, CUL7, CYP27B1, CYP2R1, DDR2, DHCR24, DHCR7, DLL3, DLX3, DMP1, DNA2, DYM, DYNC2H1, EBP, EFNB1, EIF2AK3, ENPP1, EP300, ERF, ESCO2, EVC, EVC2, FAM111A, FAM20C, FAM58A, FBLN1, FBN1, FGF10, FGF16, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FKBP10, FLNA, FLNB, FREM1, GDF3, GDF5, GDF6, GJA1, GLI3, GNA11, GNPAT, GORAB, GPC6, GSC, HOXA11, HOXD13, HPGD, HSPG2, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT80, IHH, IL11RA, IMPAD1, INPPL1, KIF22, LARP7, LBR, LEMD3, LIFR, LMBR1, LMX1B, LRP4, LRP5, MATN3, MEGF8, MEOX1, MGP, MMP13, MMP9, MSX2, MTAP, NEK1, NIN, NKX3-2, NOG, NPR2, NSDHL, OBSL1, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1, PAPSS2, PCNT, PCYT1A, PDE4D, PEX7, PHEX, PLEKHM1, PLOD2, PLS3, POC1A, POP1, POR, PPIB, PRKAR1A, PTDSS1, PTH1R, PTHLH, PYCR1, RAB23, RAB33B, RBBP8, RECQL4, RMRP, RNU4ATAC, ROR2, RUNX2, SALL1, SALL4, SBDS, SERPINF1, SERPINH1, SF3B4, SH3PXD2B, SKI, SLC26A2, SLC34A1, SLC34A3, SLC35D1, SLC39A13, SLC9A3R1, SLCO2A1, SMARCAL1, SNX10, SOST, SOX9, SP7, TBCE, TBX15, TBX3, TBX5, TBXAS1, TCF12, TCIRG1, TCTN3, TGFB1, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TP63, TRAPPC2, TRIP11, TRPS1, TRPV4, TTC21B, TWIST1, VDR, WDR19, WDR34, WDR35, WDR60, WISP3, WNT1, WNT10B, WNT5A, WNT7A, XYLT1, ZSWIM6 |