|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **S3 Table. Identified loci containing novel SNPs or genes associated with lumbar spine BMD** | | | | | | | | |
| **Locus#** | **SNP** | **Map loc.** | **Gene symbol** | **BMD**  **p-value** | **BMD FDR** | **Wald stats** | **min cond FDR** | **Driving phenotype** |
| 1 | rs12079653 | 1p36.23 | *RERE* | 2,56E-05 | 2,55E-02 | -4.11 | 9,54E-03 | LDL |
| 2 | rs12136689 | 1p36.23 | *RERE* | 9,75E-06 | 1,19E-02 | 4.32 | 6,88E-03 | DBP |
| 7 | rs1889830 | 1p32-p31 | *MACF1* | 1,18E-05 | 1,44E-02 | 4.28 | 4,91E-03 | HDL |
| 7 | rs2275187 | 1p32-p31 | *MACF1* | 1,01E-05 | 1,19E-02 | 4.31 | 4,91E-03 | HDL |
| 8 | rs2566784 | 1p31.3 | *WLS/GNG12-AS1* | 6,67E-08 | **1,72E-04** | -5.27 | 1,62E-04 | SBP |
| 10 | rs7554551 | 1p31.3 | *WLS/MIR1262/GNG12-AS1* | 1,07E-15 | **5,10E-07** | -7.83 | 3,30E-07 | SBP |
| 11 | rs2273368 | 1p13 | *WNT2B* | 1,22E-05 | 1,44E-02 | -4.27 | 4,30E-03 | DBP |
| 11 | rs17030613 | 1p13.2 | *CAPZA1* | 1,14E-05 | 1,44E-02 | -4.28 | 4,30E-03 | DBP |
| 12 | rs12120297 | 1q41 | *SUSD4* | 1,73E-06 | **2,47E-03** | 4.67 | 2,23E-03 | LDL |
| 13 | rs780110 | 2p23.3 | *IFT172* | 3,83E-05 | 3,64E-02 | -4.02 | 7,67E-03 | TG |
| 14 | rs4389358 | 2p16.2 | *C2orf73* | 2,26E-05 | 2,55E-02 | 4.14 | 6,60E-03 | HDL |
| 16 | rs17049689 | 2p16.1 | *FANCL* | 1,40E-05 | 1,44E-02 | -4.24 | 5,79E-03 | DBP |
| 19 | rs7636399 | 3p23 | *SUSD5* | 4,11E-06 | **5,45E-03** | 4.5 | 4,68E-03 | LDL |
| 19 | rs9861369 | 3p23 | *SUSD5* | 5,13E-06 | **6,62E-03** | 4.45 | 3,40E-03 | DBP |
| 19 | rs7630970 | 3p23 | *SUSD5* | 4,93E-06 | **6,62E-03** | 4.46 | 3,11E-03 | DBP |
| 21 | rs6599179 | 3p22.1 | *ULK4* | 6,83E-06 | **8,05E-03** | 4.39 | 3,90E-03 | WHR |
| 21 | rs900569 | 3p22.1 | *ULK4* | 1,81E-05 | 2,11E-02 | 4.18 | 7,60E-03 | DBP |
| 21 | rs9856088 | 3p22.1 | *ULK4* | 5,94E-06 | **8,05E-03** | 4.42 | 3,18E-03 | WHR |
| 21 | rs1717017 | 3p22.1 | *ULK4* | 8,06E-06 | **9,79E-03** | 4.36 | 3,90E-03 | WHR |
| 21 | rs1716655 | 3p22.1 | *ULK4* | 9,12E-06 | 1,19E-02 | -4.33 | 2,95E-03 | DBP |
| 21 | rs9838537 | 3p22.1 | *ULK4* | 2,73E-06 | **3,69E-03** | -4.58 | 1,30E-03 | WHR |
| 22 | rs11720264 | 3p21.31 | *BSN-AS2* | 1,29E-05 | 1,44E-02 | -4.26 | 5,12E-03 | WHR |
| 22 | rs11919311 | 3p21.31 | *BSN* | 3,51E-06 | **4,49E-03** | -4.53 | 1,88E-03 | SBP |
| 22 | rs1060962 | 3p21.31 | *BSN* | 4,41E-06 | **5,45E-03** | -4.48 | 2,33E-03 | SBP |
| 22 | rs4855881 | 3p21 | *APEH* | 3,93E-06 | **5,45E-03** | -4.5 | 1,88E-03 | SBP |
| 24 | rs6787138 | 3q27 | *ATP11B* | 8,66E-06 | **9,79E-03** | -4.34 | 3,90E-03 | WHR |
| 25 | rs6599389 | 4p16.3 | *TMEM175* | 1,30E-05 | 1,44E-02 | 4.25 | 4,28E-03 | WHR |
| 27 | rs11729056 | 4q13.3 | *AREG* | 1,33E-05 | 1,44E-02 | -4.25 | 5,18E-03 | DBP |
| 28 | rs11942753 | 4q21.3 | *DSPP* | 1,31E-05 | 1,44E-02 | 4.25 | 5,83E-03 | WHR |
| 30 | rs4862810 | 4q35.2 | *ZFP42* | 6,97E-06 | **8,05E-03** | -4.39 | 6,68E-03 | T2D |
| 31 | rs3822469 | 5p13 | *SLC1A3* | 8,23E-06 | **9,79E-03** | -4.35 | 6,14E-03 | DBP |
| 36 | rs6465508 | 7q21.3 | *C7orf76* | 3,17E-16 | **5,10E-07** | -7.97 | 2,51E-07 | LDL |
| 36 | rs10808100 | 7q21.3 | *C7orf76* | 3,48E-16 | **5,10E-07** | -7.96 | 2,74E-07 | LDL |
| 36 | rs7781370 | 7q21.3 | *C7orf76* | 3,81E-16 | **5,10E-07** | -7.95 | 1,91E-07 | LDL |
| 36 | rs4370463 | 7q21.3 | *C7orf76* | 3,81E-16 | **5,10E-07** | -7.95 | 2,30E-07 | LDL |
| 36 | rs10085588 | 7q21.3 | *C7orf76* | 2,54E-15 | **5,10E-07** | -7.72 | 2,51E-07 | LDL |
| 36 | rs6971293 | 7q21.3 | *C7orf76* | 3,48E-16 | **5,10E-07** | -7.96 | 2,51E-07 | LDL |
| 36 | rs2272224 | 7q21.3 | *SHFM1* | 1,15E-08 | **4,23E-05** | -5.57 | 3,73E-05 | T1D |
| 36 | rs2922927 | 7q21.3 | *SHFM1* | 1,31E-06 | **2,00E-03** | -4.72 | 1,27E-03 | LDL |
| 36 | rs7786855 | 7q21.3 | *SHFM1* | 2,47E-06 | **3,69E-03** | 4.6 | 2,26E-03 | DBP |
| 38 | rs10274324 | 7q31.31 | *CPED1* | 2,43E-06 | **3,69E-03** | -4.6 | 1,25E-03 | WHR |
| 38 | rs798917 | 7q31.31 | *CPED1* | 2,05E-06 | **3,02E-03** | -4.63 | 1,25E-03 | WHR |
| 38 | rs798915 | 7q31.31 | *CPED1* | 1,94E-06 | **3,02E-03** | -4.64 | 1,25E-03 | WHR |
| 38 | rs2691032 | 7q31.31 | *CPED1* | 1,74E-06 | **2,47E-03** | -4.67 | 1,25E-03 | WHR |
| 38 | rs2536150 | 7q31.31 | *CPED1* | 2,16E-06 | **3,02E-03** | 4.62 | 2,09E-03 | LDL |
| 41 | rs980695 | 8q23-q24.1 | *COLEC10* | 2,07E-06 | **3,02E-03** | -4.63 | 3,54E-03 | LDL |
| 42 | rs10956415 | 8q24 | *MIR1208* | 5,05E-06 | **6,62E-03** | -4.45 | 6,49E-03 | WHR |
| 43 | rs592203 | 9p24.1-p23 | *PTPRD/SNORD27* | 1,93E-05 | **2,11E-02** | 4.17 | 8,12E-03 | SBP |
| 44 | rs2388855 | 10p15 | *KLF6* | 3,56E-06 | **5,45E-03** | -4.52 | 3,80E-03 | LDL |
| 47 | rs4350264 | 10q21.3-q22.1 | *SLC25A16* | 1,01E-05 | 1,19E-02 | 4.31 | 3,98E-03 | LDL |
| 49 | rs11602954 | 11p15.5 | *BET1L* | 1,38E-07 | **2,99E-04** | -5.14 | 2,06E-04 | T1D |
| 53 | rs4514364 | 11p14-p13 | *LGR4* | 3,02E-06 | **4,49E-03** | 4.56 | 1,51E-03 | SBP |
| 53 | rs10767646 | 11p14.1 | *BDNF-AS* | 3,42E-06 | **4,49E-03** | 4.53 | 2,43E-03 | HDL |
| 55 | rs10160701 | 11p11.2 | *AMBRA1/HARBI1* | 5,47E-05 | 4,32E-02 | -3.94 | 9,82E-03 | HDL |
| 55 | rs2306029 | 11p11.2 | *LRP4-AS1/LRP4* | 1,31E-06 | **2,00E-03** | 4.72 | 7,96E-04 | SBP |
| 56 | rs7119750 | 11q13 | *RELA* | 1,08E-06 | **1,61E-03** | 4.76 | 6,39E-04 | SBP |
| 56 | rs7101916 | 11q13 | *RELA* | 5,54E-07 | **8,53E-04** | 4.89 | 3,29E-04 | SBP |
| 56 | rs12421691 | 11q13 | *KAT5* | 6,29E-07 | **1,05E-03** | 4.86 | 4,09E-04 | SBP |
| 58 | rs549932 | 11q14.2 | *TMEM135* | 1,12E-05 | 1,44E-02 | 4.29 | 6,04E-03 | T1D |
| 60 | rs3730071 | 12q12-q13 | *ADCY6/MIR4701* | 2,08E-06 | **3,02E-03** | 4.63 | 1,25E-03 | WHR |
| 61 | rs7311091 | 12q13 | *DDN* | 4,76E-06 | **6,62E-03** | -4.46 | 3,30E-03 | DBP |
| 64 | rs11614913 | 12q13.13 | *MIR196A2* | 1,20E-11 | **5,10E-07** | 6.62 | 1,70E-07 | WHR |
| 64 | rs3803042 | 12q13.13 | *MIR196A2* | 1,20E-11 | **5,10E-07** | 6.62 | 1,70E-07 | WHR |
| 64 | rs894737 | 12q13.13 | *HOXC6/HOXC4/HOXC5* | 2,08E-11 | **5,10E-07** | 6.54 | 1,70E-07 | WHR |
| 64 | rs754133 | 12q13.13 | *HOXC6/HOXC4/HOXC5* | 4,55E-11 | **5,10E-07** | 6.43 | 2,10E-07 | WHR |
| 66 | rs1047796 | 12q21.31 | *MLXIP* | 2,45E-05 | 2,55E-02 | 4.12 | 9,23E-03 | DBP |
| 71 | rs12438366 | 15q21-q22 | *SMAD3* | 1,03E-05 | 1,19E-02 | -4.3 | 7,68E-03 | DBP |
| 72 | rs7173826 | 15q22.33-q23 | *AAGAB* | 1,55E-05 | 1,75E-02 | -4.22 | 6,27E-03 | DBP |
| 79 | rs7350980 | 17q21.31 | *KANSL1* | 6,31E-08 | **1,72E-04** | 5.28 | 1,10E-04 | T1D |
| 79 | rs7221390 | 17q21.31 | *KANSL1* | 6,31E-08 | **1,72E-04** | 5.28 | 1,10E-04 | T1D |
| 79 | rs9303525 | 17q21.31 | *KANSL1* | 5,48E-08 | **1,42E-04** | 5.3 | 1,40E-04 | SBP |
| 79 | rs2696689 | 17q21.31 | *KANSL1-AS1* | 1,70E-07 | **3,53E-04** | 5.1 | 1,48E-04 | WHR |
| 80 | rs4794031 | 17q21.32 | *ZNF652* | 8,59E-06 | **9,79E-03** | 4.34 | 4,30E-03 | DBP |
| 80 | rs3179840 | 17q21.32 | *ZNF652* | 1,26E-05 | 1,44E-02 | 4.26 | 4,91E-03 | HDL |
| 80 | rs2906093 | 17q21.32 | *ZNF652* | 1,60E-05 | 1,75E-02 | 4.21 | 6,10E-03 | LDL |
| 81 | rs7226305 | 17q22 | *KIF2B* | 7,24E-07 | **1,30E-03** | -4.84 | 4,49E-04 | DBP |
| 81 | rs17730919 | 17q22 | *KIF2B* | 3,33E-06 | **4,49E-03** | 4.54 | 2,55E-03 | DBP |
| 82 | rs12601958 | 17q24.2 | *CEP112* | 5,42E-05 | 4,32E-02 | -3.94 | 8,74E-03 | TG |
| 83 | rs622924 | 18q21.33 | *PIGN* | 2,83E-06 | **4,49E-03** | -4.57 | 2,91E-03 | T2D |
| 83 | rs583339 | 18q21.33 | *PIGN* | 3,02E-06 | **4,49E-03** | -4.56 | 2,91E-03 | T2D |
| 87 | rs7227107 | 18q23 | *NFATC1* | 7,21E-06 | **9,79E-03** | -4.38 | 4,16E-03 | LDL |
| 89 | rs3760846 | 19q13.32 | *ERCC1* | 6,12E-06 | **8,05E-03** | 4.41 | 2,64E-03 | LDL |
| 90 | rs4813035 | 20p12 | *SLX4IP* | 2,35E-05 | 2,55E-02 | -4.13 | 7,60E-03 | DBP |
| 94 | rs17404303 | 20q13 | *BMP7* | 5,43E-06 | **6,62E-03** | 4.44 | 3,25E-03 | T1D |
| 95 | rs4817775 | 21 | *CBR3-AS1* | 8,17E-07 | **1,30E-03** | 4.81 | 5,12E-04 | SBP |
| Independent complex or single gene loci (LD-r2 < 0.2) with SNP(s) with a conditional FDR (condFDR) < 0.01 in bone mineral density (BMD, lumbar spine) given the association in other phenotypes. We defined the most significant BMD SNP in each LD block based on the minimum condFDR (min condFDR) for each phenotype. The second phenotype which provides the minimal FDR signal (driving phenotype) is listed. All loci with SNPs with condFDR < 0.01 were used to define the number of the loci. The following abbreviations were used: type 1 diabetes (T1D), type 2 diabetes (T2D), systolic blood pressure (SBP), diastolic blood pressure (DBP), high density lipoprotein (HDL), low density lipoprotein (LDL), triglycerides (TG), waist hip ratio (WHR), chromosome location (Map Loc.). Shaded r values represent nominally significant (p<0.05) Pearson correlations (age and BMI adjusted LS BMD vs Affymetrix signal values). Wald stats: z-score transformed from p values. | | | | | | | | |