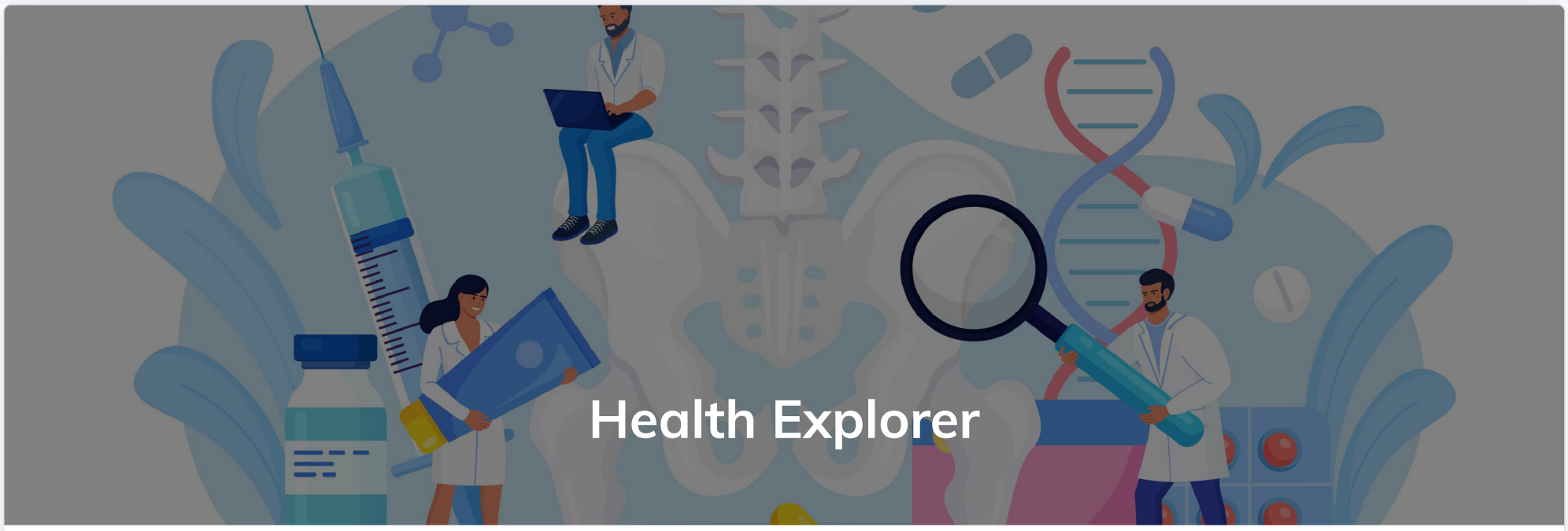

DEMO REPORT

Skeletal

The human skeleton (Osseous system) is the body's internal skeleton structure. Some several bones and cartilages make up this support system. Bands of fibrous connective tissue, such as ligaments and tendons, also attach bones. The skeleton of a healthy adult is the primary focus of this article, which explores both its physical anatomy and function. A person's skeleton serves as a foundation for their whole body. All of these functions are carried out by and via the connective tissue that consists of the human body's connective tissue. The bones of the head, neck, shoulders, arms, and legs are all part of the skeleton, including the spinal column, collarbones, and shoulder blades.

Disclaimer This report is intended as educational information. It is not intended to provide medical advice or be used solely by the customer in the diagnosis, cure, mitigation, treatment or prevention of disease. If you have any serious medical condition(s), including but not limited to, being over or under weight, or having diabetes or heart disease, you should not make any changes to your diet or exercise without consulting your doctor. Under no circumstances, should you make changes to your medication or other medical care without consulting your physician

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18 entries

Full View

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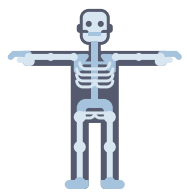
Bone mineral density, heel

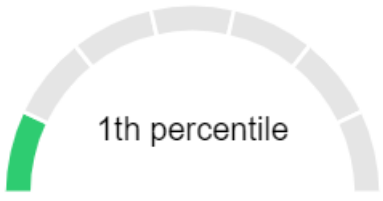


Skeletal

This result is based on **1088 genetic variants** associated with **"Bone mineral density, heel"** analyzed in the scientific paper (2019 Feb - Morris JA)

Your results

Very low genetic predisposition



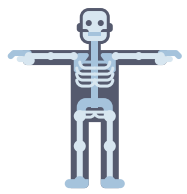
Your genetic predisposition is lower than the 99% average person from your genetic population

Study description

Osteoporosis is a common aging-related disease diagnosed primarily using bone mineral density (BMD). We assessed genetic determinants of BMD as estimated by heel quantitative ultrasound in 426,824 individuals, identifying 518 genome-wide significant loci (301 novel), explaining 20% of its variance. We identified 13 bone fracture loci, all associated with estimated BMD (eBMD), in ~1.2 million individuals. We then identified target genes enriched for genes known to influence bone density and strength (maximum odds ratio (OR) = 58, P = 1 × 10-75) from cell-specific features, including chromatin conformation and accessible chromatin sites. We next performed rapid-throughput skeletal phenotyping of 126 knockout mice with disruptions in predicted target genes and found an increased abnormal skeletal phenotype frequency compared to 526 unselected lines (P < 0.0001). In-depth analysis of one gene, DAAM2, showed a disproportionate decrease in bone strength relative to mineralization. This genetic atlas provides evidence linking associated SNPs to causal genes, offers new insight into osteoporosis pathophysiology, and highlights opportunities for drug development.

Learn more

Ossification of the posterior longitudinal ligament of the spine

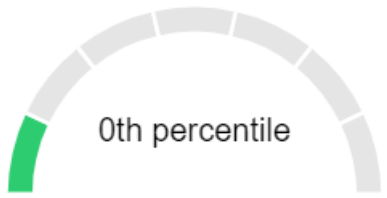


Skeletal

This result is based on **8 genetic variants** associated with **"Ossification of the posterior longitudinal ligament of the spine"** analyzed in the scientific paper [(2014 Sep - Nakajima M)]

Your results

Very low genetic predisposition



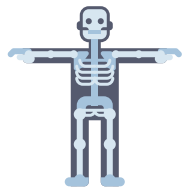
Your genetic predisposition is lower than the 100% average person from your genetic population

Study description

Ossification of the posterior longitudinal ligament of the spine (OPLL) is a common spinal disorder among the elderly that causes myelopathy and radiculopathy. To identify genetic factors for OPLL, we performed a genome-wide association study (GWAS) in ~8,000 individuals followed by a replication study using an additional ~7,000 individuals. We identified six susceptibility loci for OPLL: 20p12.3 (rs2423294: P = 1.10 × 10(-13)), 8q23.1 (rs374810: P = 1.88 × 10(-13)), 12p11.22 (rs1979679: P = 4.34 × 10(-12)), 12p12.2 (rs11045000: P = 2.95 × 10(-11)), 8q23.3 (rs13279799: P = 1.28 × 10(-10)) and 6p21.1 (rs927485: P = 9.40 × 10(-9)). Analyses of gene expression in and around the loci suggested that several genes are involved in OPLL etiology through membranous and/or endochondral ossification processes. Our results bring new insight to the etiology of OPLL.

Learn more

Cranial length, maximum

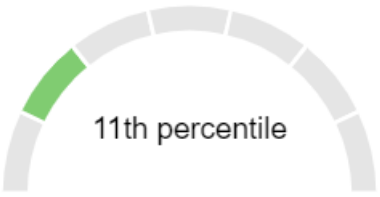


Skeletal

This result is based on **22 genetic variants** associated with **"Cranial length, maximum"** analyzed in the scientific paper [(04/26/2018 - Roosenboom J)]

Your results

Low genetic predisposition



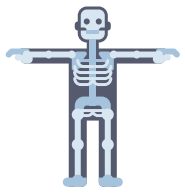
Your genetic predisposition is lower than the 89% average person from your genetic population

Study description

The shape of the cranial vault, a region comprising interlocking flat bones surrounding the cerebral cortex, varies considerably in humans. Strongly influenced by brain size and shape, cranial vault morphology has both clinical and evolutionary relevance. However, little is known about the genetic basis of normal vault shape in humans. We performed a genome-wide association study (GWAS) on three vault measures (maximum cranial width [MCW], maximum cranial length [MCL], and cephalic index [CI]) in a sample of 4419 healthy individuals of European ancestry. All measures were adjusted by sex, age, and body size, then tested for association with genetic variants spanning the genome. GWAS results for the two cohorts were combined via meta-analysis. Significant associations were observed at two loci: 15p11.2 (lead SNP rs2924767, p = 2.107 × 10-8) for MCW and 17q11.2 (lead SNP rs72841279, p = 5.29 × 10-9) for MCL. Additionally, 32 suggestive loci (p < 5x10-6) were observed. Several candidate genes were located in these loci, such as NLK, MEF2A, SOX9 and SOX11. Genome-wide linkage analysis of cranial vault shape in mice (N = 433) was performed to follow-up the associated candidate loci identified in the human GWAS. Two loci, 17q11.2 (c11.loc44 in mice) and 17q25.1 (c11.loc74 in mice), associated with cranial vault size in humans, were also linked with cranial vault size in mice (LOD scores: 3.37 and 3.79 respectively). These results provide further insight into genetic pathways and mechanisms underlying normal variation in human craniofacial morphology.

Learn more

Shoulder impingement syndrome

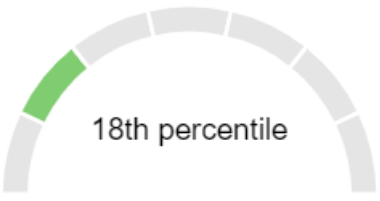


Skeletal

This result is based on **20 genetic variants** associated with **"Shoulder impingement syndrome"** analyzed in the scientific paper **(09/02/2020 - Cheng B)**

Your results

Low genetic predisposition



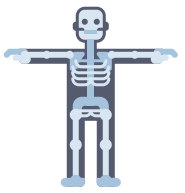
Your genetic predisposition is lower than the 82% average person from your genetic population

Study description

Shoulder impingement syndrome (SIS) is a common shoulder disorder with unclear genetic mechanism. In this study, Genome-wide Association Study (GWAS) was conducted to identify the candidate loci associated with SIS by using the UK Biobank samples (including 3,626 SIS patients and 3,626 control subjects). Based on the GWAS results, gene set enrichment analysis was further performed to detect the candidate gene ontology and pathways associated with SIS. We identified multiple risk loci associated with SIS, such as rs750968 (P = 4.82 × 10-8), rs754832 (P = 4.83 × 10-8) and rs1873119 (P = 6.39 × 10-8) of ANXA1 gene. Some candidate pathways have been identified related to SIS, including those linked to infection response and hypoxia, "ZHOU_INFLAMMATORY_RESPONSE_FIMA_DN" (P = 0.012) and "MANALO_HYPOXIA_UP" (P = 5.00 × 10-5). Our results provide novel clues for understanding the genetic mechanism of SIS.

Learn more

Bone fracture in osteoporosis

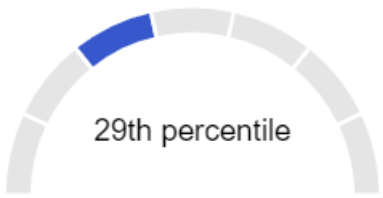


Skeletal

This result is based on **8 genetic variants** associated with **"Bone fracture in osteoporosis"** analyzed in the scientific paper **(08/27/2016 - Taylor KC)**

Your results

Below average genetic predisposition



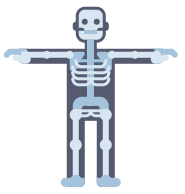
Your genetic predisposition is lower than the 71% average person from your genetic population

Study description

Osteoporosis is a major public health problem associated with excess disability and mortality. It is estimated that 50-70% of the variation in osteoporotic fracture risk is attributable to genetic factors. The purpose of this hypothesis-generating study was to identify possible genetic determinants of fracture among African American (AA) women in a GWAS meta-analysis.

Learn more

Bone mineral density (paediatric, total body less head) (higher)

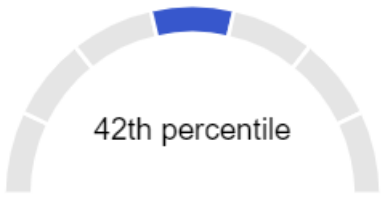


Skeletal

This result is based on **10 genetic variants** associated with **"Bone mineral density (paediatric, total body less head) (higher)"** analyzed in the scientific paper **(07/25/2017 - Medina-Gomez C)**

Your results

Average genetic predisposition



Your genetic predisposition is lower than the 58% average person from your genetic population

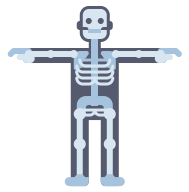
Study description

Bone mineral density is known to be a heritable, polygenic trait whereas genetic variants contributing to lean mass variation remain largely unknown. We estimated the shared SNP heritability and performed a bivariate GWAS meta-

analysis of total-body lean mass (TB-LM) and total-body less head bone mineral density (TBLH-BMD) regions in 10,414 children. The estimated SNP heritability is 43% (95% CI: 34-52%) for TBLH-BMD, and 39% (95% CI: 30-48%) for TB-LM, with a shared genetic component of 43% (95% CI: 29-56%). We identify variants with pleiotropic effects in eight loci, including seven established bone mineral density loci: WNT4, GALNT3, MEPE, CPED1/WNT16, TNFSF11, RIN3, and PPP6R3/LRP5. Variants in the TOM1L2/SREBF1 locus exert opposing effects TB-LM and TBLH-BMD, and have a stronger association with the former trait. We show that SREBF1 is expressed in murine and human osteoblasts, as well as in human muscle tissue. This is the first bivariate GWAS meta-analysis to demonstrate genetic factors with pleiotropic effects on bone mineral density and lean mass.Bone mineral density and lean skeletal mass are heritable traits. Here, Medina-Gomez and colleagues perform bivariate GWAS analyses of total body lean mass and bone mass density in children, and show genetic loci with pleiotropic effects on both traits.

Learn more

Cranial width, maximum

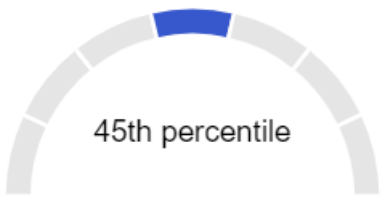


Skeletal

This result is based on **20 genetic variants** associated with **"Cranial width, maximum"** analyzed in the scientific paper **(04/26/2018 - Roosenboom J)**

Your results

Average genetic predisposition



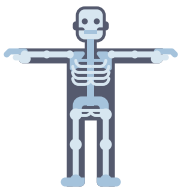
Your genetic predisposition is lower than the 55% average person from your genetic population

Study description

The shape of the cranial vault, a region comprising interlocking flat bones surrounding the cerebral cortex, varies considerably in humans. Strongly influenced by brain size and shape, cranial vault morphology has both clinical and evolutionary relevance. However, little is known about the genetic basis of normal vault shape in humans. We performed a genome-wide association study (GWAS) on three vault measures (maximum cranial width [MCW], maximum cranial length [MCL], and cephalic index [CI]) in a sample of 4419 healthy individuals of European ancestry. All measures were adjusted by sex, age, and body size, then tested for association with genetic variants spanning the genome. GWAS results for the two cohorts were combined via meta-analysis. Significant associations were observed at two loci: 15p11.2 (lead SNP rs2924767, p = 2.107 × 10-8) for MCW and 17q11.2 (lead SNP rs72841279, p = 5.29 × 10-9) for MCL. Additionally, 32 suggestive loci (p < 5x10-6) were observed. Several candidate genes were located in these loci, such as NLK, MEF2A, SOX9 and SOX11. Genome-wide linkage analysis of cranial vault shape in mice (N = 433) was performed to follow-up the associated candidate loci identified in the human GWAS. Two loci, 17q11.2 (c11.loc44 in mice) and 17q25.1 (c11.loc74 in mice), associated with cranial vault size in humans, were also linked with cranial vault size in mice (LOD scores: 3.37 and 3.79 respectively). These results provide further insight into genetic pathways and mechanisms underlying normal variation in human craniofacial morphology.

Learn more

Temporomandibular joint disorder

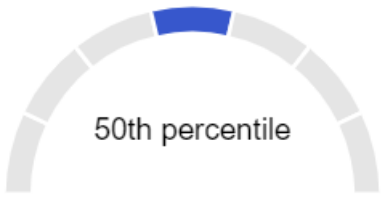


Skeletal

This result is based on **5 genetic variants** associated with **"Temporomandibular joint disorder"** analyzed in the scientific paper [(2017 Mar - Sanders AE)]

Your results

Average genetic predisposition



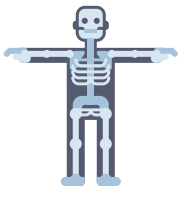
Your genetic predisposition is higher than the 50% average person from your genetic population

Study description

Temporomandibular disorder (TMD) is a musculoskeletal condition characterized by pain and reduced function in the temporomandibular joint and/or associated masticatory musculature. Prevalence in the United States is 5% and twice as high among women as men. We conducted a discovery genome-wide association study (GWAS) of TMD in 10,153 participants (769 cases, 9,384 controls) of the US Hispanic Community Health Study/Study of Latinos (HCHS/SOL). The most promising single-nucleotide polymorphisms (SNPs) were tested in meta-analysis of 4 independent cohorts. One replication cohort was from the United States, and the others were from Germany, Finland, and Brazil, totaling 1,911 TMD cases and 6,903 controls. A locus near the sarcoglycan alpha (SGCA), rs4794106, was suggestive in the discovery analysis (P = 2.6 × 106) and replicated (i.e., 1-tailed P = 0.016) in the Brazilian cohort. In the discovery cohort, sex-stratified analysis identified 2 additional genome-wide significant loci in females. One lying upstream of the relaxin/insulin-like family peptide receptor 2 (RXP2) (chromosome 13, rs60249166, odds ratio [OR] = 0.65, P = 3.6 × 10-8) was replicated among females in the meta-analysis (1-tailed P = 0.052). The other (chromosome 17, rs1531554, OR = 0.68, P = 2.9 × 10-8) was replicated among females (1-tailed P = 0.002), as well as replicated in meta-analysis of both sexes (1-tailed P = 0.021). A novel locus at genome-wide level of significance (rs73460075, OR = 0.56, P = 3.8 × 10-8) in the intron of the dystrophin gene DMD (X chromosome), and a suggestive locus on chromosome 7 (rs73271865, P = 2.9 × 10-7) upstream of the Sp4 Transcription Factor (SP4) gene were identified in the discovery cohort, but neither of these was replicated. The SGCA gene encodes SGCA, which is involved in the cellular structure of muscle fibers and, along with DMD, forms part of the dystrophin-glycoprotein complex. Functional annotation suggested that several of these variants reside in loci that regulate processes relevant to TMD pathobiologic processes.

Learn more

Bone mineral density (spine) (higher)

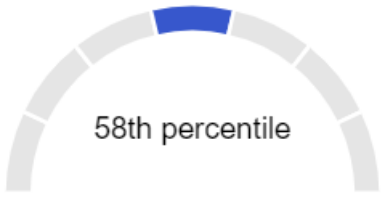


Skeletal

This result is based on **7 genetic variants** associated with **"Bone mineral density (spine) (higher)"** analyzed in the scientific paper [(01/06/2016 - Styrkarsdottir U)]

Your results

Average genetic predisposition



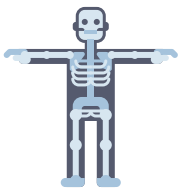
Your genetic predisposition is higher than the 58% average person from your genetic population

Study description

Bone mineral density (BMD) is a measure of osteoporosis and is useful in evaluating the risk of fracture. In a genome-wide association study of BMD among 20,100 Icelanders, with follow-up in 10,091 subjects of European and East-Asian descent, we found a new BMD locus that harbours the PTCH1 gene, represented by rs28377268 (freq. 11.4-22.6%) that associates with reduced spine BMD (P=1.0 × 10(-11), β=-0.09). We also identified a new spine BMD signal in RSPO3, rs577721086 (freq. 6.8%), that associates with increased spine BMD (P=6.6 × 10(-10), β=0.14). Importantly, both variants associate with osteoporotic fractures and affect expression of the PTCH1 and RSPO3 genes that is in line with their influence on BMD and known biological function of these genes. Additional new BMD signals were also found at the AXIN1 and SOST loci and a new lead SNP at the EN1 locus.

Learn more

Hallux valgus

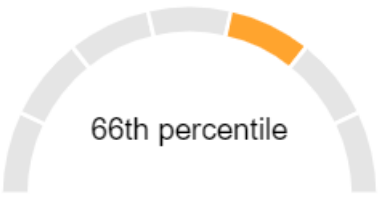


This result is based on **44 genetic variants** associated with **"Hallux valgus"** analyzed in the scientific paper **(03/04/2020 - Arbeeva L)**

Skeletal

Your results

Above average genetic predisposition



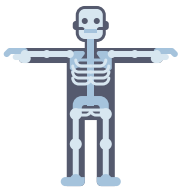
Your genetic predisposition is higher than the 66% average person from your genetic population

Study description

Hallux valgus, one of the most common structural foot deformities, is highly heritable. However, previous efforts to elucidate the genetic underpinnings of hallux valgus through a genome-wide association study (GWAS) conducted in 4409 Caucasians did not identify genome-wide significant associations with hallux valgus in both gender-specific and sex-combined GWAS meta-analyses. In this analysis, we add newly available data and more densely imputed genotypes to identify novel genetic variants associated with hallux valgus.

Learn more

Paget's disease

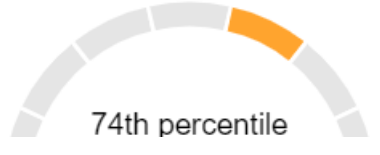


This result is based on **9 genetic variants** associated with **"Paget's disease"** analyzed in the scientific paper **(05/29/2011 - Albagha OM)**

Skeletal

Your results

Above average genetic predisposition



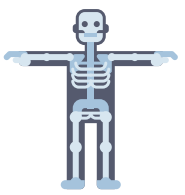
Your genetic predisposition is higher than the 74% average person from your genetic population

Study description

Paget's disease of bone (PDB) is a common disorder characterized by focal abnormalities of bone remodeling. We previously identified variants at the CSF1, OPTN and TNFRSF11A loci as risk factors for PDB by genome-wide association study. Here we extended this study, identified three new loci and confirmed their association with PDB in 2,215 affected individuals (cases) and 4,370 controls from seven independent populations. The new associations were with rs5742915 within PML on 15q24 (odds ratio (OR) = 1.34, P = 1.6 × 10(-14)), rs10498635 within RIN3 on 14q32 (OR = 1.44, P = 2.55 × 10(-11)) and rs4294134 within NUP205 on 7q33 (OR = 1.45, P = 8.45 × 10(-10)). Our data also confirmed the association of TM7SF4 (rs2458413, OR = 1.40, P = 7.38 × 10(-17)) with PDB. These seven loci explained ~13% of the familial risk of PDB. These studies provide new insights into the genetic architecture and pathophysiology of PDB.

Learn more

Lumbar spine bone mineral density (higher)

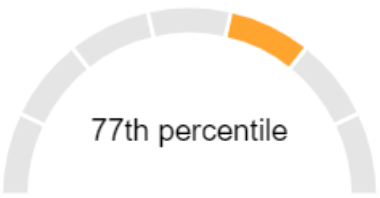


Skeletal

This result is based on **34 genetic variants** associated with **"Lumbar spine bone mineral density (higher)"** analyzed in the scientific paper [(2018 May - Pei YF)]

Your results

Above average genetic predisposition



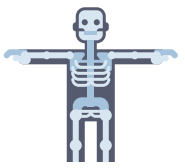
Your genetic predisposition is higher than the 77% average person from your genetic population

Study description

In the present study, aiming to identify loci associated with osteoporosis, we conducted a joint association study of 2 independent genome-wide association meta-analyses of femoral neck and lumbar spine bone mineral densities (BMDs): 1) an in-house study of 6 samples involving 7484 subjects, and 2) the GEFOS-seq study of 7 samples involving 32,965 subjects. The in-house samples were imputed by the 1000 genomes project phase 3 reference panel. SNP-based association test was applied to 7,998,108 autosomal SNPs in each meta-analysis, and for each SNP the 2 association signals were then combined for joint analysis and for mutual replication. Combining the evidence from both studies, we identified 2 novel loci associated with BMDs at the genome-wide significance level (α=5.0×10-8): 20p12.1 (rs73100693 p=2.65×10-8, closest gene MACROD2) and 20q13.33 (rs2380128 p=3.44×10-8, OSBPL2). We also replicated 7 loci that were reported by two recent studies on heel and total body BMD. Our findings provide useful insights that enhance our understanding of bone development, osteoporosis and fracture pathogenesis.

Learn more

Ankle injury



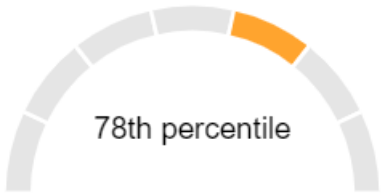
This result is based on **42 genetic variants** associated with **"Ankle injury"** analyzed in the scientific paper

(09/28/2017 - Kim SK)

Skeletal

Your results

Above average genetic predisposition



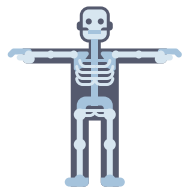
Your genetic predisposition is higher than the 78% average person from your genetic population

Study description

Ankle injuries, including sprains, strains and other joint derangements and instability, are common, especially for athletes involved in indoor court or jumping sports. Identifying genetic loci associated with these ankle injuries could shed light on their etiologies. A genome-wide association screen was performed using publicly available data from the Research Program in Genes, Environment and Health (RPGEH) including 1,694 cases of ankle injury and 97,646 controls. An indel (chr21:47156779:D) that lies close to a collagen gene, COL18A1, showed an association with ankle injury at genome-wide significance (p = 3.8x10-8; OR = 1.99; 95% CI = 1.75-2.23). A second DNA variant (rs13286037 on chromosome 9) that lies within an intron of the transcription factor gene NFIB showed an association that was nearly genome-wide significant (p = 5.1x10-8; OR = 1.63; 95% CI = 1.46-1.80). The ACTN3 R577X mutation was previously reported to show an association with acute ankle sprains, but did not show an association in this cohort. This study is the first genome-wide screen for ankle injury that yields insights regarding the genetic etiology of ankle injuries and provides DNA markers with the potential to inform athletes about their genetic risk for ankle injury.

Learn more

Bone mineral density (hip) (higher)

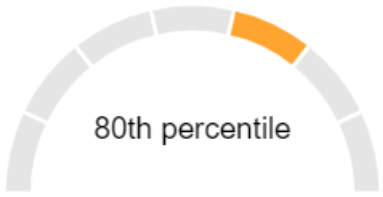


This result is based on **7 genetic variants** associated with **"Bone mineral density (hip) (higher)"** analyzed in the scientific paper (01/06/2016 - Styrkarsdottir U)

Skeletal

Your results

Above average genetic predisposition



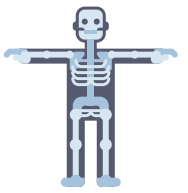
Your genetic predisposition is higher than the 80% average person from your genetic population

Study description

Bone mineral density (BMD) is a measure of osteoporosis and is useful in evaluating the risk of fracture. In a genome-wide association study of BMD among 20,100 Icelanders, with follow-up in 10,091 subjects of European and East-Asian descent, we found a new BMD locus that harbours the PTCH1 gene, represented by rs28377268 (freq. 11.4-22.6%) that associates with reduced spine BMD (P=1.0 × 10(-11), β=-0.09). We also identified a new spine BMD signal in RSPO3, rs577721086 (freq. 6.8%), that associates with increased spine BMD (P=6.6 × 10(-10), β=0.14). Importantly, both variants associate with osteoporotic fractures and affect expression of the PTCH1 and RSPO3 genes that is in line with their influence on BMD and known biological function of these genes. Additional new BMD signals were also found at the AXIN1 and SOST loci and a new lead SNP at the EN1 locus.

Learn more

Fractures

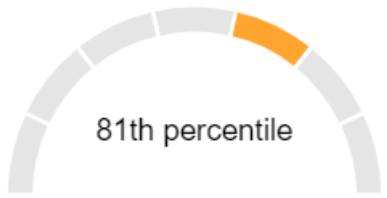


Skeletal

This result is based on **21 genetic variants** associated with **"Fractures"** analyzed in the scientific paper (08/29/2018 - Trajanoska K)

Your results

Above average genetic predisposition



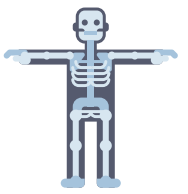
Your genetic predisposition is higher than the 81% average person from your genetic population

Study description

To identify the genetic determinants of fracture risk and assess the role of 15 clinical risk factors on osteoporotic fracture risk.

Learn more

Bone mineral density (paediatric, upper limb) (higher)

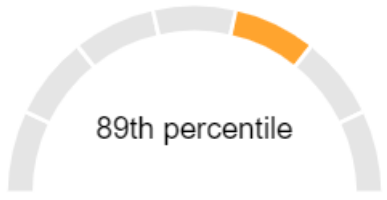


Skeletal

This result is based on **6 genetic variants** associated with **"Bone mineral density (paediatric, upper limb) (higher)"** analyzed in the scientific paper (06/19/2014 - Kemp JP)

Your results

Above average genetic predisposition



Your genetic predisposition is higher than the 89% average person from your genetic population

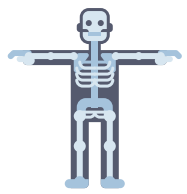
Study description

Heritability of bone mineral density (BMD) varies across skeletal sites, reflecting different relative contributions of genetic and environmental influences. To quantify the degree to which common genetic variants tag and environmental factors influence BMD, at different sites, we estimated the genetic (rg) and residual (re) correlations between BMD measured at the upper limbs (UL-BMD), lower limbs (LL-BMD) and skull (SK-BMD), using total-body DXA scans of ~ 4,890 participants recruited by the Avon Longitudinal Study of Parents and their Children (ALSPAC). Point estimates of rg indicated that appendicular sites have a greater proportion of shared genetic architecture (LL-/UL-BMD rg = 0.78) between them, than with the skull (UL-/SK-BMD rg = 0.58 and LL-/SK-BMD rg = 0.43). Likewise, the residual correlation between BMD at appendicular sites (r(e) = 0.55) was higher than the residual correlation between SK-BMD and BMD at appendicular sites (r(e) = 0.20-0.24). To explore the basis for the observed differences in rg and re, genome-wide association meta-analyses

were performed (n ~ 9,395), combining data from ALSPAC and the Generation R Study identifying 15 independent signals from 13 loci associated at genome-wide significant level across different skeletal regions. Results suggested that previously identified BMD-associated variants may exert site-specific effects (i.e. differ in the strength of their association and magnitude of effect across different skeletal sites). In particular, variants at CPED1 exerted a larger influence on SK-BMD and UL-BMD when compared to LL-BMD (P = 2.01 × 10(-37)), whilst variants at WNT16 influenced UL-BMD to a greater degree when compared to SK- and LL-BMD (P = 2.31 × 10(-14)). In addition, we report a novel association between RIN3 (previously associated with Paget's disease) and LL-BMD (rs754388: β = 0.13, SE = 0.02, P = 1.4 × 10(-10)). Our results suggest that BMD at different skeletal sites is under a mixture of shared and specific genetic and environmental influences. Allowing for these differences by performing genome-wide association at different skeletal sites may help uncover new genetic influences on BMD.

Learn more

Craniofacial microsomia

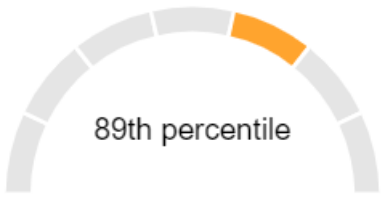


This result is based on **13 genetic variants** associated with **"Craniofacial microsomia"** analyzed in the scientific paper **(02/08/2016 - Zhang YB)**

Skeletal

Your results

Above average genetic predisposition



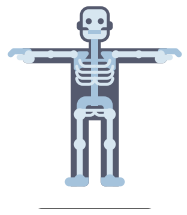
Your genetic predisposition is higher than the 89% average person from your genetic population

Study description

Craniofacial microsomia (CFM) is a rare congenital anomaly that involves immature derivatives from the first and second pharyngeal arches. The genetic pathogenesis of CFM is still unclear. Here we interrogate 0.9 million genetic variants in 939 CFM cases and 2,012 controls from China. After genotyping of an additional 443 cases and 1,669 controls, we identify 8 significantly associated loci with the most significant SNP rs13089920 (logistic regression P=2.15 × 10(-120)) and 5 suggestive loci. The above 13 associated loci, harboured by candidates of ROBO1, GATA3, GBX2, FGF3, NRP2, EDNRB, SHROOM3, SEMA7A, PLCD3, KLF12 and EPAS1, are found to be enriched for genes involved in neural crest cell (NCC) development and vasculogenesis. We then perform whole-genome sequencing on 21 samples from the case cohort, and identify several novel loss-of-function mutations within the associated loci. Our results provide new insights into genetic background of craniofacial microsomia.

Learn more

Osteoarthritis

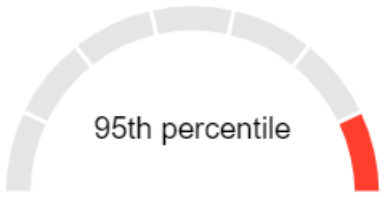


This result is based on **34 genetic variants** associated with **"Osteoarthritis"** analyzed in the scientific paper **(2019 Feb - Tachmazidou I)**

Skeletal

Your results

Very high genetic predisposition



Your genetic predisposition is higher than the 95% average person from your genetic population

Study description

Osteoarthritis is the most common musculoskeletal disease and the leading cause of disability globally. Here, we performed a genome-wide association study for osteoarthritis (77,052 cases and 378,169 controls), analyzing four phenotypes: knee osteoarthritis, hip osteoarthritis, knee and/or hip osteoarthritis, and any osteoarthritis. We discovered 64 signals, 52 of them novel, more than doubling the number of established disease loci. Six signals fine-mapped to a single variant. We identified putative effector genes by integrating expression quantitative trait loci (eQTL) colocalization, fine-mapping, and human rare-disease, animal-model, and osteoarthritis tissue expression data. We found enrichment for genes underlying monogenic forms of bone development diseases, and for the collagen formation and extracellular matrix organization biological pathways. Ten of the likely effector genes, including TGFB1 (transforming growth factor beta 1), FGF18 (fibroblast growth factor 18), CTSK (cathepsin K), and IL11 (interleukin 11), have therapeutics approved or in clinical trials, with mechanisms of action supportive of evaluation for efficacy in osteoarthritis.

Learn more