Health Explorer

DEMO REPORT

Muscular

Muscle fibres are cells that make up the muscular system. Their primary role is to be able to contract. The power to move comes from the muscles linked to the bones or internal organs and blood vessels. Muscle contraction is responsible for the vast majority of movement in the human body. Skeletal, smooth, and cardiac muscles make up the muscular system, an organ system. In addition to facilitating mobility and posture, it also helps transport and distributes blood throughout the body. The muscular system is an intricate web of muscles that play a critical role in human health and function. Everything you do has an impact on your muscles. They regulate your pulse and respiration, aid digestion, and enable mobility. The health of your muscles and the rest of your body is directly linked to regular physical activity and a healthy diet.

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

Find more information at this link

Return to the health dashboard

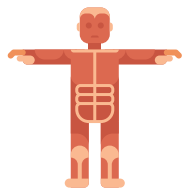
11 entries

Full View

Sort by risk | Sort by name

Polymyositis

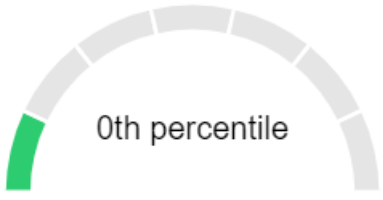
This result is based on **8 genetic variants** associated with **"Polymyositis"** analyzed in the scientific paper (2016 Aug - Rothwell S)



Muscular

Your results

Very low genetic predisposition



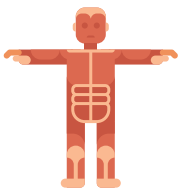
Your genetic predisposition is lower than the 100% average person from your genetic population

Study description

The idiopathic inflammatory myopathies (IIMs) are a heterogeneous group of rare autoimmune diseases characterised by muscle weakness and extramuscular manifestations such as skin rashes and interstitial lung disease. We genotyped 2566 IIM cases of Caucasian descent using the Immunochip; a custom array covering 186 established autoimmune susceptibility loci. The cohort was predominantly comprised of patients with dermatomyositis (DM, n=879), juvenile DM (JDM, n=481), polymyositis (PM, n=931) and inclusion body myositis (n=252) collected from 14 countries through the Myositis Genetics Consortium.

Learn more

Upper eyelid sagging severity

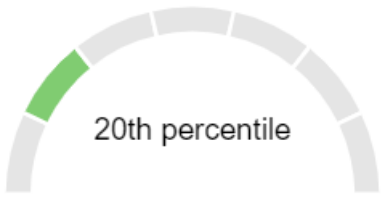


Muscular

This result is based on **22 genetic variants** associated with **"Upper eyelid sagging severity"** analyzed in the scientific paper **(2014 Aug - Jacobs LC)**

Your results

Low genetic predisposition



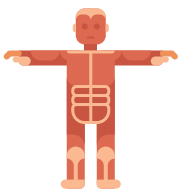
Your genetic predisposition is lower than the 80% average person from your genetic population

Study description

Sagging eyelids, or dermatochalasis, are a frequent concern in older adults. It is considered a feature of skin aging, but risk factors other than aging are largely unknown.

Learn more

Risk of Achilles Tendon Injury

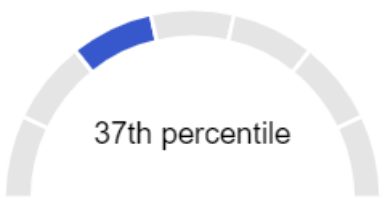


Muscular

This result is based on **5 genetic variants** associated with **"Risk of Achilles Tendon Injury"** analyzed in the scientific paper **(08/01/2021 - Kim SK)**

Your results

Below average genetic predisposition



Your genetic predisposition is lower than the 63% average person from your genetic population

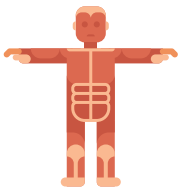
Study description

This study aimed to screen the entire genome for genetic markers associated with risk for Achilles tendon injury.

Learn more

Late-onset myasthenia gravis

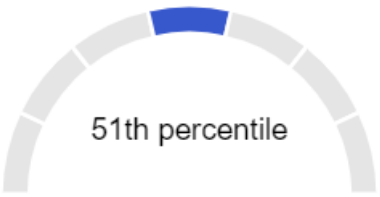
This result is based on **21 genetic variants** associated with **"Late-onset myasthenia gravis"** analyzed in the scientific paper [(2016 Mar - Seldin MF)]



Muscular

Your results

Average genetic predisposition



Your genetic predisposition is higher than the 51% average person from your genetic population

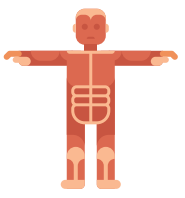
Study description

To investigate the genetics of late-onset myasthenia gravis (LOMG), we conducted a genome-wide association study imputation of>6 million single nucleotide polymorphisms (SNPs) in 532 LOMG cases (anti-acetylcholine receptor [AChR] antibody positive; onset age≥50 years) and 2,128 controls matched for sex and population substructure. The data confirm reported TNFRSF11A associations (rs4574025, P = 3.9 × 10-7, odds ratio [OR] 1.42) and identify a novel candidate gene, ZBTB10, achieving genome-wide significance (rs6998967, P = 8.9 × 10-10, OR 0.53). Several other SNPs showed suggestive significance including rs2476601 (P = 6.5 × 10-6, OR 1.62) encoding the PTPN22 R620W variant noted in early-onset myasthenia gravis (EOMG) and other autoimmune diseases. In contrast, EOMG-associated SNPs in TNIP1 showed no association in LOMG, nor did other loci suggested for EOMG. Many SNPs within the major histocompatibility complex (MHC) region showed strong associations in LOMG, but with smaller effect sizes than in EOMG (highest OR ~2 versus ~6 in EOMG). Moreover, the strongest associations were in opposite directions from EOMG, including an OR of 0.54 for DQA1*05:01 in LOMG (P = 5.9 × 10-12) versus 2.82 in EOMG (P = 3.86 × 10-45). Association and conditioning studies for the MHC region showed three distinct and largely independent association peaks for LOMG corresponding to (a) MHC class II (highest attenuation when conditioning on DQA1), (b) HLA-A and (c) MHC class III SNPs. Conditioning studies of human leukocyte antigen (HLA) amino acid residues also suggest potential functional correlates. Together, these findings emphasize the value of subgrouping myasthenia gravis patients for clinical and basic investigations and imply distinct predisposing mechanisms in LOMG.

Learn more

Tandem gait

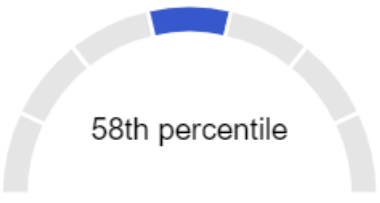
This result is based on **5 genetic variants** associated with **"Tandem gait"** analyzed in the scientific paper [(2016 Jun - Adams HH)]



Muscular

Your results

Average genetic predisposition



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

Study description

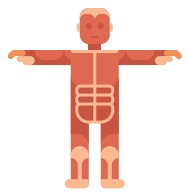
Human gait is a complex neurological and musculoskeletal function, of which the genetic basis remains largely unknown. To determine the influence of common genetic variants on gait parameters, we studied 2,946 participants of the

Rotterdam Study, a population-based cohort of unrelated elderly individuals. We assessed 30 gait parameters using an electronic walkway, which yielded seven independent gait domains after principal component analysis. Genotypes of participants were imputed to the 1,000 Genomes reference panel for generating genetic relationship matrices to estimate heritability of gait parameters, and for subsequent genome-wide association scans (GWASs) to identify specific variants. Gait domains with the highest age- and sex-adjusted heritability were Variability (h (2) = 61%), Rhythm (37%), and Tandem (32%). For other gait domains, heritability estimates attenuated after adjustment for height and weight. Genome-wide association scans identified a variant on 1p22.3 that was significantly associated with single support time, a variable from the Rhythm domain (rs72953990; N = 2,946; β [SE] = 0.0069 (0.0012), p = 2.30×10(-8)). This variant did not replicate in an independent sample (N = 362; p = .78). In conclusion, human gait has highly heritable components that are explained by common genetic variation, which are partly attributed to height and weight. Collaborative efforts are needed to identify robust single variant associations for the heritable parameters.

Learn more

Dupuytren's disease

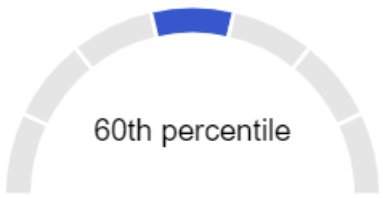
This result is based on **28 genetic variants** associated with **"Dupuytren's disease"** analyzed in the scientific paper **(09/07/2017 - Ng M)**



Muscular

Your results

Average genetic predisposition



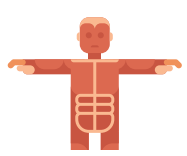
Your genetic predisposition is higher than the 60% average person from your genetic population

Study description

Individuals with Dupuytren disease (DD) are commonly seen by physicians and surgeons across multiple specialties. It is an increasingly common and disabling fibroproliferative disorder of the palmar fascia, which leads to flexion contractures of the digits, and is associated with other tissue-specific fibroses. DD affects between 5% and 25% of people of European descent and is the most common inherited disease of connective tissue. We undertook the largest GWAS to date in individuals with a surgically validated diagnosis of DD from the UK, with replication in British, Dutch, and German individuals. We validated association at all nine previously described signals and discovered 17 additional variants with p ≤ 5 × 10-8. As a proof of principle, we demonstrated correlation of the high-risk genotype at the statistically most strongly associated variant with decreased secretion of the soluble WNT-antagonist SFRP4, in surgical specimen-derived DD myofibroblasts. These results highlight important pathways involved in the pathogenesis of fibrosis, including WNT signaling, extracellular matrix modulation, and inflammation. In addition, many associated loci contain genes that were hitherto unrecognized as playing a role in fibrosis, opening up new avenues of research that may lead to novel treatments for DD and fibrosis more generally. DD represents an ideal human model disease for fibrosis research.

Learn more

Lower body strength

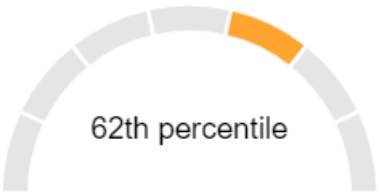


This result is based on **10 genetic variants** associated with **"Lower body strength"** analyzed in the scientific paper [(2016 Oct - Matteini AM)]

Muscular

Your results

Above average genetic predisposition



Your genetic predisposition is higher than the 62% average person from your genetic population

Study description

Decline in muscle strength with aging is an important predictor of health trajectory in the elderly. Several factors, including genetics, are proposed contributors to variability in muscle strength. To identify genetic contributors to muscle strength, a meta-analysis of genomewide association studies of handgrip was conducted. Grip strength was measured using a handheld dynamometer in 27 581 individuals of European descent over 65 years of age from 14 cohort studies. Genomewide association analysis was conducted on ~2.7 million imputed and genotyped variants (SNPs). Replication of the most significant findings was conducted using data from 6393 individuals from three cohorts. GWAS of lower body strength was also characterized in a subset of cohorts. Two genomewide significant (P-value< 5 × 10(-8)) and 39 suggestive (P-value< 5 × 10(-5)) associations were observed from meta-analysis of the discovery cohorts. After meta-analysis with replication cohorts, genomewide significant association was observed for rs752045 on chromosome 8 (β = 0.47, SE = 0.08, P-value = 5.20 × 10(-10)). This SNP is mapped to an intergenic region and is located within an accessible chromatin region (DNase hypersensitivity site) in skeletal muscle myotubes differentiated from the human skeletal muscle myoblasts cell line. This locus alters a binding motif of the CCAAT/enhancer-binding protein-β (CEBPB) that is implicated in muscle repair mechanisms. GWAS of lower body strength did not yield significant results. A common genetic variant in a chromosomal region that regulates myotube differentiation and muscle repair may contribute to variability in grip strength in the elderly. Further studies are needed to uncover the mechanisms that link this genetic variant with muscle strength.

Learn more

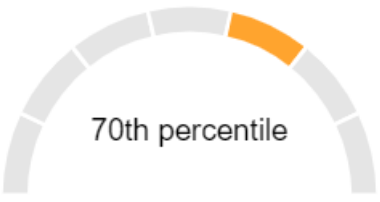
Systemic sclerosis

This result is based on **23 genetic variants** associated with **"Systemic sclerosis"** analyzed in the scientific paper [(10/31/2019 - López-Isac E)]

Muscular

Your results

Above average genetic predisposition



Your genetic predisposition is higher than the 70% average person from your genetic population

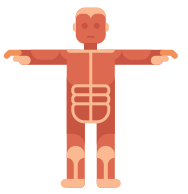
Study description

Systemic sclerosis (SSc) is an autoimmune disease that shows one of the highest mortality rates among rheumatic diseases. We perform a large genome-wide association study (GWAS), and meta-analysis with previous GWASs, in 26,679 individuals and identify 27 independent genome-wide associated signals, including 13 new risk loci. The novel associations nearly double the number of genome-wide hits reported for SSc thus far. We define 95% credible sets of less than 5 likely causal variants in 12 loci. Additionally, we identify specific SSc subtype-associated signals. Functional analysis of high-priority variants shows the potential function of SSc signals, with the identification of 43 robust target

genes through HiChIP. Our results point towards molecular pathways potentially involved in vasculopathy and fibrosis, two main hallmarks in SSc, and highlight the spectrum of critical cell types for the disease. This work supports a better understanding of the genetic basis of SSc and provides directions for future functional experiments.

Learn more

Carpal tunnel syndrome

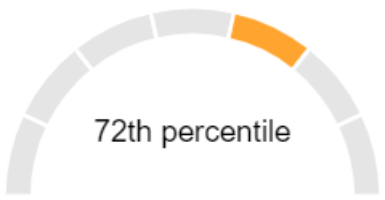


Muscular

This result is based on **16 genetic variants** associated with **"Carpal tunnel syndrome"** analyzed in the scientific paper **(03/04/2019 - Wiberg A)**

Your results

Above average genetic predisposition



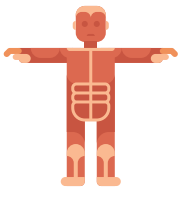
Your genetic predisposition is higher than the 72% average person from your genetic population

Study description

Carpal tunnel syndrome (CTS) is a common and disabling condition of the hand caused by entrapment of the median nerve at the level of the wrist. It is the commonest entrapment neuropathy, with estimates of prevalence ranging between 5-10%. Here, we undertake a genome-wide association study (GWAS) of an entrapment neuropathy, using 12,312 CTS cases and 389,344 controls identified in UK Biobank. We discover 16 susceptibility loci for CTS with $p < 5 \times 10\text{-}8$. We identify likely causal genes in the pathogenesis of CTS, including ADAMTS17, ADAMTS10 and EFEMP1, and using RNA sequencing demonstrate expression of these genes in surgically resected tenosynovium from CTS patients. We perform Mendelian randomisation and demonstrate a causal relationship between short stature and higher risk of CTS. We suggest that variants within genes implicated in growth and extracellular matrix architecture contribute to the genetic predisposition to CTS by altering the environment through which the median nerve transits.

Learn more

Risk of Anterior Cruciate Ligament Rupture

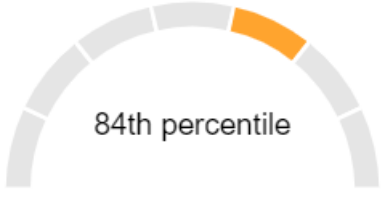


Muscular

This result is based on **3 genetic variants** associated with **"Risk of Anterior Cruciate Ligament Rupture"** analyzed in the scientific paper **(03/30/2017 - Kim SK)**

Your results

Above average genetic predisposition



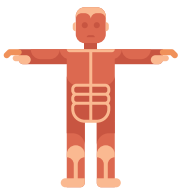
Your genetic predisposition is higher than the 84% average person from your genetic population

Study description

Achilles tendinopathy or rupture and anterior cruciate ligament (ACL) rupture are substantial injuries affecting athletes, associated with delayed recovery or inability to return to competition. To identify genetic markers that might be used to predict risk for these injuries, we performed genome-wide association screens for these injuries using data from the Genetic Epidemiology Research on Adult Health and Aging (GERA) cohort consisting of 102,979 individuals. We did not find any single nucleotide polymorphisms (SNPs) associated with either of these injuries with a p-value that was genome-wide significant (p<5x10-8). We found, however, four and three polymorphisms with p-values that were borderline significant (p<10-6) for Achilles tendon injury and ACL rupture, respectively. We then tested SNPs previously reported to be associated with either Achilles tendon injury or ACL rupture. None showed an association in our cohort with a false discovery rate of less than 5%. We obtained, however, moderate to weak evidence for replication in one case; specifically, rs4919510 in MIR608 had a p-value of 5.1x10-3 for association with Achilles tendon injury, corresponding to a 7% chance of false replication. Finally, we tested 2855 SNPs in 90 candidate genes for musculoskeletal injury, but did not find any that showed a significant association below a false discovery rate of 5%. We provide data containing summary statistics for the entire genome, which will be useful for future genetic studies on these injuries.

Learn more

Appendicular lean mass

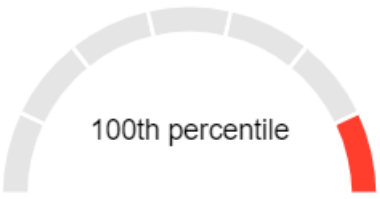


This result is based on **550 genetic variants** associated with **"Appendicular lean mass"** analyzed in the scientific paper **(12/05/2019 - Hernandez Cordero AI)**

Muscular

Your results

Very high genetic predisposition



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

Study description

Muscle bulk in adult healthy humans is highly variable even after height, age, and sex are accounted for. Low muscle mass, due to fewer and/or smaller constituent muscle fibers, would exacerbate the impact of muscle loss occurring in aging or disease. Genetic variability substantially influences muscle mass differences, but causative genes remain largely unknown. In a genome-wide association study (GWAS) on appendicular lean mass (ALM) in a population of 85,750 middle-aged (aged 38-49 years) individuals from the UK Biobank (UKB), we found 182 loci associated with ALM ($p < 5 \times 10\text{-}8$). We replicated associations for 78% of these loci ($p < 5 \times 10\text{-}8$) with ALM in a population of 181,862 elderly (aged 60-74 years) individuals from UKB. We also conducted a GWAS on hindlimb skeletal muscle mass of 1,867 mice from an advanced intercross between two inbred strains (LG/J and SM/J); this GWAS identified 23 quantitative trait loci. Thirty-eight positional candidates distributed across five loci overlapped between the two species. In vitro studies of positional candidates confirmed CPNE1 and STC2 as modifiers of myogenesis. Collectively, these findings shed light on the genetics of muscle mass variability in humans and identify targets for the development of interventions for treatment of muscle loss. The overlapping results between humans and the mouse model GWAS point to shared genetic mechanisms across species.

Learn more