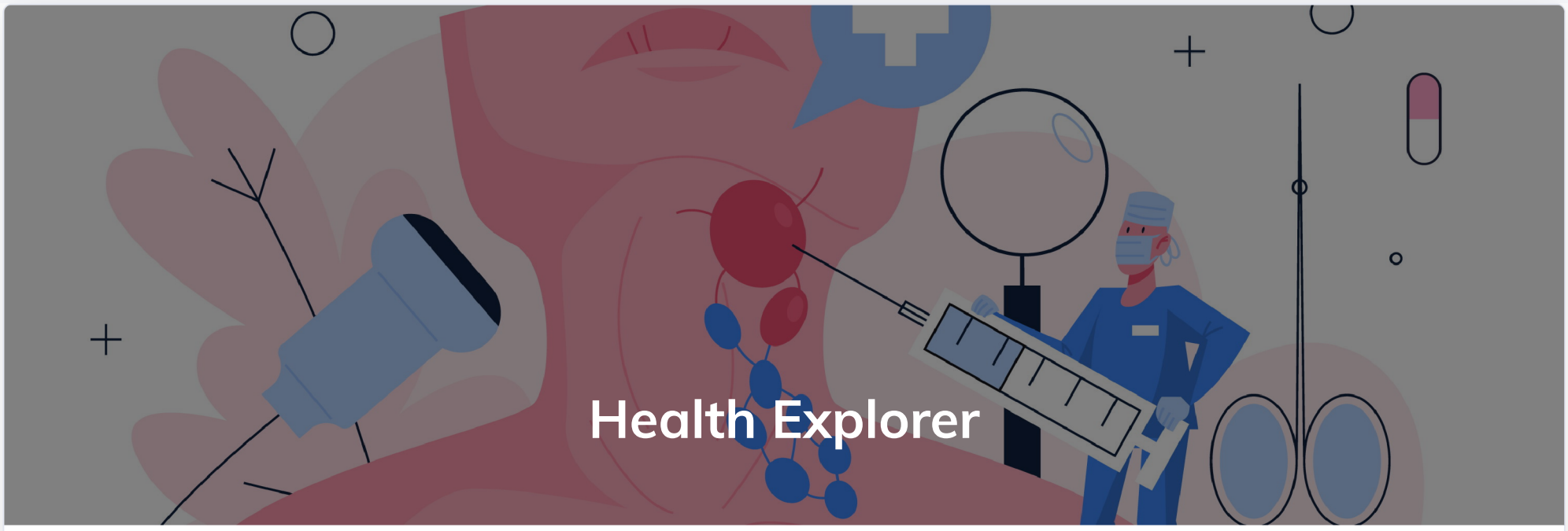


DEMO REPORT

Immune

No one can live without a functioning immune system. Our bodies would be vulnerable if we didn't have an immune system in place. As we float over a sea of germs, our immune system protects us. Cells and tissues in this massive network are always alert for potential threats. Once an adversary is identified, an elaborate assault is launched. The immune system comprises a wide variety of cells, organs, proteins, and tissues found throughout the body. The immune system also recognizes and eliminates damaged or dead cells. Anti-infective measures are taken by the immune system when it confronts an invading pathogen (such as an infectious agent such as a virus or parasite).

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

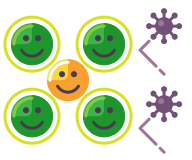
Return to the health dashboard

27 entries

Full View

Sort by risk | Sort by name

Primary biliary cholangitis

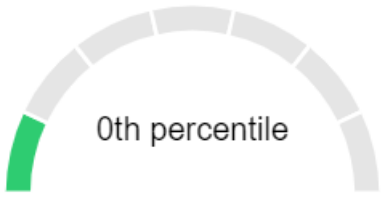


This result is based on **22 genetic variants** associated with **"Primary biliary cholangitis"** analyzed in the scientific paper **(04/20/2017 - Qiu F)**

Immune

Your results

Very low genetic predisposition



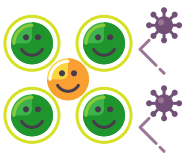
Your genetic predisposition is lower than the 100% average person from your genetic population

Study description

Primary biliary cholangitis (PBC) is an autoimmune liver disease with a strong hereditary component. Here, we report a genome-wide association study that included 1,122 PBC cases and 4,036 controls of Han Chinese descent, with subsequent replication in a separate cohort of 907 PBC cases and 2,127 controls. Our results show genome-wide association of 14 PBC risk loci including previously identified 6p21 (HLA-DRA and DPB1), 17q12 (ORMDL3), 3q13.33 (CD80), 2q32.3 (STAT1/STAT4), 3q25.33 (IL12A), 4q24 (NF-κB) and 22q13.1 (RPL3/SYNGR1). We also identified variants in IL21, IL21R, CD28/CTLA4/ICOS, CD58, ARID3A and IL16 as novel PBC risk loci. These new findings and histochemical studies showing enhanced expression of IL21 and IL21R in PBC livers (particularly in the hepatic portal tracks) support a disease mechanism in which the deregulation of the IL21 signalling pathway, in addition to CD4 T-cell activation and T-cell co-stimulation are critical components in the development of PBC.

Learn more

Primary biliary cirrhosis

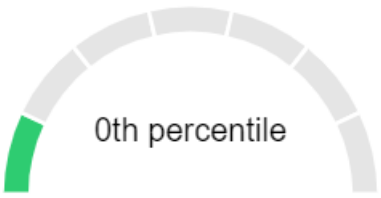


This result is based on **38 genetic variants** associated with **"Primary biliary cirrhosis"** analyzed in the scientific paper [(2012 Oct - Liu JZ)]

Immune

Your results

Very low genetic predisposition



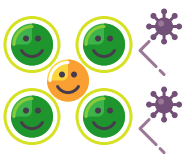
Your genetic predisposition is lower than the 100% average person from your genetic population

Study description

We genotyped 2,861 cases of primary biliary cirrhosis (PBC) from the UK PBC Consortium and 8,514 UK population controls across 196,524 variants within 186 known autoimmune risk loci. We identified 3 loci newly associated with PBC (at P<5×10(-8)), increasing the number of known susceptibility loci to 25. The most associated variant at 19p12 is a low-frequency nonsynonymous SNP in TYK2, further implicating JAK-STAT and cytokine signaling in disease pathogenesis. An additional five loci contained nonsynonymous variants in high linkage disequilibrium (LD; r2>0.8) with the most associated variant at the locus. We found multiple independent common, low-frequency and rare variant association signals at five loci. Of the 26 independent non-human leukocyte antigen (HLA) signals tagged on the Immunochip, 15 have SNPs in B-lymphoblastoid open chromatin regions in high LD (r2>0.8) with the most associated variant. This study shows how data from dense fine-mapping arrays coupled with functional genomic data can be used to identify candidate causal variants for functional follow-up.

Learn more

Acute lymphoblastic leukemia (childhood)

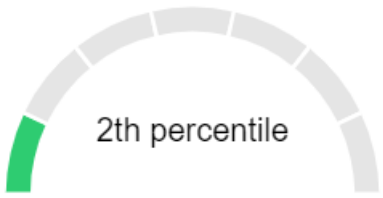


This result is based on **9 genetic variants** associated with **"Acute lymphoblastic leukemia (childhood)"** analyzed in the scientific paper [(01/18/2018 - Wiemels JL)]

Immune

Your results

Very low genetic predisposition



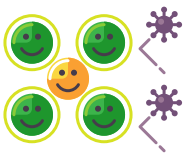
Your genetic predisposition is lower than the 98% average person from your genetic population

Study description

Childhood acute lymphoblastic leukemia (ALL) (age 0-14 years) is 20% more common in Latino Americans than non-Latino whites. We conduct a genome-wide association study in a large sample of 3263 Californian children with ALL (including 1949 of Latino heritage) and 3506 controls matched on month and year of birth, sex, and ethnicity, and an additional 12,471 controls from the Kaiser Resource for Genetic Epidemiology Research on Aging Cohort. Replication of the strongest genetic associations is performed in two independent datasets from the Children's Oncology Group and the California Childhood Leukemia Study. Here we identify new risk loci on 17q12 near IKZF3/ZPBP2/GSDMB/ORMDL3, a locus encompassing a transcription factor important for lymphocyte development (IKZF3), and at an 8q24 region known for structural contacts with the MYC oncogene. These new risk loci may impact gene expression via local (four 17q12 genes) or long-range (8q24) interactions, affecting function of well-characterized hematopoietic and growth-regulation pathways.

Learn more

Ankylosing spondylitis

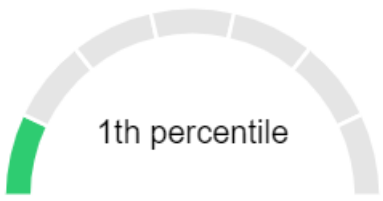


This result is based on **39 genetic variants** associated with **"Ankylosing spondylitis"** analyzed in the scientific paper **(2013 Jul - Cortes A)**

Immune

Your results

Very low genetic predisposition



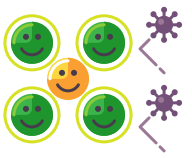
Your genetic predisposition is lower than the 99% average person from your genetic population

Study description

Ankylosing spondylitis is a common, highly heritable inflammatory arthritis affecting primarily the spine and pelvis. In addition to HLA-B*27 alleles, 12 loci have previously been identified that are associated with ankylosing spondylitis in populations of European ancestry, and 2 associated loci have been identified in Asians. In this study, we used the Illumina Immunochip microarray to perform a case-control association study involving 10,619 individuals with ankylosing spondylitis (cases) and 15,145 controls. We identified 13 new risk loci and 12 additional ankylosing spondylitis-associated haplotypes at 11 loci. Two ankylosing spondylitis-associated regions have now been identified encoding four aminopeptidases that are involved in peptide processing before major histocompatibility complex (MHC) class I presentation. Protective variants at two of these loci are associated both with reduced aminopeptidase function and with MHC class I cell surface expression.

Learn more

Pain, multisite chronic

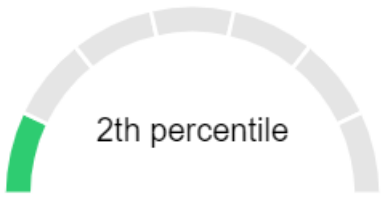


This result is based on **39 genetic variants** associated with **"Pain, multisite chronic"** analyzed in the scientific paper [(06/13/2019 - Johnston KJA)]

Immune

Your results

Very low genetic predisposition



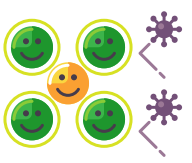
Your genetic predisposition is lower than the 98% average person from your genetic population

Study description

Chronic pain is highly prevalent worldwide and represents a significant socioeconomic and public health burden. Several aspects of chronic pain, for example back pain and a severity-related phenotype 'chronic pain grade', have been shown previously to be complex heritable traits with a polygenic component. Additional pain-related phenotypes capturing aspects of an individual's overall sensitivity to experiencing and reporting chronic pain have also been suggested as a focus for investigation. We made use of a measure of the number of sites of chronic pain in individuals within the UK general population. This measure, termed Multisite Chronic Pain (MCP), is a complex trait and its genetic architecture has not previously been investigated. To address this, we carried out a large-scale genome-wide association study (GWAS) of MCP in ~380,000 UK Biobank participants. Our findings were consistent with MCP having a significant polygenic component, with a Single Nucleotide Polymorphism (SNP) heritability of 10.2%. In total 76 independent lead SNPs at 39 risk loci were associated with MCP. Additional gene-level association analyses identified neurogenesis, synaptic plasticity, nervous system development, cell-cycle progression and apoptosis genes as enriched for genetic association with MCP. Genetic correlations were observed between MCP and a range of psychiatric, autoimmune and anthropometric traits, including major depressive disorder (MDD), asthma and Body Mass Index (BMI). Furthermore, in Mendelian randomisation (MR) analyses a causal effect of MCP on MDD was observed. Additionally, a polygenic risk score (PRS) for MCP was found to significantly predict chronic widespread pain (pain all over the body), indicating the existence of genetic variants contributing to both of these pain phenotypes. Overall, our findings support the proposition that chronic pain involves a strong nervous system component with implications for our understanding of the physiology of chronic pain. These discoveries may also inform the future development of novel treatment approaches.

Learn more

Graves' disease

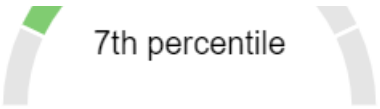


This result is based on **12 genetic variants** associated with **"Graves' disease"** analyzed in the scientific paper [(12/01/2012 - Cooper JD)]

Immune

Your results

Low genetic predisposition



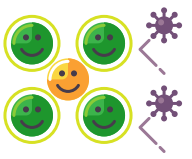
Your genetic predisposition is lower than the 93% average person from your genetic population

Study description

Autoimmune thyroid disease (AITD), including Graves' disease (GD) and Hashimoto's thyroiditis (HT), is one of the most common of the immune-mediated diseases. To further investigate the genetic determinants of AITD, we conducted an association study using a custom-made single-nucleotide polymorphism (SNP) array, the ImmunoChip. The SNP array contains all known and genotype-able SNPs across 186 distinct susceptibility loci associated with one or more immune-mediated diseases. After stringent quality control, we analysed 103 875 common SNPs (minor allele frequency >0.05) in 2285 GD and 462 HT patients and 9364 controls. We found evidence for seven new AITD risk loci ($P < 1.12 \times 10(-6)$; a permutation test derived significance threshold), five at locations previously associated and two at locations awaiting confirmation, with other immune-mediated diseases.

Learn more

Multiple sclerosis

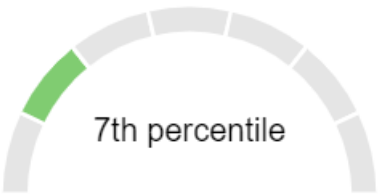


This result is based on **330 genetic variants** associated with **"Multiple sclerosis"** analyzed in the scientific paper (09/27/2019 -)

Immune

Your results

Low genetic predisposition



Your genetic predisposition is lower than the 93% average person from your genetic population

Study description

We analyzed genetic data of 47,429 multiple sclerosis (MS) and 68,374 control subjects and established a reference map of the genetic architecture of MS that includes 200 autosomal susceptibility variants outside the major histocompatibility complex (MHC), one chromosome X variant, and 32 variants within the extended MHC. We used an ensemble of methods to prioritize 551 putative susceptibility genes that implicate multiple innate and adaptive pathways distributed across the cellular components of the immune system. Using expression profiles from purified human microglia, we observed enrichment for MS genes in these brain-resident immune cells, suggesting that these may have a role in targeting an autoimmune process to the central nervous system, although MS is most likely initially triggered by perturbation of peripheral immune responses.

Learn more

Psoriasis

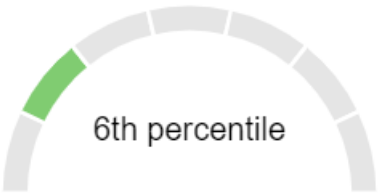
This result is based on **61 genetic variants** associated with **"Psoriasis"** analyzed in the scientific paper

(05/24/2017 - Tsoi LC)

Immune

Your results

Low genetic predisposition



Your genetic predisposition is lower than the 94% average person from your genetic population

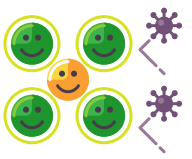
Study description

Psoriasis is a complex disease of skin with a prevalence of about 2%. We conducted the largest meta-analysis of genome-wide association studies (GWAS) for psoriasis to date, including data from eight different Caucasian cohorts, with a combined effective sample size >39,000 individuals. We identified 16 additional psoriasis susceptibility loci achieving genome-wide significance, increasing the number of identified loci to 63 for European-origin individuals. Functional analysis highlighted the roles of interferon signalling and the NFκB cascade, and we showed that the psoriasis signals are enriched in regulatory elements from different T cells (CD8+ T-cells and CD4+ T-cells including TH0, TH1 and TH17). The identified loci explain ~28% of the genetic heritability and generate a discriminatory genetic risk score (AUC=0.76 in our sample) that is significantly correlated with age at onset (p=2 × 10-89). This study provides a comprehensive layout for the genetic architecture of common variants for psoriasis.

Learn more

Diffuse large B cell lymphoma

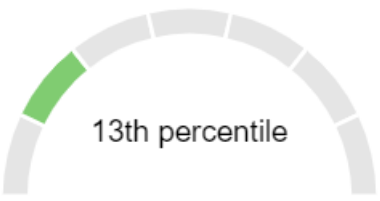
This result is based on **8 genetic variants** associated with **"Diffuse large B cell lymphoma"** analyzed in the scientific paper (2014 Nov - Cerhan JR)



Immune

Your results

Low genetic predisposition



Your genetic predisposition is lower than the 87% average person from your genetic population

Study description

Diffuse large B cell lymphoma (DLBCL) is the most common lymphoma subtype and is clinically aggressive. To identify genetic susceptibility loci for DLBCL, we conducted a meta-analysis of 3 new genome-wide association studies (GWAS) and 1 previous scan, totaling 3,857 cases and 7,666 controls of European ancestry, with additional genotyping of 9 promising SNPs in 1,359 cases and 4,557 controls. In our multi-stage analysis, five independent SNPs in four loci achieved genome-wide significance marked by rs116446171 at 6p25.3 (EXOC2; P = 2.33 × 10(-21)), rs2523607 at 6p21.33 (HLA-B; P = 2.40 × 10(-10)), rs79480871 at 2p23.3 (NCOA1; P = 4.23 × 10(-8)) and two independent SNPs, rs13255292 and rs4733601, at 8q24.21 (PVT1; P = 9.98 × 10(-13) and 3.63 × 10(-11), respectively). These data provide substantial new evidence for genetic susceptibility to this B cell malignancy and point to pathways involved in immune recognition and immune function in the pathogenesis of DLBCL.

Learn more

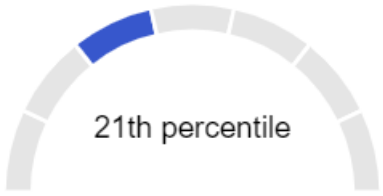
Myeloproliferative neoplasms

This result is based on **17 genetic variants** associated with **"Myeloproliferative neoplasms"** analyzed in the scientific paper [(2020 Oct - Bao EL)]

Immune

Your results

Below average genetic predisposition



Your genetic predisposition is lower than the 79% average person from your genetic population

Study description

Myeloproliferative neoplasms (MPNs) are blood cancers that are characterized by the excessive production of mature myeloid cells and arise from the acquisition of somatic driver mutations in haematopoietic stem cells (HSCs). Epidemiological studies indicate a substantial heritable component of MPNs that is among the highest known for cancers1. However, only a limited number of genetic risk loci have been identified, and the underlying biological mechanisms that lead to the acquisition of MPNs remain unclear. Here, by conducting a large-scale genome-wide association study (3,797 cases and 1,152,977 controls), we identify 17 MPN risk loci ($P < 5.0 \times 10\text{-}8$), 7 of which have not been previously reported. We find that there is a shared genetic architecture between MPN risk and several haematopoietic traits from distinct lineages; that there is an enrichment for MPN risk variants within accessible chromatin of HSCs; and that increased MPN risk is associated with longer telomere length in leukocytes and other clonal haematopoietic states-collectively suggesting that MPN risk is associated with the function and self-renewal of HSCs. We use gene mapping to identify modulators of HSC biology linked to MPN risk, and show through targeted variant-to-function assays that CHEK2 and GFI1B have roles in altering the function of HSCs to confer disease risk. Overall, our results reveal a previously unappreciated mechanism for inherited MPN risk through the modulation of HSC function.

Learn more

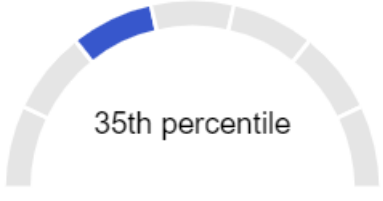
Inflammatory bowel disease

This result is based on **135 genetic variants** associated with **"Inflammatory bowel disease"** analyzed in the scientific paper [(2017 Feb - de Lange KM)]

Immune

Your results

Below average genetic predisposition



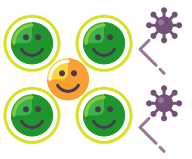
Your genetic predisposition is lower than the 65% average person from your genetic population

Study description

Genetic association studies have identified 215 risk loci for inflammatory bowel disease, thereby uncovering fundamental aspects of its molecular biology. We performed a genome-wide association study of 25,305 individuals and conducted a meta-analysis with published summary statistics, yielding a total sample size of 59,957 subjects. We identified 25 new susceptibility loci, 3 of which contain integrin genes that encode proteins in pathways that have been identified as important therapeutic targets in inflammatory bowel disease. The associated variants are correlated with expression changes in response to immune stimulus at two of these genes (ITGA4 and ITGB8) and at previously implicated loci (ITGAL and ICAM1). In all four cases, the expression-increasing allele also increases disease risk. We also identified likely causal missense variants in a gene implicated in primary immune deficiency, PLCG2, and a negative regulator of inflammation, SLAMF8. Our results demonstrate that new associations at common variants continue to identify genes relevant to therapeutic target identification and prioritization.

Learn more

Alopecia areata

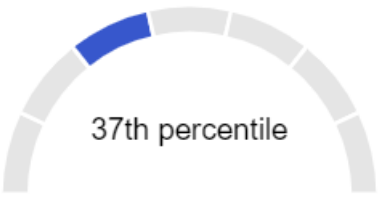


This result is based on **32 genetic variants** associated with **"Alopecia areata"** analyzed in the scientific paper (01/22/2015 - Betz RC)

Immune

Your results

Below average genetic predisposition



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

Study description

Alopecia areata (AA) is a prevalent autoimmune disease with 10 known susceptibility loci. Here we perform the first meta-analysis of research on AA by combining data from two genome-wide association studies (GWAS), and replication with supplemented ImmunoChip data for a total of 3,253 cases and 7,543 controls. The strongest region of association is the major histocompatibility complex, where we fine-map four independent effects, all implicating human leukocyte antigen-DR as a key aetiologic driver. Outside the major histocompatibility complex, we identify two novel loci that exceed the threshold of statistical significance, containing ACOXL/BCL2L11(BIM) (2q13); GARP (LRRC32) (11q13.5), as well as a third nominally significant region SH2B3(LNK)/ATXN2 (12q24.12). Candidate susceptibility gene expression analysis in these regions demonstrates expression in relevant immune cells and the hair follicle. We integrate our results with data from seven other autoimmune diseases and provide insight into the alignment of AA within these disorders. Our findings uncover new molecular pathways disrupted in AA, including autophagy/apoptosis, transforming growth factor beta/Tregs and JAK kinase signalling, and support the causal role of aberrant immune processes in AA.

Learn more

Allergic rhinitis

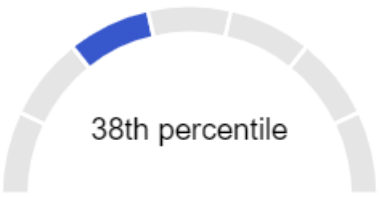
This result is based on **27 genetic variants** associated with **"Allergic rhinitis"** analyzed in the scientific

paper [(12/01/2019 - Johansson Å)]

Immune

Your results

Below average genetic predisposition



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

Study description

Even though heritability estimates suggest that the risk of asthma, hay fever and eczema is largely due to genetic factors, previous studies have not explained a large part of the genetics behind these diseases. In this genome-wide association study, we include 346 545 Caucasians from the UK Biobank to identify novel loci for asthma, hay fever and eczema and replicate novel loci in three independent cohorts. We further investigate if associated lead single nucleotide polymorphisms (SNPs) have a significantly larger effect for one disease compared to the other diseases, to highlight possible disease-specific effects. We identified 141 loci, of which 41 are novel, to be associated ($P \leq 3 \times 10\text{-}8$) with asthma, hay fever or eczema, analyzed separately or as disease phenotypes that includes the presence of different combinations of these diseases. The largest number of loci was associated with the combined phenotype (asthma/hay fever/eczema). However, as many as 20 loci had a significantly larger effect on hay fever/eczema only compared to their effects on asthma, while 26 loci exhibited larger effects on asthma compared with their effects on hay fever/eczema. At four of the novel loci, TNFRSF8, MYRF, TSPAN8, and BHMG1, the lead SNPs were in Linkage Disequilibrium (LD) (>0.8) with potentially casual missense variants. Our study shows that a large amount of the genetic contribution is shared between the diseases. Nonetheless, a number of SNPs have a significantly larger effect on one of the phenotypes, suggesting that part of the genetic contribution is more phenotype specific.

Learn more

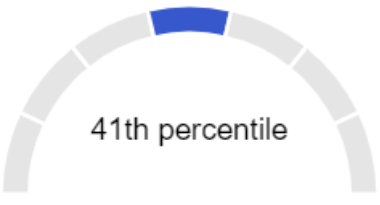
Gout

This result is based on **17 genetic variants** associated with **"Gout"** analyzed in the scientific paper [(2020 May - Nakayama A)]

Immune

Your results

Average genetic predisposition



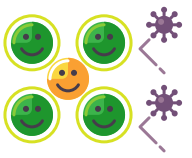
Your genetic predisposition is lower than the 59% average person from your genetic population

Study description

Genome-wide meta-analyses of clinically defined gout were performed to identify subtype-specific susceptibility loci. Evaluation using selection pressure analysis with these loci was also conducted to investigate genetic risks characteristic of the Japanese population over the last 2000-3000 years.

Learn more

Lymphocytic leukemia

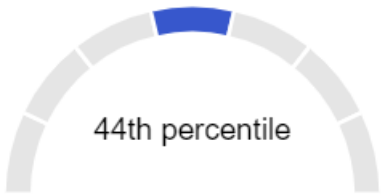


This result is based on **24 genetic variants** associated with **"Lymphocytic leukemia"** analyzed in the scientific paper [(09/04/2020 - Rashkin SR)]

Immune

Your results

Average genetic predisposition



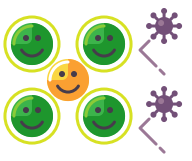
Your genetic predisposition is lower than the 56% average person from your genetic population

Study description

Deciphering the shared genetic basis of distinct cancers has the potential to elucidate carcinogenic mechanisms and inform broadly applicable risk assessment efforts. Here, we undertake genome-wide association studies (GWAS) and comprehensive evaluations of heritability and pleiotropy across 18 cancer types in two large, population-based cohorts: the UK Biobank (408,786 European ancestry individuals; 48,961 cancer cases) and the Kaiser Permanente Genetic Epidemiology Research on Adult Health and Aging cohorts (66,526 European ancestry individuals; 16,001 cancer cases). The GWAS detect 21 genome-wide significant associations independent of previously reported results. Investigations of pleiotropy identify 12 cancer pairs exhibiting either positive or negative genetic correlations; 25 pleiotropic loci; and 100 independent pleiotropic variants, many of which are regulatory elements and/or influence cross-tissue gene expression. Our findings demonstrate widespread pleiotropy and offer further insight into the complex genetic architecture of cross-cancer susceptibility.

Learn more

Multiple myeloma

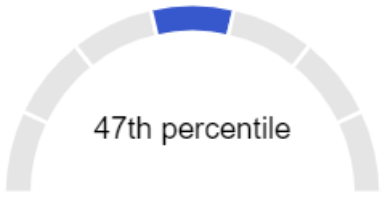


This result is based on **22 genetic variants** associated with **"Multiple myeloma"** analyzed in the scientific paper [(07/01/2016 - Mitchell JS)]

Immune

Your results

Average genetic predisposition



Your genetic predisposition is lower than the 53% average person from your genetic population

Study description

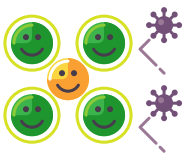
Multiple myeloma (MM) is a plasma cell malignancy with a significant heritable basis. Genome-wide association studies have transformed our understanding of MM predisposition, but individual studies have had limited power to discover risk loci. Here we perform a meta-analysis of these GWAS, add a new GWAS and perform replication analyses resulting in

loci. Here we perform a meta-analysis of these GWAS, add a new GWAS and perform replication analyses resulting in

9,866 cases and 239,188 controls. We confirm all nine known risk loci and discover eight new loci at 6p22.3 (rs34229995, P=1.31 × 10(-8)), 6q21 (rs9372120, P=9.09 × 10(-15)), 7q36.1 (rs7781265, P=9.71 × 10(-9)), 8q24.21 (rs1948915, P=4.20 × 10(-11)), 9p21.3 (rs2811710, P=1.72 × 10(-13)), 10p12.1 (rs2790457, P=1.77 × 10(-8)), 16q23.1 (rs7193541, P=5.00 × 10(-12)) and 20q13.13 (rs6066835, P=1.36 × 10(-13)), which localize in or near to JARID2, ATG5, SMARCD3, CCAT1, CDKN2A, WAC, RFWD3 and PREX1. These findings provide additional support for a polygenic model of MM and insight into the biological basis of tumour development.

Learn more

Chronic lymphocytic leukemia

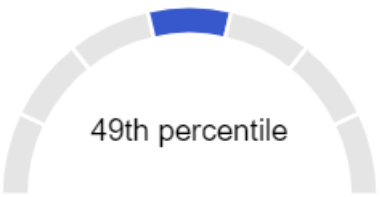


This result is based on **37 genetic variants** associated with **"Chronic lymphocytic leukemia"** analyzed in the scientific paper **(02/06/2017 - Law PJ)**

Immune

Your results

Average genetic predisposition



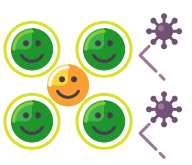
Your genetic predisposition is lower than the 51% average person from your genetic population

Study description

Several chronic lymphocytic leukaemia (CLL) susceptibility loci have been reported; however, much of the heritable risk remains unidentified. Here we perform a meta-analysis of six genome-wide association studies, imputed using a merged reference panel of 1,000 Genomes and UK10K data, totalling 6,200 cases and 17,598 controls after replication. We identify nine risk loci at 1p36.11 (rs34676223, P=5.04 × 10-13), 1q42.13 (rs41271473, P=1.06 × 10-10), 4q24 (rs71597109, P=1.37 × 10-10), 4q35.1 (rs57214277, P=3.69 × 10-8), 6p21.31 (rs3800461, P=1.97 × 10-8), 11q23.2 (rs61904987, P=2.64 × 10-11), 18q21.1 (rs1036935, P=3.27 × 10-8), 19p13.3 (rs7254272, P=4.67 × 10-8) and 22q13.33 (rs140522, P=2.70 × 10-9). These new and established risk loci map to areas of active chromatin and show an over-representation of transcription factor binding for the key determinants of B-cell development and immune response.

Learn more

Crohn's disease

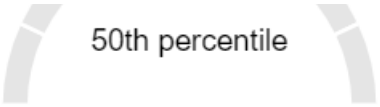


This result is based on **30 genetic variants** associated with **"Crohn's disease"** analyzed in the scientific paper **(11/01/2012 - Jostins L)**

Immune

Your results

Average genetic predisposition



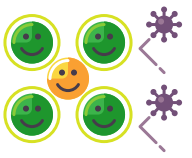
Your genetic predisposition is lower than the 50% average person from your genetic population

Study description

Crohn's disease and ulcerative colitis, the two common forms of inflammatory bowel disease (IBD), affect over 2.5 million people of European ancestry, with rising prevalence in other populations. Genome-wide association studies and subsequent meta-analyses of these two diseases as separate phenotypes have implicated previously unsuspected mechanisms, such as autophagy, in their pathogenesis and showed that some IBD loci are shared with other inflammatory diseases. Here we expand on the knowledge of relevant pathways by undertaking a meta-analysis of Crohn's disease and ulcerative colitis genome-wide association scans, followed by extensive validation of significant findings, with a combined total of more than 75,000 cases and controls. We identify 71 new associations, for a total of 163 IBD loci, that meet genome-wide significance thresholds. Most loci contribute to both phenotypes, and both directional (consistently favouring one allele over the course of human history) and balancing (favouring the retention of both alleles within populations) selection effects are evident. Many IBD loci are also implicated in other immune-mediated disorders, most notably with ankylosing spondylitis and psoriasis. We also observe considerable overlap between susceptibility loci for IBD and mycobacterial infection. Gene co-expression network analysis emphasizes this relationship, with pathways shared between host responses to mycobacteria and those predisposing to IBD.

Learn more

Psoriatic arthritis

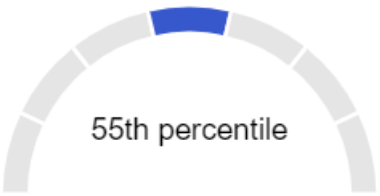


This result is based on **14 genetic variants** associated with **"Psoriatic arthritis"** analyzed in the scientific paper **(12/03/2015 - Stuart PE)**

Immune

Your results

Average genetic predisposition



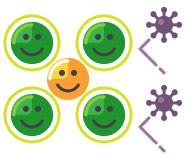
Your genetic predisposition is higher than the 55% average person from your genetic population

Study description

Psoriasis vulgaris (PsV) is a common inflammatory and hyperproliferative skin disease. Up to 30% of people with PsV eventually develop psoriatic arthritis (PsA), an inflammatory musculoskeletal condition. To discern differences in genetic risk factors for PsA and cutaneous-only psoriasis (PsC), we carried out a genome-wide association study (GWAS) of 1,430 PsA case subjects and 1,417 unaffected control subjects. Meta-analysis of this study with three other GWASs and two targeted genotyping studies, encompassing a total of 9,293 PsV case subjects, 3,061 PsA case subjects, 3,110 PsC case subjects, and 13,670 unaffected control subjects of European descent, detected 10 regions associated with PsA and 11 with PsC at genome-wide (GW) significance. Several of these association signals (IFNLR1, IFIH1, NFKBIA for PsA; TNFRSF9, LCE3C/B, TRAF3IP2, IL23A, NFKBIA for PsC) have not previously achieved GW significance. After replication, we also identified a PsV-associated SNP near CDKAL1 (rs4712528, odds ratio [OR] = 1.16, p = 8.4 × 10(-11)). Among identified psoriasis risk variants, three were more strongly associated with PsC than PsA (rs12189871 near HLA-C, p = 5.0 × 10(-19); rs4908742 near TNFRSF9, p = 0.00020; rs10888503 near LCE3A, p = 0.0014), and two were more strongly associated with PsA than PsC (rs12044149 near IL23R, p = 0.00018; rs9321623 near TNFAIP3, p = 0.00022). The PsA-specific variants were independent of previously identified psoriasis variants near IL23R and TNFAIP3. We also found multiple independent susceptibility variants in the IL12B, NOS2, and IFIH1 regions. These results provide insights into the pathogenetic similarities and differences between PsC and PsA.

Learn more

Type 2 diabetes

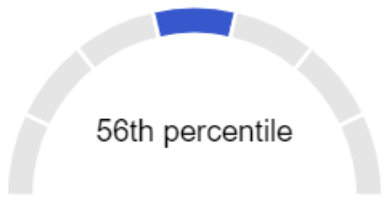


This result is based on **382 genetic variants** associated with **"Type 2 diabetes"** analyzed in the scientific paper [(2018 Nov - Mahajan A)]

Immune

Your results

Average genetic predisposition



Your genetic predisposition is higher than the 56% average person from your genetic population

Study description

We expanded GWAS discovery for type 2 diabetes (T2D) by combining data from 898,130 European-descent individuals (9% cases), after imputation to high-density reference panels. With these data, we (i) extend the inventory of T2D-risk variants (243 loci, 135 newly implicated in T2D predisposition, comprising 403 distinct association signals); (ii) enrich discovery of lower-frequency risk alleles (80 index variants with minor allele frequency <5%, 14 with estimated allelic odds ratio >2); (iii) substantially improve fine-mapping of causal variants (at 51 signals, one variant accounted for >80% posterior probability of association (PPA)); (iv) extend fine-mapping through integration of tissue-specific epigenomic information (islet regulatory annotations extend the number of variants with PPA >80% to 73); (v) highlight validated therapeutic targets (18 genes with associations attributable to coding variants); and (vi) demonstrate enhanced potential for clinical translation (genome-wide chip heritability explains 18% of T2D risk; individuals in the extremes of a T2D polygenic risk score differ more than ninefold in prevalence).

Learn more

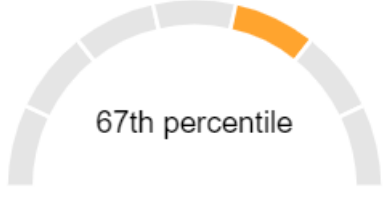
Immunoglobulin light chain (AL) amyloidosis

This result is based on **9 genetic variants** associated with **"Immunoglobulin light chain (AL) amyloidosis"** analyzed in the scientific paper [(2017 Aug - da Silva Filho MI)]

Immune

Your results

Above average genetic predisposition



Your genetic predisposition is higher than the 67% average person from your genetic population

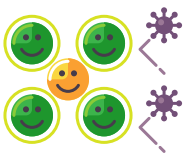
Study description

Immunoglobulin light chain (AL) amyloidosis is characterized by tissue deposition of amyloid fibers derived from immunoglobulin light chain. AL amyloidosis and multiple myeloma (MM) originate from monoclonal gammopathy of undetermined significance. We wanted to characterize germline susceptibility to AL amyloidosis using a genome-wide association study (GWAS) on 1229 AL amyloidosis patients from Germany, UK and Italy, and 7526 healthy local controls. For comparison with MM, recent GWAS data on 3790 cases were used. For AL amyloidosis, single nucleotide polymorphisms (SNPs) at 10 loci showed evidence of an association at P<10-5 with homogeneity of results from the 3 sample sets; some of these were previously documented to influence MM risk, including the SNP at the IRF4 binding site. In AL amyloidosis, rs9344 at the splice site of cyclin D1, promoting translocation (11;14), reached the highest significance, P=7.80 × 10-11; the SNP was only marginally significant in MM. SNP rs79419269 close to gene SMARCD3 involved in chromatin remodeling was also significant (P=5.2 × 10-8). These data provide evidence for common genetic susceptibility to AL amyloidosis and MM. Cyclin D1 is a more prominent driver in AL amyloidosis than in MM, but the links to aggregation of light chains need to be demonstrated.

Learn more

Follicular lymphoma

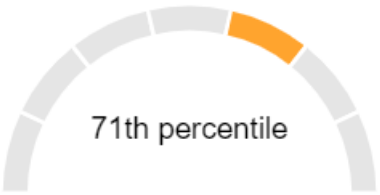
This result is based on **9 genetic variants** associated with **"Follicular lymphoma"** analyzed in the scientific paper **(10/02/2014 - Skibola CF)**



Immune

Your results

Above average genetic predisposition



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

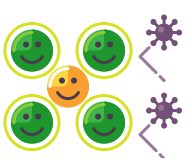
Study description

Genome-wide association studies (GWASs) of follicular lymphoma (FL) have previously identified human leukocyte antigen (HLA) gene variants. To identify additional FL susceptibility loci, we conducted a large-scale two-stage GWAS in 4,523 case subjects and 13,344 control subjects of European ancestry. Five non-HLA loci were associated with FL risk: 11q23.3 (rs4938573, p = 5.79 × 10(-20)) near CXCR5; 11q24.3 (rs4937362, p = 6.76 × 10(-11)) near ETS1; 3q28 (rs6444305, p = 1.10 × 10(-10)) in LPP; 18q21.33 (rs17749561, p = 8.28 × 10(-10)) near BCL2; and 8q24.21 (rs13254990, p = 1.06 × 10(-8)) near PVT1. In an analysis of the HLA region, we identified four linked HLA-DRβ1 multiallelic amino acids at positions 11, 13, 28, and 30 that were associated with FL risk (pomnibus = 4.20 × 10(-67) to 2.67 × 10(-70)). Additional independent signals included rs17203612 in HLA class II (odds ratio [OR(per-allele)] = 1.44; p = 4.59 × 10(-16)) and rs3130437 in HLA class I (OR(per-allele) = 1.23; p = 8.23 × 10(-9)). Our findings further expand the number of loci associated with FL and provide evidence that multiple common variants outside the HLA region make a significant contribution to FL risk.

Learn more

Type 1 diabetes

This result is based on **51 genetic variants** associated with **"Type 1 diabetes"** analyzed in the scientific

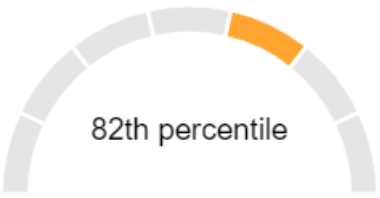


paper [(2015 Apr - Onengut-Gumuscu S)]

Immune

Your results

Above average genetic predisposition



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

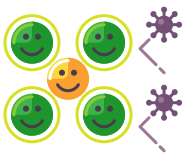
Study description

Genetic studies of type 1 diabetes (T1D) have identified 50 susceptibility regions, finding major pathways contributing to risk, with some loci shared across immune disorders. To make genetic comparisons across autoimmune disorders as informative as possible, a dense genotyping array, the Immunochip, was developed, from which we identified four new T1D-associated regions (P < 5 × 10(-8)). A comparative analysis with 15 immune diseases showed that T1D is more similar genetically to other autoantibody-positive diseases, significantly most similar to juvenile idiopathic arthritis and significantly least similar to ulcerative colitis, and provided support for three additional new T1D risk loci. Using a Bayesian approach, we defined credible sets for the T1D-associated SNPs. The associated SNPs localized to enhancer sequences active in thymus, T and B cells, and CD34(+) stem cells. Enhancer-promoter interactions can now be analyzed in these cell types to identify which particular genes and regulatory sequences are causal.

Learn more

Rheumatoid arthritis

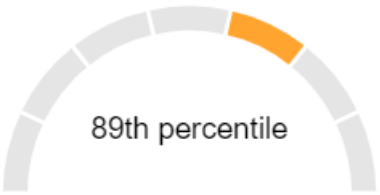
This result is based on **93 genetic variants** associated with **"Rheumatoid arthritis"** analyzed in the scientific paper [(02/20/2014 - Okada Y)]



Immune

Your results

Above average genetic predisposition



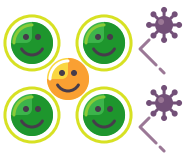
Your genetic predisposition is higher than the 89% average person from your genetic population

Study description

A major challenge in human genetics is to devise a systematic strategy to integrate disease-associated variants with diverse genomic and biological data sets to provide insight into disease pathogenesis and guide drug discovery for complex traits such as rheumatoid arthritis (RA). Here we performed a genome-wide association study meta-analysis in a total of >100,000 subjects of European and Asian ancestries (29,880 RA cases and 73,758 controls), by evaluating ~10 million single-nucleotide polymorphisms. We discovered 42 novel RA risk loci at a genome-wide level of significance, bringing the total to 101 (refs 2 - 4). We devised an in silico pipeline using established bioinformatics methods based on functional annotation, cis-acting expression quantitative trait loci and pathway analyses--as well as novel methods based on genetic overlap with human primary immunodeficiency, haematological cancer somatic mutations and knockout mouse phenotypes--to identify 98 biological candidate genes at these 101 risk loci. We demonstrate that these genes are the targets of approved therapies for RA, and further suggest that drugs approved for other indications may be repurposed for the treatment of RA. Together, this comprehensive genetic study sheds light on fundamental genes, pathways and cell types that contribute to RA pathogenesis, and provides empirical evidence that the genetics of RA can provide important information for drug discovery.

Learn more

Ulcerative colitis

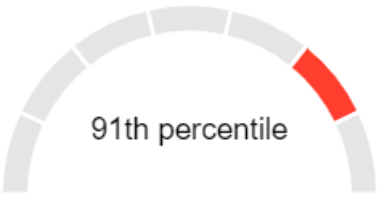


This result is based on **23 genetic variants** associated with **"Ulcerative colitis"** analyzed in the scientific paper **(11/01/2012 - Jostins L)**

Immune

Your results

High genetic predisposition



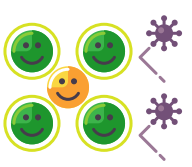
Your genetic predisposition is higher than the 91% average person from your genetic population

Study description

Crohn's disease and ulcerative colitis, the two common forms of inflammatory bowel disease (IBD), affect over 2.5 million people of European ancestry, with rising prevalence in other populations. Genome-wide association studies and subsequent meta-analyses of these two diseases as separate phenotypes have implicated previously unsuspected mechanisms, such as autophagy, in their pathogenesis and showed that some IBD loci are shared with other inflammatory diseases. Here we expand on the knowledge of relevant pathways by undertaking a meta-analysis of Crohn's disease and ulcerative colitis genome-wide association scans, followed by extensive validation of significant findings, with a combined total of more than 75,000 cases and controls. We identify 71 new associations, for a total of 163 IBD loci, that meet genome-wide significance thresholds. Most loci contribute to both phenotypes, and both directional (consistently favouring one allele over the course of human history) and balancing (favouring the retention of both alleles within populations) selection effects are evident. Many IBD loci are also implicated in other immune-mediated disorders, most notably with ankylosing spondylitis and psoriasis. We also observe considerable overlap between susceptibility loci for IBD and mycobacterial infection. Gene co-expression network analysis emphasizes this relationship, with pathways shared between host responses to mycobacteria and those predisposing to IBD.

Learn more

Eosinophil counts

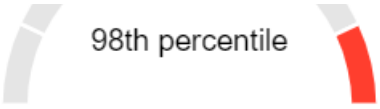


This result is based on **209 genetic variants** associated with **"Eosinophil counts"** analyzed in the scientific paper **(11/17/2016 - Astle WJ)**

Immune

Your results

Very high genetic predisposition



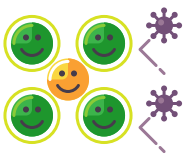
Your genetic predisposition is higher than the 98% average person from your genetic population

Study description

Many common variants have been associated with hematological traits, but identification of causal genes and pathways has proven challenging. We performed a genome-wide association analysis in the UK Biobank and INTERVAL studies, testing 29.5 million genetic variants for association with 36 red cell, white cell, and platelet properties in 173,480 European-ancestry participants. This effort yielded hundreds of low frequency (<5%) and rare (<1%) variants with a strong impact on blood cell phenotypes. Our data highlight general properties of the allelic architecture of complex traits, including the proportion of the heritable component of each blood trait explained by the polygenic signal across different genome regulatory domains. Finally, through Mendelian randomization, we provide evidence of shared genetic pathways linking blood cell indices with complex pathologies, including autoimmune diseases, schizophrenia, and coronary heart disease and evidence suggesting previously reported population associations between blood cell indices and cardiovascular disease may be non-causal.

Learn more

Non-Hodgkin's lymphoma

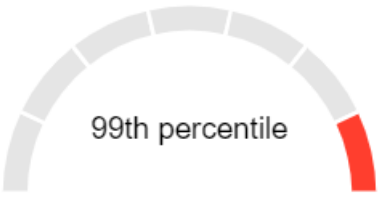


This result is based on **15 genetic variants** associated with **"Non-Hodgkin's lymphoma"** analyzed in the scientific paper [(09/04/2020 - Rashkin SR)]

Immune

Your results

Very high genetic predisposition



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

Study description

Deciphering the shared genetic basis of distinct cancers has the potential to elucidate carcinogenic mechanisms and inform broadly applicable risk assessment efforts. Here, we undertake genome-wide association studies (GWAS) and comprehensive evaluations of heritability and pleiotropy across 18 cancer types in two large, population-based cohorts: the UK Biobank (408,786 European ancestry individuals; 48,961 cancer cases) and the Kaiser Permanente Genetic Epidemiology Research on Adult Health and Aging cohorts (66,526 European ancestry individuals; 16,001 cancer cases). The GWAS detect 21 genome-wide significant associations independent of previously reported results. Investigations of pleiotropy identify 12 cancer pairs exhibiting either positive or negative genetic correlations; 25 pleiotropic loci; and 100 independent pleiotropic variants, many of which are regulatory elements and/or influence cross-tissue gene expression. Our findings demonstrate widespread pleiotropy and offer further insight into the complex genetic architecture of cross-cancer susceptibility.

Learn more