### Tom R Gaunt

### List of Publications by Citations

Source: https://exaly.com/author-pdf/7623815/tom-r-gaunt-publications-by-citations.pdf

Version: 2024-04-25

This document has been generated based on the publications and citations recorded by exaly.com. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

281 65 22,598 147 h-index g-index citations papers 6.21 29,677 8.7 332 L-index avg, IF ext. papers ext. citations

| #   | Paper  | IF   | Citations |
|-----|--|------|-----------|
| 281 | Genetic studies of body mass index yield new insights for obesity biology. <i>Nature</i> , <b>2015</b> , 518, 197-206  | 50.4 | 2687      |
| 280 | Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. <i>Nature</i> , <b>2011</b> , 478, 103-9  | 50.4 | 1564      |
| 279 | The MR-Base platform supports systematic causal inference across the human phenome. <i>ELife</i> , <b>2018</b> , 7,  | 8.9  | 1190      |
| 278 | New genetic loci link adipose and insulin biology to body fat distribution. <i>Nature</i> , <b>2015</b> , 518, 187-196   | 50.4 | 920       |
| 277 | Hardy-Weinberg equilibrium testing of biological ascertainment for Mendelian randomization studies. <i>American Journal of Epidemiology</i> , <b>2009</b> , 169, 505-14  | 3.8  | 777       |
| 276 | The UK10K project identifies rare variants in health and disease. <i>Nature</i> , <b>2015</b> , 526, 82-90   | 50.4 | 776       |
| 275 | Predicting the functional, molecular, and phenotypic consequences of amino acid substitutions using hidden Markov models. <i>Human Mutation</i> , <b>2013</b> , 34, 57-65  | 4.7  | 723       |
| 274 | LD Hub: a centralized database and web interface to perform LD score regression that maximizes the potential of summary level GWAS data for SNP heritability and genetic correlation analysis. <i>Bioinformatics</i> , <b>2017</b> , 33, 272-279 | 7.2  | 541       |
| 273 | Epigenome-wide association study of body mass index, and the adverse outcomes of adiposity. <i>Nature</i> , <b>2017</b> , 541, 81-86   | 50.4 | 511       |
| 272 | DNA Methylation in Newborns and Maternal Smoking in Pregnancy: Genome-wide Consortium Meta-analysis. <i>American Journal of Human Genetics</i> , <b>2016</b> , 98, 680-96  | 11   | 489       |
| 271 | Mendelian randomization of blood lipids for coronary heart disease. <i>European Heart Journal</i> , <b>2015</b> , 36, 539-50   | 9.5  | 417       |
| 270 | Genome-wide association study identifies loci influencing concentrations of liver enzymes in plasma. <i>Nature Genetics</i> , <b>2011</b> , 43, 1131-8   | 36.3 | 415       |
| 269 | HMG-coenzyme A reductase inhibition, type 2 diabetes, and bodyweight: evidence from genetic analysis and randomised trials. <i>Lancet, The</i> , <b>2015</b> , 385, 351-61   | 40   | 409       |
| 268 | Association between alcohol and cardiovascular disease: Mendelian randomisation analysis based on individual participant data. <i>BMJ, The</i> , <b>2014</b> , 349, g4164  | 5.9  | 406       |
| 267 | Metabolite profiling and cardiovascular event risk: a prospective study of 3 population-based cohorts. <i>Circulation</i> , <b>2015</b> , 131, 774-85  | 16.7 | 367       |
| 266 | An integrative approach to predicting the functional effects of non-coding and coding sequence variation. <i>Bioinformatics</i> , <b>2015</b> , 31, 1536-43  | 7.2  | 340       |
| 265 | Systematic identification of genetic influences on methylation across the human life course. <i>Genome Biology</i> , <b>2016</b> , 17, 61  | 18.3 | 331       |

# (2019-2016)

| 264 | Genetic associations at 53 loci highlight cell types and biological pathways relevant for kidney function. <i>Nature Communications</i> , <b>2016</b> , 7, 10023   | 17.4            | 295 |  |
|-----|--|-----------------|-----|--|
| 263 | Epigenome-wide association of DNA methylation markers in peripheral blood from Indian Asians and Europeans with incident type 2 diabetes: a nested case-control study. <i>Lancet Diabetes and Endocrinology,the</i> , <b>2015</b> , 3, 526-534 | 18.1            | 277 |  |
| 262 | C-reactive protein and its role in metabolic syndrome: mendelian randomisation study. <i>Lancet, The</i> , <b>2005</b> , 366, 1954-9   | 40              | 266 |  |
| 261 | Prenatal exposure to maternal smoking and offspring DNA methylation across the lifecourse: findings from the Avon Longitudinal Study of Parents and Children (ALSPAC). <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 2201-17             | 5.6             | 256 |  |
| 260 | Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases: A Mendelian Randomization Study. <i>JAMA Oncology</i> , <b>2017</b> , 3, 636-651   | 13.4            | 236 |  |
| 259 | Large-scale gene-centric meta-analysis across 39 studies identifies type 2 diabetes loci. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 410-25   | 11              | 214 |  |
| 258 | Cubic exact solutions for the estimation of pairwise haplotype frequencies: implications for linkage disequilibrium analyses and a web tool 'CubeX'. <i>BMC Bioinformatics</i> , <b>2007</b> , 8, 428  | 3.6             | 205 |  |
| 257 | Large-scale gene-centric meta-analysis across 32 studies identifies multiple lipid loci. <i>American Journal of Human Genetics</i> , <b>2012</b> , 91, 823-38  | 11              | 189 |  |
| 256 | Improved imputation of low-frequency and rare variants using the UK10K haplotype reference panel. <i>Nature Communications</i> , <b>2015</b> , 6, 8111   | 17.4            | 186 |  |
| 255 | Causal Associations of Adiposity and Body Fat Distribution With Coronary Heart Disease, Stroke Subtypes, and Type 2 Diabetes Mellitus: A Mendelian Randomization Analysis. <i>Circulation</i> , <b>2017</b> , 135, 2373-2388                   | 16.7            | 182 |  |
| 254 | Maternal pre-pregnancy BMI and gestational weight gain, offspring DNA methylation and later offspring adiposity: findings from the Avon Longitudinal Study of Parents and Children.  International Journal of Epidemiology, 2015, 44, 1288-304 | 7.8             | 181 |  |
| 253 | Gene-centric association signals for lipids and apolipoproteins identified via the HumanCVD BeadChip. <i>American Journal of Human Genetics</i> , <b>2009</b> , 85, 628-42   | 11              | 163 |  |
| 252 | Data Resource Profile: Accessible Resource for Integrated Epigenomic Studies (ARIES). <i>International Journal of Epidemiology</i> , <b>2015</b> , 44, 1181-90   | 7.8             | 162 |  |
| 251 | Predicting the functional consequences of cancer-associated amino acid substitutions. <i>Bioinformatics</i> , <b>2013</b> , 29, 1504-10  | 7.2             | 154 |  |
| 250 | Prenatal and early life influences on epigenetic age in children: a study of mother-offspring pairs from two cohort studies. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 191-201   | 5.6             | 153 |  |
| 249 | FATHMM-XF: accurate prediction of pathogenic point mutations via extended features. <i>Bioinformatics</i> , <b>2018</b> , 34, 511-513  | 7.2             | 147 |  |
| 248 | Association of genetic variation with systolic and diastolic blood pressure among African Americans: the Candidate Gene Association Resource study. <i>Human Molecular Genetics</i> , <b>2011</b> , 20, 2273-8                                 | 84 <sup>6</sup> | 146 |  |
| 247 | Identification of new therapeutic targets for osteoarthritis through genome-wide analyses of UK<br>Biobank data. <i>Nature Genetics</i> , <b>2019</b> , 51, 230-236  | 36.3            | 143 |  |

| 246 | Blood pressure loci identified with a gene-centric array. <i>American Journal of Human Genetics</i> , <b>2011</b> , 89, 688-700  | 11   | 137 |
|-----|--|------|-----|
| 245 | Gene-centric meta-analysis in 87,736 individuals of European ancestry identifies multiple blood-pressure-related loci. <i>American Journal of Human Genetics</i> , <b>2014</b> , 94, 349-60  | 11   | 131 |
| 244 | Prenatal exposure to maternal cigarette smoking and DNA methylation: epigenome-wide association in a discovery sample of adolescents and replication in an independent cohort at birth through 17 years of age. <i>Environmental Health Perspectives</i> , <b>2015</b> , 123, 193-9      | 8.4  | 127 |
| 243 | Loci influencing blood pressure identified using a cardiovascular gene-centric array. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 1663-78  | 5.6  | 119 |
| 242 | Apolipoprotein E genotype, cardiovascular biomarkers and risk of stroke: systematic review and meta-analysis of 14,015 stroke cases and pooled analysis of primary biomarker data from up to 60,883 individuals. <i>International Journal of Epidemiology</i> , <b>2013</b> , 42, 475-92 | 7.8  | 113 |
| 241 | Genome-wide meta-analysis uncovers novel loci influencing circulating leptin levels. <i>Nature Communications</i> , <b>2016</b> , 7, 10494   | 17.4 | 107 |
| 240 | Ranking non-synonymous single nucleotide polymorphisms based on disease concepts. <i>Human Genomics</i> , <b>2014</b> , 8, 11  | 6.8  | 105 |
| 239 | Meta-analysis of Dense Genecentric Association Studies Reveals Common and Uncommon Variants Associated with Height. <i>American Journal of Human Genetics</i> , <b>2011</b> , 88, 6-18   | 11   | 103 |
| 238 | Plasma urate concentration and risk of coronary heart disease: a Mendelian randomisation analysis. <i>Lancet Diabetes and Endocrinology,the</i> , <b>2016</b> , 4, 327-36  | 18.1 | 100 |
| 237 | Polymorphisms in the interleukin-4 and interleukin-4 receptor alpha chain genes confer susceptibility to asthma and atopy in a Caucasian population. <i>Clinical and Experimental Allergy</i> , <b>2003</b> , 33, 1111-7   | 4.1  | 98  |
| 236 | Copy number variations and cognitive phenotypes in unselected populations. <i>JAMA - Journal of the American Medical Association</i> , <b>2015</b> , 313, 2044-54  | 27.4 | 96  |
| 235 | Positive associations between single nucleotide polymorphisms in the IGF2 gene region and body mass index in adult males. <i>Human Molecular Genetics</i> , <b>2001</b> , 10, 1491-501   | 5.6  | 93  |
| 234 | Secretory phospholipase A(2)-IIA and cardiovascular disease: a mendelian randomization study.<br>Journal of the American College of Cardiology, <b>2013</b> , 62, 1966-1976  | 15.1 | 91  |
| 233 | Longitudinal analysis of DNA methylation associated with birth weight and gestational age. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 3752-63   | 5.6  | 91  |
| 232 | MIDAS: software for analysis and visualisation of interallelic disequilibrium between multiallelic markers. <i>BMC Bioinformatics</i> , <b>2006</b> , 7, 227   | 3.6  | 84  |
| 231 | Epigenetic profiling of ADHD symptoms trajectories: a prospective, methylome-wide study. <i>Molecular Psychiatry</i> , <b>2017</b> , 22, 250-256   | 15.1 | 83  |
| 230 | A meta-analysis of gene expression signatures of blood pressure and hypertension. <i>PLoS Genetics</i> , <b>2015</b> , 11, e1005035  | 6    | 83  |
| 229 | The association of C-reactive protein and CRP genotype with coronary heart disease: findings from five studies with 4,610 cases amongst 18,637 participants. <i>PLoS ONE</i> , <b>2008</b> , 3, e3011  | 3.7  | 79  |

| 228 | Meta-analysis of Dense Genecentric Association Studies Reveals Common and Uncommon Variants Associated with Height. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 1116-1117  | 11      | 78   |
|-----|--|---------|------|
| 227 | MR-Base: a platform for systematic causal inference across the phenome using billions of genetic associated as | ciation | s 77 |
| 226 | Sixty-five common genetic variants and prediction of type 2 diabetes. <i>Diabetes</i> , <b>2015</b> , 64, 1830-40  | 0.9     | 76   |
| 225 | DNA Methylation and BMI: Investigating Identified Methylation Sites at HIF3A in a Causal Framework. <i>Diabetes</i> , <b>2016</b> , 65, 1231-44  | 0.9     | 76   |
| 224 | Age-related DNA methylation changes are tissue-specific with ELOVL2 promoter methylation as exception. <i>Epigenetics and Chromatin</i> , <b>2018</b> , 11, 25   | 5.8     | 76   |
| 223 | Psychosocial adversity and socioeconomic position during childhood and epigenetic age: analysis of two prospective cohort studies. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 1301-1308   | 5.6     | 75   |
| 222 | Phenome-wide Mendelian randomization mapping the influence of the plasma proteome on complex diseases. <i>Nature Genetics</i> , <b>2020</b> , 52, 1122-1131  | 36.3    | 75   |
| 221 | Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 865-884  | 11      | 74   |
| 220 | Gene-centric meta-analyses of 108 912 individuals confirm known body mass index loci and reveal three novel signals. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 184-201   | 5.6     | 73   |
| 219 | TAS2R38 (phenylthiocarbamide) haplotypes, coronary heart disease traits, and eating behavior in the British Women's Heart and Health Study. <i>American Journal of Clinical Nutrition</i> , <b>2005</b> , 81, 1005-11  | 7       | 73   |
| 218 | DNA methylation and substance-use risk: a prospective, genome-wide study spanning gestation to adolescence. <i>Translational Psychiatry</i> , <b>2016</b> , 6, e976  | 8.6     | 73   |
| 217 | Software Application Profile: PHESANT: a tool for performing automated phenome scans in UK Biobank. <i>International Journal of Epidemiology</i> , <b>2018</b> , 47, 29-35   | 7.8     | 70   |
| 216 | Causal Effect of Plasminogen Activator Inhibitor Type 1 on Coronary Heart Disease. <i>Journal of the American Heart Association</i> , <b>2017</b> , 6,   | 6       | 65   |
| 215 | Adult height, coronary heart disease and stroke: a multi-locus Mendelian randomization meta-analysis. <i>International Journal of Epidemiology</i> , <b>2016</b> , 45, 1927-1937   | 7.8     | 65   |
| 214 | Cohort Profile: Pregnancy And Childhood Epigenetics (PACE) Consortium. <i>International Journal of Epidemiology</i> , <b>2018</b> , 47, 22-23u   | 7.8     | 62   |
| 213 | An epigenome-wide association meta-analysis of prenatal maternal stress in neonates: A model approach for replication. <i>Epigenetics</i> , <b>2016</b> , 11, 140-9  | 5.7     | 62   |
| 212 | The MRC IEU OpenGWAS data infrastructure   |         | 61   |
| 211 | The epigenetic clock and physical development during childhood and adolescence: longitudinal analysis from a UK birth cohort. <i>International Journal of Epidemiology</i> , <b>2017</b> , 46, 549-558   | 7.8     | 60   |

| 210 | Mendelian randomization studies do not support a role for raised circulating triglyceride levels influencing type 2 diabetes, glucose levels, or insulin resistance. <i>Diabetes</i> , <b>2011</b> , 60, 1008-18  | 0.9                | 60 |
|-----|---|--------------------|----|
| 209 | Molecular genetics of human growth hormone, insulin-like growth factors and their pathways in common disease. <i>Human Genetics</i> , <b>2007</b> , 122, 1-21   | 6.3                | 57 |
| 208 | The association of the PON1 Q192R polymorphism with coronary heart disease: findings from the British Women's Heart and Health cohort study and a meta-analysis. <i>BMC Genetics</i> , <b>2004</b> , 5, 17  | 2.6                | 57 |
| 207 | The effects of height and BMI on prostate cancer incidence and mortality: a Mendelian randomization study in 20,848 cases and 20,214 controls from the PRACTICAL consortium. <i>Cancer Causes and Control</i> , <b>2015</b> , 26, 1603-16   | 2.8                | 56 |
| 206 | Whole-genome sequence-based analysis of thyroid function. <i>Nature Communications</i> , <b>2015</b> , 6, 5681  | 17.4               | 56 |
| 205 | The relationship between plasma angiopoietin-like protein 4 levels, angiopoietin-like protein 4 genotype, and coronary heart disease risk. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , <b>2010</b> , 30, 2277-82  | 9.4                | 53 |
| 204 | A pathway-based data integration framework for prediction of disease progression. <i>Bioinformatics</i> , <b>2014</b> , 30, 838-45  | 7.2                | 52 |
| 203 | Does milk intake promote prostate cancer initiation or progression via effects on insulin-like growth factors (IGFs)? A systematic review and meta-analysis. <i>Cancer Causes and Control</i> , <b>2017</b> , 28, 497-  | ·5 <mark>28</mark> | 51 |
| 202 | Prenatal unhealthy diet, insulin-like growth factor 2 gene (IGF2) methylation, and attention deficit hyperactivity disorder symptoms in youth with early-onset conduct problems. <i>Journal of Child Psychology and Psychiatry and Allied Disciplines</i> , <b>2017</b> , 58, 19-27 | 7.9                | 51 |
| 201 | Discovery and refinement of genetic loci associated with cardiometabolic risk using dense imputation maps. <i>Nature Genetics</i> , <b>2016</b> , 48, 1303-1312   | 36.3               | 51 |
| 200 | Haplotypic analyses of the IGF2-INS-TH gene cluster in relation to cardiovascular risk traits. <i>Human Molecular Genetics</i> , <b>2004</b> , 13, 715-25   | 5.6                | 49 |
| 199 | A rare variant in APOC3 is associated with plasma triglyceride and VLDL levels in Europeans. <i>Nature Communications</i> , <b>2014</b> , 5, 4871   | 17.4               | 46 |
| 198 | Variants in the human insulin gene that affect pre-mRNA splicing: is -23HphI a functional single nucleotide polymorphism at IDDM2?. <i>Diabetes</i> , <b>2006</b> , 55, 260-4   | 0.9                | 45 |
| 197 | Mendelian Randomization Analysis Identifies CpG Sites as Putative Mediators for Genetic Influences on Cardiovascular Disease Risk. <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 590-602   | 11                 | 44 |
| 196 | Epigenome-wide association study of asthma and wheeze in childhood and adolescence. <i>Clinical Epigenetics</i> , <b>2017</b> , 9, 112  | 7.7                | 43 |
| 195 | Variation in DNA methylation of the oxytocin receptor gene predicts children's resilience to prenatal stress. <i>Development and Psychopathology</i> , <b>2017</b> , 29, 1663-1674  | 4.3                | 42 |
| 194 | TCTEX1D2 mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. <i>Nature Communications</i> , <b>2015</b> , 6, 7074   | 17.4               | 41 |
| 193 | Searching for the causal effects of body mass index in over 300 000 participants in UK Biobank, using Mendelian randomization. <i>PLoS Genetics</i> , <b>2019</b> , 15, e1007951  | 6                  | 40 |

| 192 | Hypertensive Disorders of Pregnancy and DNA Methylation in Newborns. <i>Hypertension</i> , <b>2019</b> , 74, 375-3   | <b>83</b> .5        | 40 |
|-----|--|---------------------|----|
| 191 | Systematic Mendelian randomization framework elucidates hundreds of CpG sites which may mediate the influence of genetic variants on disease. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 3293-3304            | 5.6                 | 40 |
| 190 | Mosaic structural variation in children with developmental disorders. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 2733-45  | 5.6                 | 39 |
| 189 | Liver Function and Risk of Type 2 Diabetes: Bidirectional Mendelian Randomization Study. <i>Diabetes</i> , <b>2019</b> , 68, 1681-1691   | 0.9                 | 36 |
| 188 | Gene-centric analysis identifies variants associated with interleukin-6 levels and shared pathways with other inflammation markers. <i>Circulation: Cardiovascular Genetics</i> , <b>2013</b> , 6, 163-70              |                     | 34 |
| 187 | Automating Mendelian randomization through machine learning to construct a putative causal map of the human phenome  |                     | 34 |
| 186 | Population genomics of cardiometabolic traits: design of the University College London-London School of Hygiene and Tropical Medicine-Edinburgh-Bristol (UCLEB) Consortium. <i>PLoS ONE</i> , <b>2013</b> , 8, e7      | 13:45               | 33 |
| 185 | The long-term impact of folic acid in pregnancy on offspring DNA methylation: follow-up of the Aberdeen Folic Acid Supplementation Trial (AFAST). <i>International Journal of Epidemiology</i> , <b>2018</b> , 47, 928 | 8- <del>7</del> 9§7 | 32 |
| 184 | Rare Variant Analysis of Human and Rodent Obesity Genes in Individuals with Severe Childhood Obesity. <i>Scientific Reports</i> , <b>2017</b> , 7, 4394  | 4.9                 | 31 |
| 183 | Influence of common genetic variation on blood lipid levels, cardiovascular risk, and coronary events in two British prospective cohort studies. <i>European Heart Journal</i> , <b>2013</b> , 34, 972-81              | 9.5                 | 28 |
| 182 | Appraising the causal relevance of DNA methylation for risk of lung cancer. <i>International Journal of Epidemiology</i> , <b>2019</b> , 48, 1493-1504   | 7.8                 | 27 |
| 181 | Variants of ADRA2A are associated with fasting glucose, blood pressure, body mass index and type 2 diabetes risk: meta-analysis of four prospective studies. <i>Diabetologia</i> , <b>2011</b> , 54, 1710-9            | 10.3                | 27 |
| 180 | IGF2BP1, IGF2BP2 and IGF2BP3 genotype, haplotype and genetic model studies in metabolic syndrome traits and diabetes. <i>Growth Hormone and IGF Research</i> , <b>2010</b> , 20, 310-8                                 | 2                   | 27 |
| 179 | A transcriptome-wide Mendelian randomization study to uncover tissue-dependent regulatory mechanisms across the human phenome. <i>Nature Communications</i> , <b>2020</b> , 11, 185                                    | 17.4                | 27 |
| 178 | PhenoSpD: an integrated toolkit for phenotypic correlation estimation and multiple testing correction using GWAS summary statistics. <i>GigaScience</i> , <b>2018</b> , 7,   | 7.6                 | 27 |
| 177 | Genomic and phenotypic insights from an atlas of genetic effects on DNA methylation. <i>Nature Genetics</i> , <b>2021</b> , 53, 1311-1321  | 36.3                | 27 |
| 176 | Neonatal DNA methylation and early-onset conduct problems: A genome-wide, prospective study. <i>Development and Psychopathology</i> , <b>2018</b> , 30, 383-397  | 4.3                 | 26 |
| 175 | Using the MR-Base platform to investigate risk factors and drug targets for thousands of phenotypes. <i>Wellcome Open Research</i> , <b>2019</b> , 4, 113  | 4.8                 | 26 |

| 174 | Discovery of rare variants associated with blood pressure regulation through meta-analysis of 1.3 million individuals. <i>Nature Genetics</i> , <b>2020</b> , 52, 1314-1332   | 36.3             | 26     |
|-----|---|------------------|--------|
| 173 | Role of DNA Methylation in Type 2 Diabetes Etiology: Using Genotype as a Causal Anchor. <i>Diabetes</i> , <b>2017</b> , 66, 1713-1722   | 0.9              | 25     |
| 172 | Alcohol dehydrogenase type 1C (ADH1C) variants, alcohol consumption traits, HDL-cholesterol and risk of coronary heart disease in women and men: British Women's Heart and Health Study and Caerphilly cohorts. <i>Atherosclerosis</i> , <b>2008</b> , 196, 871-8 | 3.1              | 25     |
| 171 | Nonsense mutation in coiled-coil domain containing 151 gene (CCDC151) causes primary ciliary dyskinesia. <i>Human Mutation</i> , <b>2014</b> , 35, 1446-8   | 4.7              | 24     |
| 170 | Genetic variants associated with Von Willebrand factor levels in healthy men and women identified using the HumanCVD BeadChip. <i>Annals of Human Genetics</i> , <b>2011</b> , 75, 456-67   | 2.2              | 24     |
| 169 | Late life metabolic syndrome, early growth, and common polymorphism in the growth hormone and placental lactogen gene cluster. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2004</b> , 89, 5569-76  | 5 <sup>5.6</sup> | 24     |
| 168 | Deciphering osteoarthritis genetics across 826,690 individuals from 9 populations. <i>Cell</i> , <b>2021</b> , 184, 4784  | 1- <b>48.1</b> 8 | .e3147 |
| 167 | Exploiting horizontal pleiotropy to search for causal pathways within a Mendelian randomization framework. <i>Nature Communications</i> , <b>2020</b> , 11, 1010  | 17.4             | 23     |
| 166 | Texture classification using feature selection and kernel-based techniques. <i>Soft Computing</i> , <b>2015</b> , 19, 2469-2480   | 3.5              | 23     |
| 165 | The association of the PON1 Q192R polymorphism with complications and outcomes of pregnancy: findings from the British Women's Heart and Health cohort study. <i>Paediatric and Perinatal Epidemiology</i> , <b>2006</b> , 20, 244-50                             | 2.7              | 23     |
| 164 | Angiotensin II type I receptor gene polymorphism: anthropometric and metabolic syndrome traits. <i>Journal of Medical Genetics</i> , <b>2005</b> , 42, 396-401  | 5.8              | 23     |
| 163 | Importance of Genetic Studies in Consanguineous Populations for the Characterization of Novel Human Gene Functions. <i>Annals of Human Genetics</i> , <b>2016</b> , 80, 187-96  | 2.2              | 23     |
| 162 | Gene-centric meta-analyses for central adiposity traits in up to 57 412 individuals of European descent confirm known loci and reveal several novel associations. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 2498-510                                    | 5.6              | 22     |
| 161 | CScape: a tool for predicting oncogenic single-point mutations in the cancer genome. <i>Scientific Reports</i> , <b>2017</b> , 7, 11597   | 4.9              | 22     |
| 160 | Four genetic loci influencing electrocardiographic indices of left ventricular hypertrophy. <i>Circulation: Cardiovascular Genetics</i> , <b>2011</b> , 4, 626-35   |                  | 22     |
| 159 | Manual 768 or 384 well microplate gel 'dry' electrophoresis for PCR checking and SNP genotyping. <i>Nucleic Acids Research</i> , <b>2003</b> , 31, e48  | 20.1             | 22     |
| 158 | Replication of IGF2-INS-TH*5 haplotype effect on obesity in older men and study of related phenotypes. <i>European Journal of Human Genetics</i> , <b>2006</b> , 14, 109-16   | 5.3              | 21     |
| 157 | Phenome-wide Mendelian randomization mapping the influence of the plasma proteome on complex diseases   |                  | 21     |

# (2006-2016)

| 156 | Lipids, obesity and gallbladder disease in women: insights from genetic studies using the cardiovascular gene-centric 50K SNP array. <i>European Journal of Human Genetics</i> , <b>2016</b> , 24, 106-12  | 5.3  | 20 |  |
|-----|--|------|----|--|
| 155 | Linkage analysis of the 5q31-33 candidate region for asthma in 240 UK families. <i>Genes and Immunity</i> , <b>2001</b> , 2, 20-4  | 4.4  | 20 |  |
| 154 | Metabolic Characterization of a Rare Genetic Variation Within APOC3 and Its Lipoprotein Lipase-Independent Effects. <i>Circulation: Cardiovascular Genetics</i> , <b>2016</b> , 9, 231-9   |      | 20 |  |
| 153 | Prioritizing putative influential genes in cardiovascular disease susceptibility by applying tissue-specific Mendelian randomization. <i>Genome Medicine</i> , <b>2019</b> , 11, 6   | 14.4 | 19 |  |
| 152 | An integrative approach to predicting the functional effects of small indels in non-coding regions of the human genome. <i>BMC Bioinformatics</i> , <b>2017</b> , 18, 442  | 3.6  | 19 |  |
| 151 | Inflammation-related epigenetic risk and child and adolescent mental health: A prospective study from pregnancy to middle adolescence. <i>Development and Psychopathology</i> , <b>2018</b> , 30, 1145-1156  | 4.3  | 19 |  |
| 150 | Influence of adiposity-related genetic markers in a population of saudi arabians where other variables influencing obesity may be reduced. <i>Disease Markers</i> , <b>2014</b> , 2014, 758232   | 3.2  | 19 |  |
| 149 | Canonical correlation analysis for gene-based pleiotropy discovery. <i>PLoS Computational Biology</i> , <b>2014</b> , 10, e1003876   | 5    | 19 |  |
| 148 | Homogeneous assay of rs4343, an ACE I/D proxy, and an analysis in the British Women's Heart and Health Study (BWHHS). <i>Disease Markers</i> , <b>2008</b> , 24, 11-7  | 3.2  | 19 |  |
| 147 | Null mutation in human ciliary neurotrophic factor gene confers higher body mass index in males. <i>European Journal of Human Genetics</i> , <b>2002</b> , 10, 749-52  | 5.3  | 19 |  |
| 146 | SNP genotyping by combination of 192-well MADGE, ARMS and computerized gel image analysis. <i>BioTechniques</i> , <b>2000</b> , 29, 500-4, 505-6   | 2.5  | 19 |  |
| 145 | Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. <i>Nature Genetics</i> , <b>2021</b> , 53, 1636-1648                            | 36.3 | 19 |  |
| 144 | Associations between high blood pressure and DNA methylation. <i>PLoS ONE</i> , <b>2020</b> , 15, e0227728   | 3.7  | 18 |  |
| 143 | From a single whole exome read to notions of clinical screening: primary ciliary dyskinesia and RSPH9 p.Lys268del in the Arabian Peninsula. <i>Annals of Human Genetics</i> , <b>2012</b> , 76, 211-20   | 2.2  | 18 |  |
| 142 | Using the MR-Base platform to investigate risk factors and drug targets for thousands of phenotypes. <i>Wellcome Open Research</i> , <b>2019</b> , 4, 113  | 4.8  | 18 |  |
| 141 | Assessing the role of insulin-like growth factors and binding proteins in prostate cancer using Mendelian randomization: Genetic variants as instruments for circulating levels. <i>International Journal of Cancer</i> , <b>2016</b> , 139, 1520-33 | 7.5  | 18 |  |
| 140 | Association of copy number variation across the genome with neuropsychiatric traits in the general population. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , <b>2018</b> , 177, 489-502                            | 3.5  | 17 |  |
| 139 | Refined association mapping for a quantitative trait: weight in the H19-IGF2-INS-TH region. <i>Annals of Human Genetics</i> , <b>2006</b> , 70, 848-56   | 2.2  | 17 |  |

| 138 | Author response: The MR-Base platform supports systematic causal inference across the human phenome <b>2018</b> ,   |                  | 17 |
|-----|---|------------------|----|
| 137 | Integrating Mendelian randomization and multiple-trait colocalization to uncover cell-specific inflammatory drivers of autoimmune and atopic disease. <i>Human Molecular Genetics</i> , <b>2019</b> , 28, 3293-3300                                       | o <sup>5.6</sup> | 16 |
| 136 | Metabolic Profiling of Adiponectin Levels in Adults: Mendelian Randomization Analysis. <i>Circulation: Cardiovascular Genetics</i> , <b>2017</b> , 10,  |                  | 16 |
| 135 | The association of the paraoxonase (PON1) Q192R polymorphism with depression in older women: findings from the British Women's Heart and Health Study. <i>Journal of Epidemiology and Community Health</i> , <b>2007</b> , 61, 85-7                       | 5.1              | 16 |
| 134 | Replication and Characterization of Association between ABO SNPs and Red Blood Cell Traits by Meta-Analysis in Europeans. <i>PLoS ONE</i> , <b>2016</b> , 11, e0156914  | 3.7              | 16 |
| 133 | A genome-wide association study of mitochondrial DNA copy number in two population-based cohorts. <i>Human Genomics</i> , <b>2019</b> , 13, 6   | 6.8              | 15 |
| 132 | Characterizing the Causal Pathway for Genetic Variants Associated with Neurological Phenotypes Using Human Brain-Derived Proteome Data. <i>American Journal of Human Genetics</i> , <b>2020</b> , 106, 885-892  | 11               | 15 |
| 131 | Genome-wide survey of parent-of-origin effects on DNA methylation identifies candidate imprinted loci in humans. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 2927-2939  | 5.6              | 15 |
| 130 | Molecular and population analysis of natural selection on the human haptoglobin duplication. <i>Annals of Human Genetics</i> , <b>2012</b> , 76, 352-62   | 2.2              | 15 |
| 129 | Complexity of a complex trait locus: HP, HPR, haemoglobin and cholesterol. <i>Gene</i> , <b>2012</b> , 499, 8-13  | 3.8              | 15 |
| 128 | Quantitated transcript haplotypes (QTH) of AGTR1, reduced abundance of mRNA haplotypes containing 1166C (rs5186:A>C), and relevance to metabolic syndrome traits. <i>Human Mutation</i> , <b>2007</b> , 28, 365-73  | 4.7              | 15 |
| 127 | Texture analysis in gel electrophoresis images using an integrative kernel-based approach. <i>Scientific Reports</i> , <b>2016</b> , 6, 19256   | 4.9              | 15 |
| 126 | Epigenome-wide change and variation in DNA methylation in childhood: trajectories from birth to late adolescence. <i>Human Molecular Genetics</i> , <b>2021</b> , 30, 119-134   | 5.6              | 15 |
| 125 | HIPred: an integrative approach to predicting haploinsufficient genes. <i>Bioinformatics</i> , <b>2017</b> , 33, 1751-17  | <del>5</del> 72  | 14 |
| 124 | A gene-centric analysis of activated partial thromboplastin time and activated protein C resistance using the HumanCVD focused genotyping array. <i>European Journal of Human Genetics</i> , <b>2013</b> , 21, 779-83                                     | 5.3              | 14 |
| 123 | Combined analysis of CHRNA5, CHRNA3 and CYP2A6 in relation to adolescent smoking behaviour.<br>Journal of Psychopharmacology, <b>2011</b> , 25, 915-23  | 4.6              | 14 |
| 122 | Sex-associated autosomal DNA methylation differences are wide-spread and stable throughout childho  | od               | 14 |
| 121 | Developing the WCRF International/University of Bristol Methodology for Identifying and Carrying Out Systematic Reviews of Mechanisms of Exposure-Cancer Associations. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2017</b> , 26, 1667-1675 | 4                | 13 |

| 120 | Circulating Fatty Acids and Risk of Coronary Heart Disease and Stroke: Individual Participant Data Meta-Analysis in Up to 16126 Participants. <i>Journal of the American Heart Association</i> , <b>2020</b> , 9, e013131         | 6                  | 13 |
|-----|---|--------------------|----|
| 119 | The EWAS Catalog: a database of epigenome-wide association studies  |                    | 13 |
| 118 | A Methylome-Wide Association Study of Trajectories of Oppositional Defiant Behaviors and Biological Overlap With Attention Deficit Hyperactivity Disorder. <i>Child Development</i> , <b>2018</b> , 89, 1839-18                   | 5 <del>\$</del> .9 | 13 |
| 117 | Low-frequency variation in TP53 has large effects on head circumference and intracranial volume. <i>Nature Communications</i> , <b>2019</b> , 10, 357   | 17.4               | 12 |
| 116 | Smoking, DNA Methylation, and Lung Function: a Mendelian Randomization Analysis to Investigate Causal Pathways. <i>American Journal of Human Genetics</i> , <b>2020</b> , 106, 315-326  | 11                 | 12 |
| 115 | Integration of genetics into a systems model of electrocardiographic traits using HumanCVD BeadChip. <i>Circulation: Cardiovascular Genetics</i> , <b>2012</b> , 5, 630-8   |                    | 12 |
| 114 | Genotype of galectin 2 (LGALS2) is associated with insulin-glucose profile in the British Women's Heart and Health Study. <i>Diabetologia</i> , <b>2006</b> , 49, 673-7   | 10.3               | 12 |
| 113 | Machine learning improves mortality risk prediction after cardiac surgery: Systematic review and meta-analysis. <i>Journal of Thoracic and Cardiovascular Surgery</i> , <b>2020</b> ,   | 1.5                | 12 |
| 112 | A pathway-centric approach to rare variant association analysis. <i>European Journal of Human Genetics</i> , <b>2016</b> , 25, 123-129  | 5.3                | 11 |
| 111 | Maternal eating disorders affect offspring cord blood DNA methylation: a prospective study. <i>Clinical Epigenetics</i> , <b>2017</b> , 9, 120  | 7.7                | 11 |
| 110 | Cardiometabolic phenotypes and mitochondrial DNA copy number in two cohorts of UK women. <i>Mitochondrion</i> , <b>2018</b> , 39, 9-19  | 4.9                | 11 |
| 109 | A study of relationships between single nucleotide polymorphisms from the growth hormone-insulin-like growth factor axis and bone mass: the Hertfordshire cohort study. <i>Journal of Rheumatology</i> , <b>2009</b> , 36, 1520-6 | 4.1                | 11 |
| 108 | A study of TH01 and IGF2-INS-TH haplotypes in relation to smoking initiation in three independent surveys. <i>Pharmacogenetics and Genomics</i> , <b>2006</b> , 16, 15-23   | 1.9                | 11 |
| 107 | Interaction between birthweight and polymorphism in the calcium-sensing receptor gene in determination of adult bone mass: the Hertfordshire cohort study. <i>Journal of Rheumatology</i> , <b>2007</b> , 34, 769-75              | 4.1                | 11 |
| 106 | Can machine learning improve mortality prediction following cardiac surgery?. <i>European Journal of Cardio-thoracic Surgery</i> , <b>2020</b> , 58, 1130-1136  | 3                  | 11 |
| 105 | Mendelian Randomisation study of the influence of eGFR on coronary heart disease. <i>Scientific Reports</i> , <b>2016</b> , 6, 28514  | 4.9                | 11 |
| 104 | MELODI: Mining Enriched Literature Objects to Derive Intermediates. <i>International Journal of Epidemiology</i> , <b>2018</b> ,  | 7.8                | 10 |
| 103 | A multi-cohort study of polymorphisms in the GH/IGF axis and physical capability: the HALCyon programme. <i>PLoS ONE</i> , <b>2012</b> , 7, e29883  | 3.7                | 10 |

| 102 | Identifying drug targets for neurological and psychiatric disease via genetics and the brain transcriptome. <i>PLoS Genetics</i> , <b>2021</b> , 17, e1009224  | 6                 | 10 |
|-----|--|-------------------|----|
| 101 | Haplotype of growth hormone and angiotensin I-converting enzyme genes, serum angiotensin I-converting enzyme and ventricular growth: pathway inference in pharmacogenetics. <i>Pharmacogenetics and Genomics</i> , <b>2007</b> , 17, 291-4 | 1.9               | 9  |
| 100 | Triglyceride-containing lipoprotein sub-fractions and risk of coronary heart disease and stroke: A prospective analysis in 11,560 adults. <i>European Journal of Preventive Cardiology</i> , <b>2020</b> , 27, 1617-1626                   | 3.9               | 9  |
| 99  | DNA methylation derived systemic inflammation indices are associated with head and neck cancer development and survival. <i>Oral Oncology</i> , <b>2018</b> , 85, 87-94  | 4.4               | 9  |
| 98  | Identifying low density lipoprotein cholesterol associated variants in the Annexin A2 (ANXA2) gene. <i>Atherosclerosis</i> , <b>2017</b> , 261, 60-68  | 3.1               | 8  |
| 97  | Leveraging brain cortex-derived molecular data to elucidate epigenetic and transcriptomic drivers of complex traits and disease. <i>Translational Psychiatry</i> , <b>2019</b> , 9, 105  | 8.6               | 8  |
| 96  | Longitudinal analysis strategies for modelling epigenetic trajectories. <i>International Journal of Epidemiology</i> , <b>2018</b> , 47, 516-525   | 7.8               | 8  |
| 95  | Amplification ratio control system for copy number variation genotyping. <i>Nucleic Acids Research</i> , <b>2011</b> , 39, e54   | 20.1              | 8  |
| 94  | An expectation-maximization program for determining allelic spectrum from CNV data (CoNVEM): insights into population allelic architecture and its mutational history. <i>Human Mutation</i> , <b>2010</b> , 31, 414-2                     | 20 <sup>4.7</sup> | 8  |
| 93  | The variant call format provides efficient and robust storage of GWAS summary statistics. <i>Genome Biology</i> , <b>2021</b> , 22, 32   | 18.3              | 8  |
| 92  | Genetic determinants of circulating haptoglobin concentration. Clinica Chimica Acta, 2019, 494, 138-143  | 26.2              | 7  |
| 91  | eNOS and coronary artery disease: publication bias and the eclipse of hypothesis-driven meta-analysis in genetic association studies. <i>Gene</i> , <b>2015</b> , 556, 257-8   | 3.8               | 7  |
| 90  | Gene-centric association signals for haemostasis and thrombosis traits identified with the HumanCVD BeadChip. <i>Thrombosis and Haemostasis</i> , <b>2013</b> , 110, 995-1003  | 7                 | 7  |
| 89  | Incorporating Non-Coding Annotations into Rare Variant Analysis. <i>PLoS ONE</i> , <b>2016</b> , 11, e0154181  | 3.7               | 7  |
| 88  | Cholesteryl ester transfer protein (CETP) as a drug target for cardiovascular disease. <i>Nature Communications</i> , <b>2021</b> , 12, 5640   | 17.4              | 7  |
| 87  | Multi-omics study revealing putative drug targets of COVID-19 severity and other viral infection disease   | es                | 7  |
| 86  | Genomic and phenomic insights from an atlas of genetic effects on DNA methylation  |                   | 7  |
| 85  | Collapsed methylation quantitative trait loci analysis for low frequency and rare variants. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 4339-4349  | 5.6               | 7  |

# (2017-2017)

| 84 | Physical activity phenotyping with activity bigrams, and their association with BMI. <i>International Journal of Epidemiology</i> , <b>2017</b> , 46, 1857-1870   | 7.8                           | 6  |  |
|----|---|-------------------------------|----|--|
| 83 | Identifying Highly Penetrant Disease Causal Mutations Using Next Generation Sequencing: Guide to Whole Process. <i>BioMed Research International</i> , <b>2015</b> , 2015, 923491   | 3                             | 6  |  |
| 82 | Questioning INS VNTR role in obesity and diabetes: subclasses tag IGF2-INS-TH haplotypes; and -23HphI as a STEP (splicing and translational efficiency polymorphism). <i>Physiological Genomics</i> , <b>2006</b> , 28, 113 | 3.6                           | 6  |  |
| 81 | Global Biobank Meta-analysis Initiative: powering genetic discovery across human diseases   |                               | 6  |  |
| 80 | Using genetic variation to disentangle the complex relationship between food intake and health outco  | mes                           | 6  |  |
| 79 | Genome-wide association study of circulating interleukin 6 levels identifies novel loci. <i>Human Molecular Genetics</i> , <b>2021</b> , 30, 393-409  | 5.6                           | 6  |  |
| 78 | Haptoglobin genotype and outcome after aneurysmal subarachnoid haemorrhage. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2020</b> , 91, 305-313  | 5.5                           | 5  |  |
| 77 | Discovery and replication of SNP-SNP interactions for quantitative lipid traits in over 60,000 individuals. <i>BioData Mining</i> , <b>2017</b> , 10, 25  | 4.3                           | 5  |  |
| 76 | Sequential sentinel SNP Regional Association Plots (SSS-RAP): an approach for testing independence of SNP association signals using meta-analysis data. <i>Annals of Human Genetics</i> , <b>2013</b> , 77, 67-79           | 2.2                           | 5  |  |
| 75 | Haptoglobin duplicon, hemoglobin, and vitamin C: analyses in the british women's heart and health study and Caerphilly prospective study. <i>Disease Markers</i> , <b>2014</b> , 2014, 529456                               | 3.2                           | 5  |  |
| 74 | The Effect of Circulating Zinc, Selenium, Copper and Vitamin K on COVID-19 Outcomes: A Mendelian Randomization Study <i>Nutrients</i> , <b>2022</b> , 14,   | 6.7                           | 5  |  |
| 73 | MR-TRYX: A Mendelian randomization framework that exploits horizontal pleiotropy to infer novel causal pathways   |                               | 5  |  |
| 72 | A Protein Domain and Family Based Approach to Rare Variant Association Analysis. <i>PLoS ONE</i> , <b>2016</b> , 11, e0153803   | 3.7                           | 5  |  |
| 71 | EpiGraphDB: a database and data mining platform for health data science. <i>Bioinformatics</i> , <b>2021</b> , 37, 130  | 04 <del>7</del> . <u>1</u> 31 | 15 |  |
| 7° | MELODI Presto: a fast and agile tool to explore semantic triples derived from biomedical literature. <i>Bioinformatics</i> , <b>2021</b> , 37, 583-585  | 7.2                           | 5  |  |
| 69 | Identifying epigenetic biomarkers of established prognostic factors and survival in a clinical cohort of individuals with oropharyngeal cancer. <i>Clinical Epigenetics</i> , <b>2020</b> , 12, 95                          | 7.7                           | 4  |  |
| 68 | CScape-somatic: distinguishing driver and passenger point mutations in the cancer genome. <i>Bioinformatics</i> , <b>2020</b> , 36, 3637-3644   | 7.2                           | 4  |  |
| 67 | HAPRAP: a haplotype-based iterative method for statistical fine mapping using GWAS summary statistics. <i>Bioinformatics</i> , <b>2017</b> , 33, 79-86  | 7.2                           | 4  |  |

| 66 | Functional Analysis of the Coronary Heart Disease Risk Locus on Chromosome 21q22. <i>Disease Markers</i> , <b>2017</b> , 2017, 1096916   | 3.2  | 4 |
|----|--|------|---|
| 65 | Proxy molecular diagnosis from whole-exome sequencing reveals Papillon-Lefevre syndrome caused by a missense mutation in CTSC. <i>PLoS ONE</i> , <b>2015</b> , 10, e0121351  | 3.7  | 4 |
| 64 | Large-Scale Gene-Centric Meta-Analysis across 39 Studies Identifies Type 2 Diabetes Loci. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 753  | 11   | 4 |
| 63 | Genome-wide data-mining of candidate human splice translational efficiency polymorphisms (STEPs) and an online database. <i>PLoS ONE</i> , <b>2010</b> , 5, e13340   | 3.7  | 4 |
| 62 | Linking Physical Activity to Breast Cancer via Sex Hormones, Part 1: The Effect of Physical Activity on Sex Steroid Hormones. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2021</b> ,                                     | 4    | 4 |
| 61 | Linking Physical Activity to Breast Cancer: Text Mining Results and a Protocol for Systematically Reviewing Three Potential Mechanistic Pathways. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2021</b> ,                 | 4    | 4 |
| 60 | Imprinted loci may be more widespread in humans than previously appreciated and enable limited assignment of parental allelic transmissions in unrelated individuals   |      | 4 |
| 59 | The variant call format provides efficient and robust storage of GWAS summary statistics   |      | 4 |
| 58 | Brain expression quantitative trait locus and network analysis reveals downstream effects and putative drivers for brain-related diseases  |      | 4 |
| 57 | The causal effects of serum lipids and apolipoproteins on kidney function: multivariable and bidirectional Mendelian-randomization analyses. <i>International Journal of Epidemiology</i> , <b>2021</b> , 50, 1569-                    | 1379 | 4 |
| 56 | Diagnosis of Coronary Heart Diseases Using Gene Expression Profiling; Stable Coronary Artery Disease, Cardiac Ischemia with and without Myocardial Necrosis. <i>PLoS ONE</i> , <b>2016</b> , 11, e0149475                              | 3.7  | 4 |
| 55 | Navigating sample overlap, winner curse and weak instrument bias in Mendelian randomization studies using the UK Biobank   |      | 4 |
| 54 | MendelVar: gene prioritization at GWAS loci using phenotypic enrichment of Mendelian disease genes. <i>Bioinformatics</i> , <b>2021</b> , 37, 1-8  | 7.2  | 4 |
| 53 | Estimating the Frequency of Single Point Driver Mutations across Common Solid Tumours. <i>Scientific Reports</i> , <b>2019</b> , 9, 13452  | 4.9  | 3 |
| 52 | Haptoglobin genotype and outcome after spontaneous intracerebral haemorrhage. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2020</b> , 91, 298-304   | 5.5  | 3 |
| 51 | GTB - an online genome tolerance browser. <i>BMC Bioinformatics</i> , <b>2017</b> , 18, 20   | 3.6  | 3 |
| 50 | Comment on: Marchand and Polychronakos (2007) Evaluation of polymorphic splicing in the mechanism of the association of the insulin gene with diabetes: Diabetes 56:709-713. <i>Diabetes</i> , <b>2007</b> , 56, e16; author reply e17 | 0.9  | 3 |
| 49 | Rare SLC13A1 variants associate with intervertebral disc disorder highlighting role of sulfate in disc pathology <i>Nature Communications</i> , <b>2022</b> , 13, 634  | 17.4 | 3 |

| 48 | The EWAS Catalog: a database of epigenome-wide association studies <i>Wellcome Open Research</i> , <b>2022</b> , 7, 41  | 4.8  | 3 |
|----|---|------|---|
| 47 | An informatics consult approach for generating clinical evidence for treatment decisions. <i>BMC Medical Informatics and Decision Making</i> , <b>2021</b> , 21, 281  | 3.6  | 3 |
| 46 | Texture Classification Using Kernel-Based Techniques. Lecture Notes in Computer Science, 2013, 427-434  | 10.9 | 3 |
| 45 | PhenoSpD: an integrated toolkit for phenotypic correlation estimation and multiple testing correction using GWAS summary statistics   |      | 3 |
| 44 | Systematic Mendelian randomization framework elucidates hundreds of genetic loci which may influence disease through changes in DNA methylation levels  |      | 3 |
| 43 | Causal epigenome-wide association study identifies CpG sites that influence cardiovascular disease risk   |      | 3 |
| 42 | Triangulating Molecular Evidence to Prioritize Candidate Causal Genes at Established Atopic Dermatitis Loci. <i>Journal of Investigative Dermatology</i> , <b>2021</b> , 141, 2620-2629                     | 4.3  | 3 |
| 41 | Platelet Glycoprotein Ib EChain as a Putative Therapeutic Target for Juvenile Idiopathic Arthritis: A Mendelian Randomization Study. <i>Arthritis and Rheumatology</i> , <b>2021</b> , 73, 693-701          | 9.5  | 3 |
| 40 | Association of physical activity intensity and bout length with mortality: An observational study of 79,503 UK Biobank participants. <i>PLoS Medicine</i> , <b>2021</b> , 18, e1003757                      | 11.6 | 3 |
| 39 | Availability of public databases for triangulation of findings. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2019</b> , 116, 15766-15767                     | 11.5 | 2 |
| 38 | Early life adiposity and telomere length across the life course: a systematic review and meta-analysis. <i>Wellcome Open Research</i> , <b>2017</b> , 2, 118  | 4.8  | 2 |
| 37 | Validation of lipid-related therapeutic targets for coronary heart disease prevention using human genetics. <i>Nature Communications</i> , <b>2021</b> , 12, 6120   | 17.4 | 2 |
| 36 | Linking Physical Activity to Breast Cancer via Sex Steroid Hormones, Part 2: The Effect of Sex Steroid Hormones on Breast Cancer Risk. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2021</b> , | 4    | 2 |
| 35 | LD Hub: a centralized database and web interface to perform LD score regression that maximizes the potential of summary level GWAS data for SNP heritability and genetic correlation analysis               |      | 2 |
| 34 | Epigenome-wide change and variation in DNA methylation from birth to late adolescence   |      | 2 |
| 33 | Identification of new therapeutic targets for osteoarthritis through genome-wide analyses of UK Bioba   | nk   | 2 |
| 32 | A transcriptome-wide Mendelian randomization study to uncover tissue-dependent regulatory mechanisms across the human phenome   |      | 2 |
| 31 | Computational Tools for Causal Inference in Genetics. <i>Cold Spring Harbor Perspectives in Medicine</i> , <b>2021</b> , 11,  | 5.4  | 2 |

| 30 | Opportunities and Challenges in Functional Genomics Research in Osteoporosis: Report From a Workshop Held by the Causes Working Group of the Osteoporosis and Bone Research Academy of the Royal Osteoporosis Society on October 5th 2020. <i>Frontiers in Endocrinology</i> , <b>2020</b> , 11, 630875 | 5.7   | 2 |
|----|---|-------|---|
| 29 | A proteome-wide genetic investigation identifies several SARS-CoV-2-exploited host targets of clinical relevance. <i>ELife</i> , <b>2021</b> , 10,  | 8.9   | 2 |
| 28 | Reply to Janssen et al. Comment on "Sobczyk, M.K.; Gaunt, T.R. The Effect of Circulating Zinc, Selenium, Copper and Vitamin K on COVID-19 Outcomes: A Mendelian Randomization Study. 2022, , 233" <i>Nutrients</i> , <b>2022</b> , 14,  | 6.7   | 2 |
| 27 | Sequential data selection for predicting the pathogenic effects of sequence variation 2015,   |       | 1 |
| 26 | Analysis of potential genomic confounding in genetic association studies and an online genomic confounding browser (GCB). <i>Annals of Human Genetics</i> , <b>2011</b> , 75, 723-31  | 2.2   | 1 |
| 25 | Evaluating and implementing block jackknife resampling Mendelian randomization to mitigate bias induced by overlapping samples  |       | 1 |
| 24 | Separating the direct effects of traits on atherosclerotic cardiovascular disease from those mediated by type 2 diabetes <i>Diabetologia</i> , <b>2022</b> , 1  | 10.3  | 1 |
| 23 | Early life adiposity and telomere length across the life course: a systematic review and meta-analysis. <i>Wellcome Open Research</i> , <b>2017</b> , 2, 118  | 4.8   | 1 |
| 22 | Validation of lipid-related therapeutic targets for coronary heart disease prevention using human gene  | etics | 1 |
| 21 | Identifying epigenetic biomarkers of established prognostic factors and survival in a clinical cohort of individuals with oropharyngeal cancer  |       | 1 |
| 20 | Trans-ethnic Mendelian randomization study reveals causal relationships between cardio-metabolic factors and chronic kidney disease   |       | 1 |
| 19 | Cholesteryl Ester Transfer Protein as a Drug Target for Cardiovascular Disease  |       | 1 |
| 18 | Prediction of driver variants in the cancer genome via machine learning methodologies. <i>Briefings in Bioinformatics</i> , <b>2021</b> , 22,   | 13.4  | 1 |
| 17 | Trans-Ethnic Mendelian Randomization Study Reveals Causal Relationships Between Cardiometabolic Factors and Chronic Kidney Disease. SSRN Electronic Journal,  | 1     | 1 |
| 16 | Trans-ethnic Mendelian-randomization study reveals causal relationships between cardiometabolic factors and chronic kidney disease. <i>International Journal of Epidemiology</i> , <b>2021</b> ,  | 7.8   | 1 |
| 15 | Mendelian Randomization Analysis Reveals a Causal Effect of Urinary Sodium/Urinary Creatinine Ratio on Kidney Function in Europeans. <i>Frontiers in Bioengineering and Biotechnology</i> , <b>2020</b> , 8, 662  | 5.8   | O |
| 14 | Establishing reference intervals for triglyceride-containing lipoprotein subfraction metabolites measured using nuclear magnetic resonance spectroscopy in a UK population. <i>Annals of Clinical Biochemistry</i> , <b>2021</b> , 58, 47-53  | 2.2   | 0 |
| 13 | Using genetic variation to disentangle the complex relationship between food intake and health outcomes. <i>PLoS Genetics</i> , <b>2022</b> , 18, e1010162  | 6     | O |

### LIST OF PUBLICATIONS

| MeltMADGE for mutation scanning of specific genes in population studies. <i>Nature Protocols</i> , <b>2010</b> , 18.8 5, 1800-12  |  |
|---|--|
| Population mutation scanning of human GHR by meltMADGE and identification of a paucimorphic variant. <i>Genetic Testing and Molecular Biomarkers</i> , <b>2011</b> , 15, 855-60 |  |
| Microplate-array diagonal-gel electrophoresis (MADGE) systems for high-throughput electrophoresis. <i>Technical Tips Online</i> , <b>2000</b> , 5, 12-18                        |  |
| Microplate Array Diagonal Gel Electrophoresis for SNP and Microsatellite Genotyping and for Mutation Scanning <b>2004</b> , 836-841   |  |
| Predicting the Pathogenic Impact of Sequence Variation in the Human Genome. <i>Studies in Health Technology and Informatics</i> , <b>2017</b> , 235, 91-95                      |  |
| 6 Associations between high blood pressure and DNA methylation <b>2020</b> , 15, e0227728   |  |
| 5 Associations between high blood pressure and DNA methylation <b>2020</b> , 15, e0227728   |  |
| Associations between high blood pressure and DNA methylation <b>2020</b> , 15, e0227728   |  |
| 3 Associations between high blood pressure and DNA methylation <b>2020</b> , 15, e0227728   |  |
| 2 Associations between high blood pressure and DNA methylation <b>2020</b> , 15, e0227728   |  |
| Associations between high blood pressure and DNA methylation <b>2020</b> , 15, e0227728   |  |