Advancing human genetics research and drug discovery UK Biobank

Nature Genetics 53, 942-948

DOI: 10.1038/s41588-021-00885-0

Citation Report

| # | Article | IF | CITATIONS |
|----|--|------|-----------|
| 1 | Analysis of 200,000 Exome-Sequenced UK Biobank Subjects Implicates Genes Involved in Increased and Decreased Risk of Hypertension. Pulse, 2021, 9, 17-29. | 0.9 | 10 |
| 2 | Whole-exome sequencing reveals a role of HTRA1 and EGFL8 in brain white matter hyperintensities. Brain, 2021, 144, 2670-2682. | 3.7 | 21 |
| 3 | Investigation of Association of Rare, Functional Genetic Variants With Heavy Drinking and Problem Drinking in Exome Sequenced UK Biobank Participants. Alcohol and Alcoholism, 2021, , . | 0.9 | 1 |
| 5 | Rare Missense Functional Variants at <i>COL4A1</i> and <i>COL4A2</i> in Sporadic Intracerebral Hemorrhage. Neurology, 2021, 97, . | 1.5 | 6 |
| 6 | Genetic pleiotropy of <i>ERCC6</i> lossâ€ofâ€function and deleterious missense variants links retinal dystrophy, arrhythmia, and immunodeficiency in diverse ancestries. Human Mutation, 2021, 42, 969-977. | 1.1 | 3 |
| 7 | Fast numerical optimization for genome sequencing data in population biobanks. Bioinformatics, 2021, 37, 4148-4155. | 1.8 | 9 |
| 9 | The power of large-scale exome sequencing. Nature Reviews Genetics, 2021, 22, 549-549. | 7.7 | 6 |
| 11 | Clonal myelopoiesis promotes adverse outcomes in chronic kidney disease. Leukemia, 2022, 36, 507-515. | 3.3 | 49 |
| 12 | Genetic insights into biological mechanisms governing human ovarian ageing. Nature, 2021, 596, 393-397. | 13.7 | 183 |
| 14 | Rare variant contribution to human disease in 281,104 UK Biobank exomes. Nature, 2021, 597, 527-532. | 13.7 | 224 |
| 16 | Phenotypic Expression and Outcomes in Individuals With Rare Genetic Variants of Hypertrophic Cardionyopathy. Journal of the American College of Cardiology, 2021, 78, 1097-1110. | 1.2 | 55 |
| 18 | Coding and Noncoding Variation in <scp><i>LRRK2</i></scp> and Parkinson's Disease Risk. Movement Disorders, 2022, 37, 95-105. | 2.2 | 14 |
| 20 | Clinical translation of hidradenitis suppurativa genetic studies requires global collaboration. British Journal of Dermatology, 2022, 186, 183-185. | 1.4 | 7 |
| 21 | Rare, Damaging DNA Variants in <i>CORIN</i> and Risk of Coronary Artery Disease: Insights From Functional Genomics and Large-Scale Sequencing Analyses. Circulation Genomic and Precision Medicine, 2021, 14, e003399. | 1.6 | 10 |
| 28 | Exome sequencing and analysis of 454,787 UK Biobank participants. Nature, 2021, 599, 628-634. | 13.7 | 377 |
| 29 | Polygenic basis and biomedical consequences of telomere length variation. Nature Genetics, 2021, 53, 1425-1433. | 9.4 | 145 |
| 31 | Gene-level analysis of rare variants in 379,066 whole exome sequences identifies an association of GIGYF1 loss of function with type 2 diabetes. Scientific Reports, 2021, 11, 21565. | 1.6 | 25 |
| 32 | Mutation saturation for fitness effects at human CpG sites. ELife, 2021, 10, . | 2.8 | 23 |

ARTICLE IF CITATIONS # Effect of Loss-of-Function Genetic Variants in <i>PCSK9</i> 4.3 1 34 Impairment, and Hepatobiliary Function. Diabetes Care, 2022, 45, 251-254. Rare coding variants in DNA damage repair genes associated with timing of natural menopause. Human 1.0 Genetics and Genomics Advances, 2022, 3, 100079. Rare coding variants in 35 genes associate with circulating lipid levelsâ€"A multi-ancestry analysis of 40 2.6 24 170,000 exomes. American Journal of Human Genetics, 2022, 109, 81-96. Genetic data sharing and artificial intelligence in the era of personalized medicine based on a crossâ€sectional analysis of the Saudi human genome program. Scientific Reports, 2022, 12, 1405. Evaluation of Evidence for Pathogenicity Demonstrates That <i>BLK</i>, <i>KLF11</i>, and <i>PAX4</i> 42 0.3 27 Should Not Be Included in Diagnostic Testing for MODY. Diabetes, 2022, 71, 1128-1136. Clinical pharmacogenetic analysis in 5,001 individuals with diagnostic Exome Sequencing data. Npj 1.7 Genomic Medicine, 2022, 7, 12. Genetic associations of protein-coding variants in human disease. Nature, 2022, 603, 95-102. 44 13.7 67 Analysis of rare genetic variation underlying cardiometabolic diseases and traits among 200,000 9.4 68 individuals in the UK Biobank. Nature Génetics, 2022, 54, 240-250. 47 Human lifespan is linked to truncating variants in single genes. Nature Aging, 0, , . 5.3 0 Reduced reproductive success is associated with selective constraint on human genes. Nature, 2022, 13.7 29 603, 858-863. The burden of rare protein-truncating genetic variants on human lifespan. Nature Aging, 2022, 2, 49 5.36 289-294. Exome-wide screening identifies novel rare risk variants for major depression disorder. Molecular 4.1 Psychiatry, 2022, 27, 3069-3074. Identification of Rare Loss-of-Function Genetic Variation Regulating Body Fat Distribution. Journal of 51 1.8 12 Clinical Endocrinology and Metabolism, 2022, 107, 1065-1077. Ribosomal protein S6 kinase beta-1 gene variants cause hypertrophic cardiomyopathy. Journal of 1.5 Medical Genetics, 2021, , jmedgenet-2021-107866. Gene-SCOUT: identifying genes with similar continuous trait fingerprints from phenome-wide 54 6.5 3 association analyses. Nucleic Acids Research, 2022, 50, 4289-4301. Genetic and environmental determinants of diastolic heart function., 2022, 1, 361-371. The Value of Rare Genetic Variation in the Prediction of Common Obesity in European Ancestry 56 1.57 Populations. Frontiers in Endocrinology, 2022, 13, 863893. Gene-based whole genome sequencing meta-analysis of 250 circulating proteins in three isolated European populations. Molecular Metabolism, 2022, 61, 101509.

| # | Article | IF | CITATIONS |
|----|--|------|-----------|
| 58 | Opportunities and challenges for the use of common controls in sequencing studies. Nature Reviews Genetics, 2022, 23, 665-679. | 7.7 | 13 |
| 59 | Rare and Common Genetic Variation Underlying the Risk of Hypertrophic Cardiomyopathy in a National Biobank. JAMA Cardiology, 2022, 7, 715. | 3.0 | 22 |
| 60 | Genome-wide association of polygenic risk extremes for Alzheimer's disease in the UK Biobank. Scientific Reports, 2022, 12, 8404. | 1.6 | 27 |
| 63 | A spectrum of recessiveness among Mendelian disease variants in UK Biobank. American Journal of Human Genetics, 2022, 109, 1298-1307. | 2.6 | 26 |
| 64 | Exome-wide analysis of copy number variation shows association of the human leukocyte antigen region with asthma in UK Biobank. BMC Medical Genomics, 2022, 15, . | 0.7 | 6 |
| 68 | Detection and characterization of male sex chromosome abnormalities in the UK Biobank study. Genetics in Medicine, 2022, 24, 1909-1919. | 1.1 | 14 |
| 69 | Open problems in human trait genetics. Genome Biology, 2022, 23, . | 3.8 | 33 |
| 70 | Association of Common and Rare Genetic Variation in the 3â€Hydroxyâ€3â€Methylglutaryl Coenzyme A Reductase Gene and Cataract Risk. Journal of the American Heart Association, 2022, 11, . | 1.6 | 4 |
| 72 | Clinical features of UK Biobank subjects carrying protein-truncating variants in genes implicated in schizophrenia pathogenesis. Psychiatric Genetics, 2022, 32, 156-161. | 0.6 | 3 |
| 77 | Preface to theme issue on pragmatic and virtual trials: Progress and challenges. Contemporary Clinical Trials, 2022, 119, 106816. | 0.8 | 2 |
| 78 | Small dataset solves big problem: An outlier-insensitive binary classifier for inhibitory potency prediction. Knowledge-Based Systems, 2022, 251, 109242. | 4.0 | 10 |
| 80 | Inherited basis of visceral, abdominal subcutaneous and gluteofemoral fat depots. Nature Communications, 2022, 13, . | 5.8 | 43 |
| 81 | Investigating the characteristics of genes and variants associated with self-reported hearing difficulty in older adults in the UK Biobank. BMC Biology, 2022, 20, . | 1.7 | 7 |
| 82 | Cenome-wide analyses of 200,453 individuals yield new insights into the causes and consequences of clonal hematopoiesis. Nature Genetics, 2022, 54, 1155-1166. | 9.4 | 109 |
| 83 | Frequency, Penetrance, and Variable Expressivity of Dilated Cardiomyopathy–Associated Putative Pathogenic Gene Variants in UK Biobank Participants. Circulation, 2022, 146, 110-124. | 1.6 | 25 |
| 85 | New Horizons: the value of UK Biobank to research on endocrine and metabolic disorders. Journal of Clinical Endocrinology and Metabolism, 0, , . | 1.8 | 3 |
| 86 | The sequences of 150,119 genomes in the UK Biobank. Nature, 2022, 607, 732-740. | 13.7 | 173 |
| 87 | Gene-Based Variant Analysis of Whole-Exome Sequencing in Relation to Eosinophil Count. Frontiers in Immunology, 0, 13, . | 2.2 | 3 |

| | | EPORT | |
|-----|--|-------|----------------|
| # | ARTICLE Evaluating the role of rare genetic variation in sleep duration. Sleep Health, 2022, 8, 536-541. | IF | CITATIONS 2 |
| 88 | CNest: A novel copy number association discovery method uncovers 862 new associations from | 1.3 | 2 |
| 89 | 200,629 whole-exome sequence datasets in the UK Biobank. Cell Genomics, 2022, 2, 100167. | 3.0 | 10 |
| 90 | Systematic single-variant and gene-based association testing of thousands of phenotypes in 394,841ÂUK Biobank exomes. Cell Genomics, 2022, 2, 100168. | 3.0 | 89 |
| 91 | Standard operating procedures for biobank in oncology. Frontiers in Molecular Biosciences, 0, 9, . | 1.6 | 13 |
| 92 | Clonal hematopoiesis and risk of prostate cancer in large samples of European ancestry men. Human Molecular Genetics, 2023, 32, 489-495. | 1.4 | 1 |
| 93 | Immunodeficiency, autoimmunity, and increased risk of B cell malignancy in humans with <i>TRAF3</i> mutations. Science Immunology, 2022, 7, . | 5.6 | 9 |
| 94 | A genome sequencing system for universal newborn screening, diagnosis, and precision medicine for severe genetic diseases. American Journal of Human Genetics, 2022, 109, 1605-1619. | 2.6 | 58 |
| 95 | Rare coding variation provides insight into the genetic architecture and phenotypic context of autism. Nature Genetics, 2022, 54, 1320-1331. | 9.4 | 155 |
| 97 | Human genotype-to-phenotype predictions: Boosting accuracy with nonlinear models. PLoS ONE, 2022, 17, e0273293. | 1.1 | 6 |
| 98 | Study of variants in genes implicated in rare familial migraine syndromes and their association with migraine in 200,000 exomeâ€sequenced UK Biobank participants. Annals of Human Genetics, 0, , . | 0.3 | 4 |
| 101 | Multimodal biomedical AI. Nature Medicine, 2022, 28, 1773-1784. | 15.2 | 191 |
| 102 | The impact of Mendelian sleep and circadian genetic variants in a population setting. PLoS Genetics, 2022, 18, e1010356. | 1.5 | 2 |
| 103 | Identifying interpretable gene-biomarker associations with functionally informed kernel-based tests in 190,000 exomes. Nature Communications, 2022, 13, . | 5.8 | 6 |
| 104 | Whole-Exome Sequencing Study of Familial Nasopharyngeal Carcinoma and Its Implication for Identifying High-Risk Individuals. Journal of the National Cancer Institute, 2022, 114, 1689-1697. | 3.0 | 7 |
| 105 | Assessment of genetic susceptibility to multiple primary cancers through whole-exome sequencing in two large multi-ancestry studies. BMC Medicine, 2022, 20, . | 2.3 | 3 |
| 106 | Dispatches from Biotech beginning <scp>BeginNGS</scp> : Rapid newborn genome sequencing to end the diagnostic and therapeutic odyssey. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 243-256. | 0.7 | 12 |
| 107 | Whole Exome Sequencing Study Identifies Novel Rare Risk Variants for Habitual Coffee Consumption Involved in Olfactory Receptor and Hyperphagia. Nutrients, 2022, 14, 4330. | 1.7 | 0 |
| 109 | The contribution of functional HNF1A variants and polygenic susceptibility to risk of type 2 diabetes in ancestrally diverse populations. Diabetologia, 0, , . | 2.9 | 1 |

| # | Article | IF | CITATIONS |
|-----|---|------|-----------|
| 110 | Prevalence and Disease Expression of Pathogenic and Likely Pathogenic Variants Associated With Inherited Cardiomyopathies in the General Population. Circulation Genomic and Precision Medicine, 2022, 15, . | 1.6 | 18 |
| 111 | Genetic map of regional sulcal morphology in the human brain from UK biobank data. Nature Communications, 2022, 13, . | 5.8 | 9 |
| 112 | Mapping the genetic architecture of cortical morphology through neuroimaging: progress and perspectives. Translational Psychiatry, 2022, 12, . | 2.4 | 10 |
| 113 | Gene Sequencing Identifies Perturbation in Nitric Oxide Signaling as a Nonlipid Molecular Subtype of Coronary Artery Disease. Circulation Genomic and Precision Medicine, 2022, 15, . | 1.6 | 4 |
| 114 | Using human genetics to improve safety assessment of therapeutics. Nature Reviews Drug Discovery, 2023, 22, 145-162. | 21.5 | 20 |
| 115 | Framework and baseline examination of the German National Cohort (NAKO). European Journal of Epidemiology, 2022, 37, 1107-1124. | 2.5 | 32 |
| 116 | Influences of rare copy-number variation on human complex traits. Cell, 2022, 185, 4233-4248.e27. | 13.5 | 15 |
| 118 | Reduced penetrance of MODY-associated HNF1A/HNF4A variants but not GCK variants in clinically unselected cohorts. American Journal of Human Genetics, 2022, 109, 2018-2028. | 2.6 | 31 |
| 119 | From Calcium Channels to New Therapeutics. , 2022, , 687-706. | | 0 |
| 120 | Damaging missense variants in IGF1R implicate a role for IGF-1 resistance in the etiology of type 2 diabetes. Cell Genomics, 2022, 2, 100208. | 3.0 | 12 |
| 121 | Identifying shared genetic factors underlying epilepsy and congenital heart disease in Europeans. Human Genetics, 0, , . | 1.8 | 0 |
| 122 | Identification of PCSK9-like human gene knockouts using metabolomics, proteomics, and whole-genome sequencing in a consanguineous population. Cell Genomics, 2023, 3, 100218. | 3.0 | 4 |
| 123 | Novel genotyping algorithms for rare variants significantly improve the accuracy of Applied Biosystemsâ"¢ Axiomâ"¢ array genotyping calls: Retrospective evaluation of UK Biobank array data. PLoS ONE, 2022, 17, e0277680. | 1.1 | 2 |
| 124 | UK Biobank: a globally important resource for cancer research. British Journal of Cancer, 2023, 128, 519-527. | 2.9 | 32 |
| 125 | Whole-exome sequencing study identifies rare variants and genes associated with intraocular pressure and glaucoma. Nature Communications, 2022, 13, . | 5.8 | 6 |
| 126 | Gene-Level Associations in Patients With and Without Pathogenic Germline Variants in <i>CDKN2A</i> and Pancreatic Cancer. JCO Precision Oncology, 2022, , . | 1.5 | 1 |
| 127 | Public awareness and attitudes toward biobank and sample donation: A regional Chinese survey. Frontiers in Public Health, 0, 10, . | 1.3 | 4 |
| 129 | Common and rare variant associations with clonal haematopoiesis phenotypes. Nature, 2022, 612, 301-309. | 13.7 | 74 |

| # 130 | ARTICLE Association of <i>PCSK9</i> Loss-of-Function Variants With Risk of Heart Failure. JAMA Cardiology, 0, , | IF 3.0 | CITATIONS |
|----------|---|-----------|-----------|
| 131 | Clonal Hematopoiesis and Risk of Incident Lung Cancer. Journal of Clinical Oncology, 2023, 41, 1423-1433. | 0.8 | 10 |
| 132 | Efficient identification of traitâ€associated lossâ€ofâ€function variants in the UK Biobank cohort by exomeâ€sequencing based genotype imputation. Genetic Epidemiology, 2023, 47, 121-134. | 0.6 | 3 |
| 133 | Powerful, scalable and resource-efficient meta-analysis of rare variant associations in large whole genome sequencing studies. Nature Genetics, 2023, 55, 154-164. | 9.4 | 12 |
| 134 | Human loss-of-function variants in the serotonin 2C receptor associated with obesity and maladaptive behavior. Nature Medicine, 2022, 28, 2537-2546. | 15.2 | 10 |
| 135 | The collective effects of genetic variants and complex traits. Journal of Human Genetics, 2023, 68, 255-262. | 1.1 | 2 |
| 137 | CanRisk-Prostate: A Comprehensive, Externally Validated Risk Model for the Prediction of Future Prostate Cancer. Journal of Clinical Oncology, 2023, 41, 1092-1104. | 0.8 | 9 |
| 139 | Relating pathogenic loss-of-function mutations in humans to their evolutionary fitness costs. ELife, 0, 12, . | 2.8 | 12 |
| 140 | Harnessing the Power of Electronic Health Records and Genomics for Drug Discovery. Annual Review of Pharmacology and Toxicology, 2023, 63, 65-76. | 4.2 | 4 |
| 141 | Study of effect modifiers of genetically predicted CETP reduction. Genetic Epidemiology, 0, , . | 0.6 | 0 |
| 143 | From genetic variation to precision medicine. , 2023, 1, . | | 2 |
| 144 | LoFTK: a framework for fully automated calculation of predicted Loss-of-Function variants and genes. BioData Mining, 2023, 16, . | 2.2 | 1 |
| 145 | Rare variant analyses in large-scale cohorts identified SLC13A1 associated with chronic pain. Pain, 2023, 164, 1841-1851. | 2.0 | 3 |
| 146 | A Rapid Review on the Value of Biobanks Containing Genetic Information. Value in Health, 2023, , . | 0.1 | 2 |
| 151 | Association of Rare Protein-Truncating DNA Variants in <i>APOB</i> or <i>PCSK9</i> With Low-density Lipoprotein Cholesterol Level and Risk of Coronary Heart Disease. JAMA Cardiology, 2023, 8, 258. | 3.0 | 10 |
| 152 | Genomic analysis of lean individuals with NAFLD identifies monogenic disorders in a prospective cohort study. JHEP Reports, 2023, 5, 100692. | 2.6 | 5 |
| 153 | Atlas of plasma NMR biomarkers for health and disease in 118,461 individuals from the UK Biobank. Nature Communications, 2023, 14, . | 5.8 | 71 |
| 154 | Rare variant aggregation in 148,508 exomes identifies genes associated with proxy dementia. Scientific Reports, 2023, 13, . | 1.6 | 3 |

| # | Article | IF | CITATIONS |
|-----|--|-----|-----------|
| 155 | Whole-Exome Sequencing Analyses Support a Role of Vitamin D Metabolism in Ischemic Stroke. Stroke, 2023, 54, 800-809. | 1.0 | 1 |
| 157 | Effects of protein-coding variants on blood metabolite measurements and clinical biomarkers in the UK Biobank. American Journal of Human Genetics, 2023, 110, 487-498. | 2.6 | 7 |
| 158 | Rare-variant association analysis reveals known and new age-related hearing loss genes. European Journal of Human Genetics, 2023, 31, 638-647. | 1.4 | 5 |
| 159 | Proteogenomic links to human metabolic diseases. Nature Metabolism, 2023, 5, 516-528. | 5.1 | 9 |
| 160 | Predicting Cancer Risk from Germline Whole-exome Sequencing Data Using a Novel Context-based Variant Aggregation Approach. Cancer Research Communications, 2023, 3, 483-488. | 0.7 | 1 |
| 162 | Exome-wide screening identifies novel rare risk variants for bone mineral density. Osteoporosis International, 2023, 34, 965-975. | 1.3 | 0 |
| 163 | Imputation-powered whole-exome analysis identifies genes associated with kidney function and disease in the UK Biobank. Nature Communications, 2023, 14, . | 5.8 | 3 |
| 165 | Adjusting for common variant polygenic scores improves yield in rare variant association analyses. Nature Genetics, 2023, 55, 544-548. | 9.4 | 9 |
| 167 | The potential of integrating human and mouse discovery platforms to advance our understanding of cardiometabolic diseases. ELife, 0, 12, . | 2.8 | 3 |
| 168 | Systematic elucidation of genetic mechanisms underlying cholesterol uptake. Cell Genomics, 2023, , 100304. | 3.0 | 2 |
| 169 | Bioinformatics for Medicinal Chemistry. , 2023, , 485-513. | | 0 |
| 193 | Exome sequencing identifies breast cancer susceptibility genes and defines the contribution of coding variants to breast cancer risk. Nature Genetics, 2023, 55, 1435-1439. | 9.4 | 6 |