# Mario Falchi

List of Publications by Year
in descending order



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|---|---|---|---|
| 107 papers | 11,899 citations | 53789 45 h-index | 31843 101 g-index |
| 118 all docs | 118 docs citations | 118 times ranked | 23144 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Phenotype, genotype, and worldwide genetic penetrance of LRRK2-associated Parkinson's disease: a case-control study. Lancet Neurology, The, 2008, 7, 583-590. | 10.2 | 1,340 |
| 2 | Real-time tracking of self-reported symptoms to predict potential COVID-19. Nature Medicine, 2020, 26, 1037-1040. | 30.7 | 1,173 |
| 3 | Loci influencing lipid levels and coronary heart disease risk in 16 European population cohorts. Nature Genetics, 2009, 41, 47-55. | 21.4 | 776 |
| 4 | Bone mineral density, osteoporosis, and osteoporotic fractures: a genome-wide association study. Lancet, The, 2008, 371, 1505-1512. | 13.7 | 612 |
| 5 | A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium. Nature Genetics, 2009, 41, 1182-1190. | 21.4 | 481 |
| 6 | Repeatability of published microarray gene expression analyses. Nature Genetics, 2009, 41, 149-155. | 21.4 | 477 |
| 7 | A new highly penetrant form of obesity due to deletions on chromosome 16p11.2. Nature, 2010, 463, 671-675. | 27.8 | 476 |
| 8 | Interplay between the human gut microbiome and host metabolism. Nature Communications, 2019, 10, 4505. | 12.8 | 450 |
| 9 | New gene functions in megakaryopoiesis and platelet formation. Nature, 2011, 480, 201-208. | 27.8 | 401 |
| 10 | Genome-wide Association Study Identifies Genes for Biomarkers of Cardiovascular Disease: Serum Urate and Dyslipidemia. American Journal of Human Genetics, 2008, 82, 139-149. | 6.2 | 397 |
| 11 | New loci for body fat percentage reveal link between adiposity and cardiometabolic disease risk. Nature Communications, 2016, 7, 10495. | 12.8 | 245 |
| 12 | Mapping Genetic Loci That Determine Leukocyte Telomere Length in a Large Sample of Unselected Female Sibling Pairs. American Journal of Human Genetics, 2006, 78, 480-486. | 6.2 | 242 |
| 13 | A reference map of potential determinants for the human serum metabolome. Nature, 2020, 588, 135-140. | 27.8 | 230 |
| 14 | Familial amyotrophic lateral sclerosis is associated with a mutation in D-amino acid oxidase. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 7556-7561. | 7.1 | 229 |
| 15 | Low copy number of the salivary amylase gene predisposes to obesity. Nature Genetics, 2014, 46, 492-497. | 21.4 | 214 |
| 16 | Genome-wide association study identifies variants at 9p21 and 22q13 associated with development of cutaneous nevi. Nature Genetics, 2009, 41, 915-919. | 21.4 | 204 |
| 17 | Genome-Wide Linkage Scan for Athlete Status in 700 British Female DZ Twin Pairs. Twin Research and Human Genetics, 2007, 10, 812-820. | 0.6 | 164 |
| 18 | Genome-wide association meta-analyses combining multiple risk phenotypes provide insights into the genetic architecture of cutaneous melanoma susceptibility. Nature Genetics, 2020, 52, 494-504. | 21.4 | 138 |

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|---|---|---|---|
| 19 | Systems genetics identifies a convergent gene network for cognition and neurodevelopmental disease. Nature Neuroscience, 2016, 19, 223-232. | 14.8 | 131 |
| 20 | Systems biology of the IMIDIA biobank from organ donors and pancreatectomised patients defines a novel transcriptomic signature of islets from individuals with type 2 diabetes. Diabetologia, 2018, 61, 641-657. | 6.3 | 131 |
| 21 | A novel variant on chromosome 7q22.3 associated with mean platelet volume, counts, and function. Blood, 2009, 113, 3831-3837. | 1.4 | 117 |
| 22 | TwinsUK: The UK Adult Twin Registry Update. Twin Research and Human Genetics, 2019, 22, 523-529. | 0.6 | 116 |
| 23 | Nevus Size and Number Are Associated with Telomere Length and Represent Potential Markers of a Decreased Senescence In vivo. Cancer Epidemiology Biomarkers and Prevention, 2007, 16, 1499-1502. | 2.5 | 115 |
| 24 | Symptom clusters in COVID-19: A potential clinical prediction tool from the COVID Symptom Study app. Science Advances, 2021, 7, . | 10.3 | 115 |
| 25 | Epigenetic associations of type 2 diabetes and BMI in an Arab population. Clinical Epigenetics, 2016, 8, 13. | 4.1 | 110 |
| 26 | Current smoking and COVID-19 risk: results from a population symptom app in over 2.4 million people. Thorax, 2021, 76, 714-722. | 5.6 | 105 |
| 27 | Circulating complement factor Hâ€“related proteins 1Âand 5 correlate with disease activity in IgA nephropathy. Kidney International, 2017, 92, 942-952. | 5.2 | 99 |
| 28 | Heritability of serum TSH, free T4 and free T3 concentrations: a study of a large UK twin cohort. Clinical Endocrinology, 2008, 68, 652-659. | 2.4 | 98 |
| 29 | Beneficial effect of a high number of copies of salivary amylase AMY1 gene on obesity risk in Mexican children. Diabetologia, 2015, 58, 290-294. | 6.3 | 89 |
| 30 | Novel pleiotropic risk loci for melanoma and nevus density implicate multiple biological pathways. Nature Communications, 2018, 9, 4774. | 12.8 | 87 |
| 31 | Comparison of Cardioprotective Abilities between the Flesh and Skin of Grapes. Journal of Agricultural and Food Chemistry, 2006, 54, 6613-6622. | 5.2 | 86 |
| 32 | Genome-wide association meta-analysis of individuals of European ancestry identifies new loci explaining a substantial fraction of hair color variation and heritability. Nature Genetics, 2018, 50, 652-656. | 21.4 | 86 |
| 33 | Circulating Levels of the Short-Chain Fatty Acid Acetate Mediate the Effect of the Gut Microbiome on Visceral Fat. Frontiers in Microbiology, 2021, 12, 711359. | 3.5 | 86 |
| 34 | Genetic variants in LEP, LEPR, and <scp>MC4R</scp> explain 30% of severe obesity in children from a consanguineous population. Obesity, 2015, 23, 1687-1695. | 3.0 | 82 |
| 35 | Genome-wide association study in 176,678 Europeans reveals genetic loci for tanning response to sun exposure. Nature Communications, 2018, 9, 1684. | 12.8 | 80 |
| 36 | Association of DNA methylation with age, gender, and smoking in an Arab population. Clinical Epigenetics, 2015, 7, 6. | 4.1 | 78 |

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|---|---|---|---|
| 37 | A New Essential Hypertension Susceptibility Locus on Chromosome 2p24-p25, Detected by Genomewide Search. American Journal of Human Genetics, 2002, 71, 893-905. | 6.2 | 74 |
| 38 | Association of Sirtuin 1 (SIRT1) Gene SNPs and Transcript Expression Levels With Severe Obesity. Obesity, 2012, 20, 178-185. | 3.0 | 68 |
| 39 | Identification of a Novel Gene and a Common Variant Associated with Uric Acid Nephrolithiasis in a Sardinian Genetic Isolate. American Journal of Human Genetics, 2003, 72, 1479-1491. | 6.2 | 65 |
| 40 | Decreased STARD10 Expression Is Associated with Defective Insulin Secretion in Humans and Mice. American Journal of Human Genetics, 2017, 100, 238-256. | 6.2 | 60 |
| 41 | GUESS-ing Polygenic Associations with Multiple Phenotypes Using a GPU-Based Evolutionary Stochastic Search Algorithm. PLoS Genetics, 2013, 9, e1003657. | 3.5 | 58 |
| 42 | Self-Reported Symptoms of COVID-19, Including Symptoms Most Predictive of SARS-CoV-2 Infection, Are Heritable. Twin Research and Human Genetics, 2020, 23, 316-321. | 0.6 | 57 |
| 43 | Novel LEPR mutations in obese Pakistani children identified by PCRâ€based enrichment and next generation sequencing. Obesity, 2014, 22, 1112-1117. | 3.0 | 51 |
| 44 | A Genome-Wide Association Study of Monozygotic Twin-Pairs Suggests a Locus Related to Variability of Serum High-Density Lipoprotein Cholesterol. Twin Research and Human Genetics, 2012, 15, 691-699. | 0.6 | 50 |
| 45 | The relationship between DXA-based and anthropometric measures of visceral fat and morbidity in women. BMC Cardiovascular Disorders, 2013, 13, 25. | 1.7 | 50 |
| 46 | A Genomewide Search Using an Original Pairwise Sampling Approach for Large Genealogies Identifies a New Locus for Total and Low-Density Lipoprotein Cholesterol in Two Genetically Differentiated Isolates of Sardinia. American Journal of Human Genetics, 2004, 75, 1015-1031. | 6.2 | 48 |
| 47 | Diagnostic value of cutaneous manifestation of SARSâ€CoVâ€2 infection*. British Journal of Dermatology, 2021, 184, 880-887. | 1.5 | 45 |
| 48 | Differential coexpression analysis of obesity-associated networks in human subcutaneous adipose tissue. International Journal of Obesity, 2012, 36, 137-147. | 3.4 | 42 |
| 49 | Genome-wide search for nevus density shows linkage to two melanoma loci on chromosome 9 and identifies a new QTL on 5q31 in an adult twin cohort. Human Molecular Genetics, 2006, 15, 2975-2979. | 2.9 | 41 |
| 50 | A Genome-Wide Linkage Scan for Age at Menarche in Three Populations of European Descent. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3965-3970. | 3.6 | 40 |
| 51 | Not all isolates are equal: linkage disequilibrium analysis on Xq13.3 reveals different patterns in Sardinian sub-populations. Human Genetics, 2002, 111, 9-15. | 3.8 | 39 |
| 52 | Novel association approach for variable number tandem repeats (VNTRs) identifies DOCK5 as a susceptibility gene for severe obesity. Human Molecular Genetics, 2012, 21, 3727-3738. | 2.9 | 37 |
| 53 | A Genome-Wide Association Study Identifies rs2000999 as a Strong Genetic Determinant of Circulating Haptoglobin Levels. PLoS ONE, 2012, 7, e32327. | 2.5 | 34 |
| 54 | O- and N-Glycosylation of Serum Immunoglobulin A is Associated with IgA Nephropathy and Glomerular Function. Journal of the American Society of Nephrology: JASN, 2021, 32, 2455-2465. | 6.1 | 33 |

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|---|---|---|---|
| 55 | Metabolomic Profile of Lowâ€“Copy Number Carriers at the Salivary Î±-Amylase Gene Suggests a Metabolic Shift Toward Lipid-Based Energy Production. Diabetes, 2016, 65, 3362-3368. | 0.6 | 32 |
| 56 | Yoghurt consumption is associated with changes in the composition of the human gut microbiome and metabolome. BMC Microbiology, 2022, 22, 39. | 3.3 | 31 |
| 57 | Genetic Loci Linked to Pituitary-Thyroid Axis Set Points: A Genome-Wide Scan of a Large Twin Cohort. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3519-3523. | 3.6 | 30 |
| 58 | Deep molecular phenotypes link complex disorders and physiological insult to CpG methylation. Human Molecular Genetics, 2018, 27, 1106-1121. | 2.9 | 30 |
| 59 | Extensive weight loss reduces glycan age by altering IgG N-glycosylation. International Journal of Obesity, 2021, 45, 1521-1531. | 3.4 | 29 |
| 60 | IgA1 Glycosylation Is Heritable in Healthy Twins. Journal of the American Society of Nephrology: JASN, 2017, 28, 64-68. | 6.1 | 27 |
| 61 | Conducting metagenomic studies in microbiology and clinical research. Applied Microbiology and Biotechnology, 2018, 102, 8629-8646. | 3.6 | 26 |
| 62 | Laser capture microdissection of human pancreatic islets reveals novel eQTLs associated with type 2 diabetes. Molecular Metabolism, 2019, 24, 98-107. | 6.5 | 26 |
| 63 | Genome-wide scan identifies a copy number variable region at 3p21.1 that influences the TLR9 expression levels in IgA nephropathy patients. European Journal of Human Genetics, 2015, 23, 940-948. | 2.8 | 23 |
| 64 | Omics studies for comprehensive understanding of immunoglobulin A nephropathy: state-of-the-art and future directions. Nephrology Dialysis Transplantation, 2018, 33, 2101-2112. | 0.7 | 23 |
| 65 | YAMP: a containerized workflow enabling reproducibility in metagenomics research. GigaScience, 2018, 7, . | 6.4 | 22 |
| 66 | CDKN2B expression and subcutaneous adipose tissue expandability: Possible influence of the 9p21 atherosclerosis locus. Biochemical and Biophysical Research Communications, 2014, 446, 1126-1131. | 2.1 | 20 |
| 67 | Changes in levels of peripheral hormones controlling appetite are inconsistent with hyperphagia in leptin-deficient subjects. Endocrine, 2014, 45, 401-408. | 2.3 | 19 |
| 68 | Investigating the effects of copy number variants on reading and language performance. Journal of Neurodevelopmental Disorders, 2016, 8, 17. | 3.1 | 19 |
| 69 | Impact of statistical models on the prediction of type 2 diabetes using non-targeted metabolomics profiling. Molecular Metabolism, 2016, 5, 918-925. | 6.5 | 18 |
| 70 | Associations of Genetic Variants Contributing to Gut Microbiota Composition in Immunoglobin A Nephropathy. MSystems, 2021, 6, . | 3.8 | 18 |
| 71 | Immune Trait Shifts in Association With Tobacco Smoking: A Study in Healthy Women. Frontiers in Immunology, 2021, 12, 637974. | 4.8 | 18 |
| 72 | Metabolome Genome-Wide Association Study Identifies 74 Novel Genomic Regions Influencing Plasma Metabolites Levels. Metabolites, 2022, 12, 61. | 2.9 | 18 |

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|---|---|---|---|
| 73 | ACE2 expression in adipose tissue is associated with cardio-metabolic risk factors and cell type compositionâ€"implications for COVID-19. International Journal of Obesity, 2022, 46, 1478-1486. | 3.4 | 18 |
| 74 | Multiple rare genetic variants coâ€segregating with familial IgA nephropathy all act within a single immuneâ€related network. Journal of Internal Medicine, 2017, 281, 189-205. | 6.0 | 17 |
| 75 | Rare variant analysis in eczema identifies exonic variants in DUSP1, NOTCH4 and SLC9A4. Nature Communications, 2021, 12, 6618. | 12.8 | 17 |
| 76 | Mendelian inheritance of trimodal CpG methylation sites suggests distal cis-acting genetic effects. Clinical Epigenetics, 2016, 8, 124. | 4.1 | 16 |
| 77 | Jenti: an efficient tool for mining complex inbred genealogies. Bioinformatics, 2008, 24, 724-726. | 4.1 | 15 |
| 78 | Body siteâ€specific genetic effects influence naevus count distribution in women. Pigment Cell and Melanoma Research, 2020, 33, 326-333. | 3.3 | 15 |
| 79 | Hyperalgesic effect of intrathecally administered interleukin-1 in rats. Drugs Under Experimental and Clinical Research, 2001, 27, 97-101. | 0.3 | 15 |
| 80 | Identification of QTLs for serum lipid levels in a female sib-pair cohort: a novel application to improve the power of two-locus linkage analysis. Human Molecular Genetics, 2005, 14, 2971-2979. | 2.9 | 14 |
| 81 | Identification of Quantitative Trait Loci for Fibrin Clot Phenotypes. Arteriosclerosis, Thrombosis, and Vascular Biology, 2009, 29, 600-605. | 2.4 | 14 |
| 82 | Low-Frequency Variants in HMGA1 Are Not Associated With Type 2 Diabetes Risk. Diabetes, 2012, 61, 524-530. | 0.6 | 14 |
| 83 | Plasma N-glycome shows continuous deterioration as the diagnosis of insulin resistance approaches. BMJ Open Diabetes Research and Care, 2021, 9, e002263. | 2.8 | 13 |
| 84 | Statistical tools for linkage analysis and genetic association studies. Expert Review of Molecular Diagnostics, 2005, 5, 781-796. | 3.1 | 11 |
| 85 | Quantitative Linkage Analysis for Pancreatic B-cell Function and Insulin Resistance in a Large Twin Cohort. Diabetes, 2008, 57, 1120-1124. | 0.6 | 11 |
| 86 | Intrauterine, Environmental, and Genetic Influences in the Relationship Between Birth Weight and Lipids in a Female Twin Cohort. Arteriosclerosis, Thrombosis, and Vascular Biology, 2006, 26, 2373-2379. | 2.4 | 10 |
| 87 | famCNV: copy number variant association for quantitative traits in families. Bioinformatics, 2011, 27, 1873-1875. | 4.1 | 10 |
| 88 | Fine-Mapping of the Human Blood Plasma N-Glycome onto Its Proteome. Metabolites, 2019, 9, 122. | 2.9 | 10 |
| 89 | Genome-wide Scan for Prospective Memory Suggests Linkage to Chromosome 12q22. Behavior Genetics, 2006, 36, 18-28. | 2.1 | 9 |
| 90 | PedVizApi: a Java API for the interactive, visual analysis of extended pedigrees. Bioinformatics, 2008, 24, 279-281. | 4.1 | 8 |

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|---|---|---|---|
| 91 | Ethnic-specific association of amylase gene copy number with adiposity traits in a large Middle Eastern biobank. Npj Genomic Medicine, 2021, 6, 8. | 3.8 | 8 |
| 92 | PopPAnTe: population and pedigree association testing for quantitative data. BMC Genomics, 2017, 18, 150. | 2.8 | 6 |
| 93 | Acne and Telomere Length: A New Spectrum between Senescence and Apoptosis Pathways. Journal of Investigative Dermatology, 2017, 137, 513-515. | 0.7 | 6 |
| 94 | Looking for Sunshine: Genetic Predisposition to Sun Seeking in 265,000 Individuals of European Ancestry. Journal of Investigative Dermatology, 2021, 141, 779-786. | 0.7 | 5 |
| 95 | Association Between Medication-Taking and Refractive Error in a Large General Population-Based Cohort. , 2021, 62, 15. | | 5 |
| 96 | Genomic expression differences between cutaneous cells from red hair color individuals and black hair color individuals based on bioinformatic analysis. Oncotarget, 2017, 8, 11589-11599. | 1.8 | 5 |
| 97 | miRNAs as Molecular Biomarkers for Prostate Cancer. Journal of Molecular Diagnostics, 2022, 24, 1171-1180. | 2.8 | 5 |
| 98 | Diclofenac-Choline Antioxidant Activity Investigated by means of Luminol Amplified Chemiluminescence of Human Neutrophil Bursts and Electron Paramagnetic Resonance Spectroscopy. Drug Research, 2015, 65, 244-251. | 1.7 | 4 |
| 99 | PowQ: a user-friendly package for the design of variance component multipoint linkage analysis studies. Bioinformatics, 2006, 22, 1404-1405. | 4.1 | 3 |
| 100 | Genetic Mapping of Quantitative Trait Loci for Disease-Related Phenotypes. Methods in Molecular Biology, 2012, 871, 281-311. | 0.9 | 2 |
| 101 | Genetics plays a role in nevi distribution in women. Melanoma Management, 2020, 7, MMT35. | 0.5 | 2 |
| 102 | Real-time tracking of self-reported symptoms to predict potential COVID-19. , 0, . | | 1 |
| 103 | A Common Single Nucleotide Polymorphism in the Chromosome 7q22.3 Region, Which Is Frequently Deleted in Myeloid Malignancies, Is Associated with Mean Platelet Volume and Platelet Function in Healthy Individuals. Blood, 2008, 112, 86-86. | 1.4 | 1 |
| 104 | Analysis of Quantitative Trait Loci. Methods in Molecular Biology, 2008, 453, 297-326. | 0.9 | 0 |
| 105 | A statistical test for detecting parent-of-origin effects when parental information is missing. Statistical Applications in Genetics and Molecular Biology, 2017, 16, 275-289. | 0.6 | 0 |
| 106 | Intracerebroventricular capsaicin and food intake in the rat. Drugs Under Experimental and Clinical Research, 2001, 27, 61-7. | 0.3 | 0 |
| 107 | A genome-wide analysis of 340â€‰318 participants identifies four novel loci associated with the age of first spectacle wear. Human Molecular Genetics, 2022, , . | 2.9 | 0 |