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List of Publications by Year in descending order

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46799 57758 8,481 127 44 89 citations h-index g-index papers 133 133 133 9612 docs citations times ranked citing authors all docs

#	Article	lF	CITATIONS
1	Mutations in IRF6 cause Van der Woude and popliteal pterygium syndromes. Nature Genetics, 2002, 32, 285-289.	21.4	784
2	Interferon Regulatory Factor 6 ($\langle i \rangle$ IRF6 $\langle i \rangle$) Gene Variants and the Risk of Isolated Cleft Lip or Palate. New England Journal of Medicine, 2004, 351, 769-780.	27.0	534
3	Epilepsy, Ataxia, Sensorineural Deafness, Tubulopathy, and <i>KCNJ10</i> Mutations. New England Journal of Medicine, 2009, 360, 1960-1970.	27.0	518
4	A genome-wide association study of cleft lip with and without cleft palate identifies risk variants near MAFB and ABCA4. Nature Genetics, 2010, 42, 525-529.	21.4	518
5	Risk HLA-DQA1 and PLA ₂ R1 Alleles in Idiopathic Membranous Nephropathy. New England Journal of Medicine, 2011, 364, 616-626.	27.0	442
6	Mutations in SLC6A19, encoding BOAT1, cause Hartnup disorder. Nature Genetics, 2004, 36, 999-1002.	21,4	272
7	A common variant of the latrophilin 3 gene, LPHN3, confers susceptibility to ADHD and predicts effectiveness of stimulant medication. Molecular Psychiatry, 2010, 15, 1053-1066.	7.9	245
8	Genetics of population isolates. Clinical Genetics, 2002, 61, 233-247.	2.0	227
9	Young adult outcomes in the followâ€up of the multimodal treatment study of attentionâ€deficit/hyperactivity disorder: symptom persistence, source discrepancy, and height suppression. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2017, 58, 663-678.	5.2	207
10	Meta-Analysis of 13 Genome Scans Reveals Multiple Cleft Lip/Palate Genes with Novel Loci on 9q21 and 2q32-35. American Journal of Human Genetics, 2004, 75, 161-173.	6.2	200
11	Attention-Deficit/Hyperactivity Disorder in a Population Isolate: Linkage to Loci at 4q13.2, 5q33.3, 11q22, and 17p11. American Journal of Human Genetics, 2004, 75, 998-1014.	6.2	192
12	Support for association between ADHD and two candidate genes:NET1andDRD1. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2005, 134B, 67-72.	1.7	180
13	A multi-ethnic genome-wide association study identifies novel loci for non-syndromic cleft lip with or without cleft palate on 2p24.2, 17q23 and 19q13. Human Molecular Genetics, 2016, 25, ddw104.	2.9	163
14	Metaâ€analysis of genomeâ€wide linkage scans of attention deficit hyperactivity disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1392-1398.	1.7	160
15	Early Interstitial Lung Disease in Familial Pulmonary Fibrosis. American Journal of Respiratory and Critical Care Medicine, 2007, 176, 698-705.	5.6	157
16	A Genome-wide Association Study of Nonsyndromic Cleft Palate Identifies an Etiologic Missense Variant in GRHL3. American Journal of Human Genetics, 2016, 98, 744-754.	6.2	146
17	Linear clinical progression, independent of age of onset, in Niemann–Pick disease, type C. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 132-140.	1.7	145
18	Myalgic Encephalomyelitis/Chronic Fatigue Syndrome: A Comprehensive Review. Diagnostics, 2019, 9, 91.	2.6	140

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19	Gray platelet syndrome: natural history of a large patient cohort and locus assignment to chromosome 3p. Blood, 2010, 116, 4990-5001.	1.4	137
20	FOXE1 association with both isolated cleft lip with or without cleft palate, and isolated cleft palate. Human Molecular Genetics, 2009, 18, 4879-4896.	2.9	136
21	Genome Scan, Fine-Mapping, and Candidate Gene Analysis of Non-Syndromic Cleft Lip with or without Cleft Palate Reveals Phenotype-Specific Differences in Linkage and Association Results. Human Heredity, 2009, 68, 151-170.	0.8	113
22	Contribution of LPHN3 to the genetic susceptibility to ADHD in adulthood: a replication study. Genes, Brain and Behavior, 2011, 10, 149-157.	2.2	103
23	Mistargeting of Peroxisomal EHHADH and Inherited Renal Fanconi's Syndrome. New England Journal of Medicine, 2014, 370, 129-138.	27.0	99
24	APOE*E2 allele delays age of onset in PSEN1 E280A Alzheimer's disease. Molecular Psychiatry, 2016, 21, 916-924.	7.9	89
25	Vitiligo: complex segregation and linkage disequilibrium analyses with respect to microsatellite loci spanning the HLA. Human Genetics, 2002, 110, 334-342.	3.8	81
26	Prediction of susceptibility to major depression by a model of interactions of multiple functional genetic variants and environmental factors. Molecular Psychiatry, 2012, 17, 624-633.	7.9	79
27	CRISPLD2: a novel NSCLP candidate gene. Human Molecular Genetics, 2007, 16, 2241-2248.	2.9	78
28	Latent Class Subtyping of Attention-Deficit/Hyperactivity Disorder and Comorbid Conditions. Journal of the American Academy of Child and Adolescent Psychiatry, 2008, 47, 797-807.	0.5	73
29	Attention-Deficit/Hyperactivity Disorder and Comorbid Disruptive Behavior Disorders: Evidence of Pleiotropy and New Susceptibility Loci. Biological Psychiatry, 2007, 61, 1329-1339.	1.3	69
30	Environmental influences that affect attention deficit/hyperactivity disorder. European Child and Adolescent Psychiatry, 2007, 16, 337-346.	4.7	69
31	Attention deficit/hyperactivity disorder (ADHD): Complex phenotype, simple genotype?. Genetics in Medicine, 2004, 6, 1-15.	2.4	65
32	Polyautoimmunity in Sjögren Syndrome. Rheumatic Disease Clinics of North America, 2016, 42, 457-472.	1.9	65
33	The Multiple Autoimmune Syndromes. A Clue for the Autoimmune Tautology. Clinical Reviews in Allergy and Immunology, 2012, 43, 256-264.	6.5	64
34	Pedigree disequilibrium test (PDT) replicates association and linkage between DRD4 and ADHD in multigenerational and extended pedigrees from a genetic isolate. Molecular Psychiatry, 2004, 9, 252-259.	7.9	61
35	A common genetic network underlies substance use disorders and disruptive or externalizing disorders. Human Genetics, 2012, 131, 917-929.	3.8	60
36	<i>LPHN</i> 3 and attentionâ€deficit/hyperactivity disorder: a susceptibility and pharmacogenetic study. Genes, Brain and Behavior, 2015, 14, 419-427.	2.2	58

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37	The PHF21B gene is associated with major depression and modulates the stress response. Molecular Psychiatry, 2017, 22, 1015-1025.	7.9	56
38	Attention-Deficit/Hyperactivity Disorder and Comorbidities in 18 Paisa Colombian Multigenerational Families. Journal of the American Academy of Child and Adolescent Psychiatry, 2004, 43, 1506-1515.	0.5	52
39	A cooperative interaction between LPHN3 and 11q doubles the risk for ADHD. Molecular Psychiatry, 2012, 17, 741-747.	7.9	52
40	Genetic analysis of candidate loci in non-syndromic cleft lip families from Antioquia-Colombia and Ohio. American Journal of Medical Genetics Part A, 2004, 125A, 135-144.	2.4	51
41	Screening of human <i>LPHN3</i> for variants with a potential impact on ADHD susceptibility. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 11-18.	1.7	49
42	An Ultraconserved Brain-Specific Enhancer Within ADGRL3 (LPHN3) Underpins Attention-Deficit/Hyperactivity Disorder Susceptibility. Biological Psychiatry, 2016, 80, 943-954.	1.3	48
43	Neurocysticercosis in Persons with Epilepsy in Medellin, Colombia. Epilepsia, 1998, 39, 1334-1339.	5.1	47
44	Distinctive adaptive response to repeated exposure to hydrogen peroxide associated with upregulation of DNA repair genes and cell cycle arrest. Redox Biology, 2016, 9, 124-133.	9.0	47
45	Genetics of non-syndromic childhood obesity and the use of high-throughput DNA sequencing technologies. Journal of Diabetes and Its Complications, 2017, 31, 1549-1561.	2.3	43
46	ADHD latent class clusters: DSM-IV subtypes and comorbidity. Psychiatry Research, 2009, 170, 192-198.	3.3	42
47	Origin of the <i>PSEN1</i> E280A mutation causing earlyâ€onset Alzheimer's disease. Alzheimer's and Dementia, 2014, 10, S277-S283.e10.	0.8	39
48	Role of the IL-1 Pathway in Dopaminergic Neurodegeneration and Decreased Voluntary Movement. Molecular Neurobiology, 2017, 54, 4486-4495.	4.0	38
49	A two-locus genetic interaction between LPHN3 and $11q$ predicts ADHD severity and long-term outcome. Translational Psychiatry, 2011, 1, e17-e17.	4.8	37
50	GWAS reveals new recessive loci associated with non-syndromic facial clefting. European Journal of Medical Genetics, 2012, 55, 510-514.	1.3	37
51	Pooling/bootstrap-based GWAS (pbGWAS) identifies new loci modifying the age of onset in PSEN1 p.Glu280Ala Alzheimer's disease. Molecular Psychiatry, 2013, 18, 568-575.	7.9	37
52	Toward a better understanding of ADHD: LPHN3 gene variants and the susceptibility to develop ADHD. ADHD Attention Deficit and Hyperactivity Disorders, 2010, 2, 139-147.	1.7	36
53	Tuning major gene variants conditioning human behavior: the anachronism of ADHD. Current Opinion in Genetics and Development, 2007, 17, 234-238.	3.3	35
54	Influence of a Latrophilin 3 (LPHN3) risk haplotype on event-related potential measures of cognitive response control in attention-deficit hyperactivity disorder (ADHD). European Neuropsychopharmacology, 2013, 23, 458-468.	0.7	35

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55	<i> <scp>ADGRL</scp> 3 (<scp>LPHN</scp> 3) </i> variants are associated with a refined phenotype of <scp>ADHD</scp> in the <scp>MTA</scp> study. Molecular Genetics & Genomic Medicine, 2016, 4, 540-547.	1.2	35
56	Clinical Outcomes and Genome-Wide Association for a Brain Methylation Site in an Antidepressant Pharmacogenetics Study in Mexican Americans. American Journal of Psychiatry, 2014, 171, 1297-1309.	7.2	33
57	A multifactorial model of pathology for age of onset heterogeneity in familial Alzheimer's disease. Acta Neuropathologica, 2021, 141, 217-233.	7.7	33
58	TAP, HLA-DQB1, and HLA-DRB1 polymorphism in Colombian patients with primary Sjögren's syndrome. Seminars in Arthritis and Rheumatism, 2002, 31, 396-405.	3.4	31
59	Rheumatoid arthritis association in Colombian population is restricted to HLA-DRB1*04 QRRAA alleles. Genes and Immunity, 2002, 3, 56-58.	4.1	31
60	Novel and rare functional genomic variants in multiple autoimmune syndrome and Sjögren's syndrome. Journal of Translational Medicine, 2015, 13, 173.	4.4	30
61	Rare DNA variants in the brain-derived neurotrophic factor gene increase risk for attention-deficit hyperactivity disorder: a next-generation sequencing study. Molecular Psychiatry, 2017, 22, 580-584.	7.9	30
62	Attention-deficit/hyperactivity disorder (ADHD): feasibility of linkage analysis in a genetic isolate using extended and multigenerational pedigrees. Clinical Genetics, 2002, 61, 335-343.	2.0	29
63	From the black widow spider to human behavior: Latrophilins, a relatively unknown class of G proteinâ€coupled receptors, are implicated in psychiatric disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 1-10.	1.7	29
64	Candidate gene discovery in autoimmunity by using extreme phenotypes, next generation sequencing and whole exome capture. Autoimmunity Reviews, 2015, 14, 204-209.	5.8	29
65	ADGRL3 (LPHN3) variants predict substance use disorder. Translational Psychiatry, 2019, 9, 42.	4.8	29
66	TAP1 and TAP2 polymorphisms analysis in northwestern Colombian patients with systemic lupus erythematosus. Annals of the Rheumatic Diseases, 2003, 62, 363-365.	0.9	28
67	An ¹ Hâ€MRS framework predicts the onset of Alzheimer's disease symptoms in <i>PSEN1</i> mutation carriers. Alzheimer's and Dementia, 2014, 10, 552-561.	0.8	26
68	A Mutation in <i>DAOA</i> Modifies the Age of Onset in <i>PSEN1</i> E280A Alzheimer's Disease. Neural Plasticity, 2016, 2016, 1-7.	2.2	25
69	Low-frequency and rare variants may contribute to elucidate the genetics of major depressive disorder. Translational Psychiatry, 2018, 8, 70.	4.8	25
70	Association of PDE11A global haplotype with major depression and antidepressant drug response. Neuropsychiatric Disease and Treatment, 2009, 5, 163.	2.2	24
71	What is next after the genes for autoimmunity?. BMC Medicine, 2013, 11, 197.	5. 5	23
72	Congenital Leptin Deficiency and Leptin Gene Missense Mutation Found in Two Colombian Sisters with Severe Obesity. Genes, 2019, 10, 342.	2.4	21

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73	Analysis of brain metabolism by proton magnetic resonance spectroscopy (1H-MRS) in attention-deficit/hyperactivity disorder suggests a generalized differential ontogenic pattern from controls. ADHD Attention Deficit and Hyperactivity Disorders, 2012, 4, 205-212.	1.7	20
74	Mutations modifying sporadic Alzheimer's disease age of onset. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 1116-1130.	1.7	20
75	Multiple sclerosis in the tropics: genetic association to STR's loci spanning the HLA and TNF. Multiple Sclerosis Journal, 2002, 8, 249-255.	3.0	19
76	Polymorphisms in the neural nicotinic acetylcholine receptor $\hat{l}\pm 4$ subunit (CHRNA4) are associated with ADHD in a genetic isolate. ADHD Attention Deficit and Hyperactivity Disorders, 2009, 1, 19-24.	1.7	19
77	Whole Exome Sequencing of Extreme Morbid Obesity Patients: Translational Implications for Obesity and Related Disorders. Genes, 2014, 5, 709-725.	2.4	19
78	Definition of mutations in polyautoimmunity. Journal of Autoimmunity, 2016, 72, 65-72.	6.5	19
79	Linkage and association analysis of ADHD endophenotypes in extended and multigenerational pedigrees from a genetic isolate. Molecular Psychiatry, 2016, 21, 1434-1440.	7.9	19
80	A latent genetic subtype of major depression identified by whole-exome genotyping data in a Mexican-American cohort. Translational Psychiatry, 2017, 7, e1134-e1134.	4.8	19
81	Chagas' disease susceptibility/resistance: linkage disequilibrium analysis suggests epistasis between major histocompatibility complex and interleukin-10. Tissue Antigens, 2004, 64, 18-24.	1.0	18
82	Pharmacogenetic Impact of VKORC1 and CYP2C9 Allelic Variants on Warfarin Dose Requirements in a Hispanic Population Isolate. Clinical and Applied Thrombosis/Hemostasis, 2010, 16, 83-90.	1.7	18
83	Exploratory data from complete genomes of familial alzheimer disease age-at-onset outliers. Human Mutation, 2012, 33, 1630-1634.	2.5	18
84	Uveitis and Multiple Sclerosis: Potential Common Causal Mutations. Molecular Neurobiology, 2019, 56, 8008-8017.	4.0	18
85	Potential cognitive endophenotypes in multigenerational families: segregating ADHD from a genetic isolate. ADHD Attention Deficit and Hyperactivity Disorders, 2011, 3, 291-299.	1.7	17
86	A multigenerational pedigree of late-onset Alzheimer's disease implies new genetic causes. Brain, 2005, 128, 1707-1715.	7.6	14
87	Genetic Variation Underpinning ADHD Risk in a Caribbean Community. Cells, 2019, 8, 907.	4.1	14
88	A New Method for Detecting Significant p-values with Applications to Genetic Data. Revista Colombiana De Estadistica, 2014, 37, 69.	0.4	13
89	Clonal Population Structure of Colombian Sylvatic Trypanosoma cruzi. Journal of Parasitology, 1998, 84, 1143.	0.7	11
90	Multiple Sclerosis: Association to HLA DQ $\hat{l}\pm$ in a Tropical Population. Experimental and Clinical Immunogenetics, 1999, 16, 131-138.	1.2	11

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91	Machine Learning Prediction of ADHD Severity: Association and Linkage to <i>ADGRL3</i> , <i>DRD4</i> , and <i>SNAP25</i> . Journal of Attention Disorders, 2022, 26, 587-605.	2.6	11
92	Chikungunya outbreak (2015) in the Colombian Caribbean: Latent classes and gender differences in virus infection. PLoS Neglected Tropical Diseases, 2020, 14, e0008281.	3.0	10
93	Planning in borderline personality disorder: Evidence for distinct subpopulations. World Journal of Biological Psychiatry, 2009, 10, 512-517.	2.6	8
94	Neural Plasticity during Aging. Neural Plasticity, 2019, 2019, 1-3.	2.2	8
95	A novel alteration in metaxin 1, F202L, is associated with N370S in Gaucher disease. Journal of Human Genetics, 2004, 49, 220-222.	2.3	7
96	Targeting Neuroplasticity, Cardiovascular, and Cognitive-Associated Genomic Variants in Familial Alzheimer's Disease. Molecular Neurobiology, 2019, 56, 3235-3243.	4.0	7
97	ADHD Endophenotypes in Caribbean Families. Journal of Attention Disorders, 2020, 24, 2100-2114.	2.6	7
98	Familial Alzheimer's Disease and Recessive Modifiers. Molecular Neurobiology, 2020, 57, 1035-1043.	4.0	7
99	Mutations in sphingolipid metabolism genes are associated with ADHD. Translational Psychiatry, 2020, 10, 231.	4.8	7
100	Complex segregation analysis of nonsyndromic cleft lip/palate in a Chilean population. Genetics and Molecular Biology, 1998, 21, 139-144.	1.3	7
101	Locus homogeneity between syndactyly type 1A and craniosynostosis Philadelphia type?. American Journal of Medical Genetics, Part A, 2008, 146A, 2308-2311.	1.2	6
102	Complex segregation analysis of non-myoclonic idiopathic generalized epilepsy in families ascertained from probands affected with idiopathic epilepsy with tonic-clonic seizures in Antioquia, Colombia. Human Genetics, 1996, 98, 214-218.	3.8	5
103	Calpainopathy: Description of a Novel Mutation and Clinical Presentation with Early Severe Contractures. Genes, 2020, 11, 129.	2.4	5
104	Temporal Gene Expression in the Hippocampus and Peripheral Organs to Endotoxin-Induced Systemic Inflammatory Response in Caspase-1-Deficient Mice. NeuroImmunoModulation, 2015, 22, 263-273.	1.8	4
105	Retrospective assessment of childhood ADHD symptoms for diagnosis in adults: validity of a short 8-item version of the Wender-Utah Rating Scale. ADHD Attention Deficit and Hyperactivity Disorders, 2016, 8, 215-223.	1.7	4
106	Generation of one iPSC line (IMEDEAi006-A) from an early-onset familial Alzheimer's Disease (fAD) patient carrying the E280A mutation in the PSEN1 gene. Stem Cell Research, 2019, 37, 101440.	0.7	4
107	ADGRL3, FGF1 and DRD4: Linkage and Association with Working Memory and Perceptual Organization Candidate Endophenotypes in ADHD. Brain Sciences, 2021, 11, 854.	2.3	4
108	Impulsive and Omission Errors: Potential Temporal Processing Endophenotypes in ADHD. Brain Sciences, 2021, 11, 1218.	2.3	4

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109	Structural Protein Effects Underpinning Cognitive Developmental Delay of the PURA p.Phe233del Mutation Modelled by Artificial Intelligence and the Hybrid Quantum Mechanics–Molecular Mechanics Framework. Brain Sciences, 2022, 12, 871.	2.3	4
110	Complex segregation analysis of schizophrenia in Santiago, Chile. Schizophrenia Research, 1997, 26, 65-69.	2.0	3
111	Rare Functional Variants Associated with Antidepressant Remission in Mexican-Americans. Journal of Affective Disorders, 2021, 279, 491-500.	4.1	3
112	A Comprehensive Machine Learning Framework for the Exact Prediction of the Age of Onset in Familial and Sporadic Alzheimer's Disease. Diagnostics, 2021, 11, 887.	2.6	2
113	Utility of a Short Neuropsychological Protocol for Detecting HIV-Associated Neurocognitive Disorders in Patients with Asymptomatic HIV-1 Infection. Brain Sciences, 2021, 11, 1037.	2.3	2
114	Executive function deficit in bipolar offspring: A neurocognitive endophenotype?. Journal of Affective Disorders, 2022, 297, 246-249.	4.1	2
115	The Mendelian Legacy to Mental and Behavioral Disorders. International Journal of Psychological Research, 2020, 13, 6-8.	0.6	2
116	D6S439 microsatellite identifies a new susceptibility region for primary Sj \tilde{A} ¶gren's syndrome. Journal of Rheumatology, 2003, 30, 2152-6.	2.0	2
117	Human Genetic Host Factors and Its Role in the Pathogenesis of Chikungunya Virus Infection. Frontiers in Medicine, 2022, 9, 654395.	2.6	2
118	Response to Uher et al American Journal of Psychiatry, 2015, 172, 396-398.	7. 2	1
119	Neural Plasticity in Obesity and Psychiatric Disorders. Neural Plasticity, 2016, 2016, 1-3.	2.2	1
120	The role of psychosocial adversity in the aetiology and course of attention deficit hyperactivity disorder. Revista Colombiana De PsiquiatrÃa, 2021, , .	0.3	1
121	Clinical features of multiple sclerosis in a genetically homogeneous tropical population. Multiple Sclerosis Journal, 2001, 7, 227-229.	3.0	1
122	Reproductive success is predicted by social dynamics and kinship in managed animal populations. F1000Research, 2016, 5, 870.	1.6	1
123	Frequency of actionable Exomic secondary findings in 160 Colombian patients: Impact in the healthcare system. Gene, 2022, 838, 146699.	2.2	1
124	Psychopathological Risk in Siblings of Subjects with Attention-Deficit/Hyperactivity Disorder: A cross-Sectional Study. Revista Colombiana De PsiquiatrÃa, 2021, , .	0.3	0
125	Cross validation of pooling/resampling GWAS using the WTCCC data. Molecular Biology and Genetic Engineering, 2015, 3, 1.	0.8	0
126	Ataxia with Ocular Apraxia Type 1 (AOA1) (APTX, W279* Mutation): Neurological, Neuropsychological, and Molecular Outlining of a Heterogenous Phenotype in Four Colombian Siblings. Molecular Neurobiology, 2022, , 1.	4.0	0

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127	MejorÃa del ACV con estimulación magnética recapitula la ontogenia. Acta Medica Colombiana: AMC: Organo De La Asociacion Columbiana De Medicina Interna, 2022, 47, .	0.0	O