Raquel Rabionet

List of Publications by Year
in descending order

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Version: 2024-02-01

87 papers	11,564 citations	87888 38 h-index	51608 86 g-index
100 all docs	100 docs citations	100 times ranked	21351 citing authors

#	Article	IF	Citations
1	Changes in the stool and oropharyngeal microbiome in obsessive-compulsive disorder. Scientific Reports, 2022, 12, 1448.	3.3	16
2	Gene Network of Susceptibility to Atypical Femoral Fractures Related to Bisphosphonate Treatment. Genes, 2022, 13, 146.	2.4	3
3	Diagnostic yield of next-generation sequencing in 87 families with neurodevelopmental disorders. Orphanet Journal of Rare Diseases, 2022, 17, 60.	2.7	21
4	On the association between Chiari malformation type 1, bone mineral density and bone related genes. Bone Reports, 2022, 16, 101181.	0.4	1
5	Shared genetic risk between eating disorderâ€•and substanceâ€useâ€related phenotypes: Evidence from genomeâ€wide association studies. Addiction Biology, 2021, 26, e12880.	2.6	28
6	De Novo PORCN and ZIC2 Mutations in a Highly Consanguineous Family. International Journal of Molecular Sciences, 2021, 22, 1549.	4.1	4
7	Efficient and flexible Integration of variant characteristics in rare variant association studies using integrated nested Laplace approximation. PLoS Computational Biology, 2021, 17, e1007784.	3.2	4
8	The alternative serotonin transporter promoter P2 impacts gene function in females with irritable bowel syndrome. Journal of Cellular and Molecular Medicine, 2021, 25, 8047-8061.	3.6	5
9	Functional Analyses of Four CYP1A1 Missense Mutations Present in Patients with Atypical Femoral Fractures. International Journal of Molecular Sciences, 2021, 22, 7395.	4.1	6
10	Severe Autoinflammatory Manifestations and Antibody Deficiency Due to Novel Hypermorphic PLCG2 Mutations. Journal of Clinical Immunology, 2020, 40, 987-1000.	3.8	41
11	Retrospective evaluation of whole exome and genome mutation calls in 746 cancer samples. Nature Communications, 2020, 11, 4748.	12.8	27
12	Sex differences in oncogenic mutational processes. Nature Communications, 2020, 11, 4330.	12.8	60
13	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
14	Looking into the genetic bases of OCD dimensions: a pilot genome-wide association study. Translational Psychiatry, 2020, 10, 151.	4.8	18
15	Pan-cancer analysis of whole genomes. Nature, 2020, 578, 82-93.	27.8	1,966
16	Genetic identification of cell types underlying brain complex traits yields insights into the etiology of Parkinsonâ€™s disease. Nature Genetics, 2020, 52, 482-493.	21.4	216
17	Exploring genetic variants in obsessive compulsive disorder severity: A GWAS approach. Journal of Affective Disorders, 2020, 267, 23-32.	4.1	2
18	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. Nature Genetics, 2019, 51, 1207-1214.	21.4	641

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19	Epigenetic modification of the pentose phosphate pathway and the IGF-axis in women with gestational diabetes mellitus. Epigenomics, 2019, 11, 1371-1385.	2.1	12
20	Serum magnesium and calcium levels in relation to ischemic stroke. Neurology, 2019, 92, e944-e950.	1.1	38
21	Associations Between Attention-Deficit/Hyperactivity Disorder and Various Eating Disorders: A Swedish Nationwide Population Study Using Multiple Genetically Informative Approaches. Biological Psychiatry, 2019, 86, 577-586.	1.3	43
22	eDiVAâ€"Classification and prioritization of pathogenic variants for clinical diagnostics. Human Mutation, 2019, 40, 865-878.	2.5	19
23	Biallelic loss-of-function LACC1/FAMIN Mutations Presenting as Rheumatoid Factor-Negative Polyarticular Juvenile Idiopathic Arthritis. Scientific Reports, 2019, 9, 4579.	3.3	20
24	Do polygenic risk and stressful life events predict pharmacological treatment response in obsessive compulsive disorder? A geneâ€“environment interaction approach. Translational Psychiatry, 2019, 9, 70.	4.8	19
25	Spectrum of clinical heterogeneity of Î²-tubulin TUBB5 gene mutations. Gene, 2019, 695, 12-17.	2.2	11
26	Allele balance bias identifies systematic genotyping errors and false disease associations. Human Mutation, 2019, 40, 115-126.	2.5	23
27	PATJ Low Frequency Variants Are Associated With Worse Ischemic Stroke Functional Outcome. Circulation Research, 2019, 124, 114-120.	4.5	49
28	Investigation of common, low-frequency and rare genome-wide variation in anorexia nervosa. Molecular Psychiatry, 2018, 23, 1169-1180.	7.9	32
29	An Integrated Data Resource for Genomic AnalysisÂof Cutaneous T-Cell Lymphoma. Journal of Investigative Dermatology, 2018, 138, 2681-2683.	0.7	38
30	Methylation analysis in SÃ©zary syndrome and integration of exome and transcriptome data. European Journal of Cancer, 2018, 101, S13-S14.	2.8	0
31	The ASXL1 mutation p.Gly646Trpfs*12 found in a Turkish boy with Bohringâ€Opitz Syndrome. Clinical Case Reports (discontinued), 2018, 6, 1452-1456.	0.5	6
32	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
33	Severe neurocognitive and growth disorders due to variation in THOC2 , an essential component of nuclear mRNA export machinery. Human Mutation, 2018, 39, 1126-1138.	2.5	28
34	Multiancestry genome-wide association study of 520,000 subjects identifies 32 loci associated with stroke and stroke subtypes. Nature Genetics, 2018, 50, 524-537.	21.4	1,124
35	Evidence for three genetic loci involved in both anorexia nervosa risk and variation of body mass index. Molecular Psychiatry, 2017, 22, 192-201.	7.9	63
36	Significant Locus and Metabolic Genetic Correlations Revealed in Genome-Wide Association Study of Anorexia Nervosa. American Journal of Psychiatry, 2017, 174, 850-858.	7.2	410

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37	A genome-wide association study of anorexia nervosa suggests a risk locus implicated in dysregulated leptin signaling. Scientific Reports, 2017, 7, 3847.	3.3	23
38	Whole exome sequencing analysis reveals TRPV3 as a risk factor for cardioembolic stroke/subtitle. Thrombosis and Haemostasis, 2016, 116, 1165-1771.	3.4	6
39	Identification of Gene Mutations and Fusion Genes in Patients with SÃ©zary Syndrome. Journal of Investigative Dermatology, 2016, 136, 1490-1499.	0.7	77
40	Loci associated with ischaemic stroke and its subtypes (SiGN): a genome-wide association study. Lancet Neurology, The, 2016, 15, 174-184.	10.2	217
41	Epigenome-wide association study identifiesTXNIPgene associated with type 2 diabetes mellitus and sustained hyperglycemia. Human Molecular Genetics, 2016, 25, 609-619.	2.9	140
42	A family carrying a homozygous LACC1 truncated mutation expands the clinical phenotype of this disease beyond systemic-onset juvenile idiopathic arthritis. Pediatric Rheumatology, 2015, 13, .	2.1	8
43	A decade of structural variants: description, history and methods to detect structural variation. Briefings in Functional Genomics, 2015, 14, 305-314.	2.7	101
44	Using ancestry-informative markers to identify fine structure across 15 populations of European origin. European Journal of Human Genetics, 2014, 22, 1190-1200.	2.8	32
45	Efficient application of next-generation sequencing for the diagnosis of rare genetic syndromes. Journal of Clinical Pathology, 2014, 67, 1099-1103.	2.0	13
46	Whole genome and exome sequencing of monozygotic twins discordant for Crohnâ€™s disease. BMC Genomics, 2014, 15, 564.	2.8	39
47	A genome-wide association study of anorexia nervosa. Molecular Psychiatry, 2014, 19, 1085-1094.	7.9	282
48	Genome-wide analysis of single nucleotide polymorphisms and copy number variants in fibromyalgia suggest a role for the central nervous system. Pain, 2014, 155, 1102-1109.	4.2	54
49	A common 56-kilobase deletion in a primate-specific segmental duplication creates a novel butyrophilin-like protein. BMC Genetics, 2013, 14, 61.	2.7	27
50	The complex SNP and CNV genetic architecture of the increased risk of congenital heart defects in Down syndrome. Genome Research, 2013, 23, 1410-1421.	5.5	65
51	PeSV-Fisher: Identification of Somatic and Non-Somatic Structural Variants Using Next Generation Sequencing Data. PLoS ONE, 2013, 8, e63377.	2.5	17
52	Cluster Analysis of Clinical Data Identifies Fibromyalgia Subgroups. PLoS ONE, 2013, 8, e74873.	2.5	49
53	TTC7B Emerges as a Novel Risk Factor for Ischemic Stroke Through the Convergence of Several Genome-Wide Approaches. Journal of Cerebral Blood Flow and Metabolism, 2012, 32, 1061-1072.	4.3	86
54	Association of Neurexin 3 polymorphisms with smoking behavior. Genes, Brain and Behavior, 2012, 11, 704-711.	2.2	29

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55	MicroRNA profiling of Parkinson's disease brains identifies early downregulation of miR-34b/c which modulate mitochondrial function. Human Molecular Genetics, 2011, 20, 3067-3078.	2.9	433
56	DNA methylation in neurodegenerative disorders: a missing link between genome and environment?. Clinical Genetics, 2011, 80, 1-14.	2.0	53
57	APOE genotype and extent of bleeding and outcome in lobar intracerebral haemorrhage: a genetic association study. Lancet Neurology, The, 2011, 10, 702-709.	10.2	174
58	Deletion of LCE3C and LCE3B is a susceptibility factor for psoriatic arthritis: A study in Spanish and Italian populations and meta-analysis. Arthritis and Rheumatism, 2011, 63, 1860-1865.	6.7	31
59	Novel variants identified in methyl-CpG-binding domain genes in autistic individuals. Neurogenetics, 2010, 11, 291-303.	1.4	67
60	Association and geneâ€“gene interaction of SLC6A4 and ITGB3 in autism. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 477-483.	1.7	41
61	Variants at APOE influence risk of deep and lobar intracerebral hemorrhage. Annals of Neurology, 2010, 68, 934-943.	5.3	241
62	Deletion of the late cornified envelope genes, LCE3C and LCE3B, is associated with rheumatoid arthritis. Arthritis and Rheumatism, 2010, 62, 1246-1251.	6.7	26
63	Genetic Structure of Europeans: A View from the Northâ€“East. PLoS ONE, 2009, 4, e5472.	2.5	279
64	Are MYO1C and MYO1F associated with hearing loss?. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 27-32.	3.8	28
65	Identification of Copy Number Variants Defining Genomic Differences among Major Human Groups. PLoS ONE, 2009, 4, e7230.	2.5	32
66	The emerging role of structural variations in common disorders: initial findings and discovery challenges. Cytogenetic and Genome Research, 2008, 123, 108-117.	1.1	5
67	Mitochondrial 12S rRNA gene mutations affect RNA secondary structure and lead to variable penetrance in hearing impairment. Biochemical and Biophysical Research Communications, 2006, 341, 950-957.	2.1	61
68	A novel G21R mutation of the GJB2 gene causes autosomal dominant non-syndromic congenital deafness in a Cuban family. Genetics and Molecular Biology, 2006, 29, 443-445.	1.3	4
69	Lack of Association Between Autism andSLC25A12. American Journal of Psychiatry, 2006, 163, 929-931.	7.2	36
70	Analysis of the autism chromosome 2 linkage region: GAD1 and other candidate genes. Neuroscience Letters, 2004, 372, 209-214.	2.1	32
71	Connexin mutations in hearing loss, dermatological and neurological disorders. Trends in Molecular Medicine, 2002, 8, 205-212.	6.7	75
72	Mutations in the TMPRSS3 gene are a rare cause of childhood nonsyndromic deafness in Caucasian patients. Journal of Molecular Medicine, 2002, 80, 124-131.	3.9	65

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73	Human connexin26 (GJB2) deafness mutations affect the function of gap junction channels at different levels of protein expression.. Human Genetics, 2002, 111, 190-197.	3.8	86
74	MYO6, the Human Homologue of the Gene Responsible for Deafness in Snellâ€™s Waltzer Mice, Is Mutated in Autosomal Dominant Nonsyndromic Hearing Loss. American Journal of Human Genetics, 2001, 69, 635-640.	6.2	212
75	Connexin 31 (GJB3) is expressed in the peripheral and auditory nerves and causes neuropathy and hearing impairment. Human Molecular Genetics, 2001, 10, 947-952.	2.9	109
76	R32W variant in ConnexinÂ31: mutation or polymorphism for deafness and skin disease?. European Journal of Human Genetics, 2001, 9, 70-70.	2.8	12
77	Identification of seven novel SNPS (five nucleotide and two amino acid substitutions) in the connexin31 (GJB3) gene. Human Mutation, 2000, 15, 481-482.	2.5	17
78	Molecular genetics of hearing impairment due to mutations in gap junction genes encoding beta connexins. Human Mutation, 2000, 16, 190-202.	2.5	197
79	High carrier frequency of the 35delG deafness mutation in European populations. European Journal of Human Genetics, 2000, 8, 19-23.	2.8	363
80	Molecular basis of childhood deafness resulting from mutations in the GJB2 (connexin 26) gene. Human Genetics, 2000, 106, 40-44.	3.8	195
81	Mutations in the Mitochondrial tRNA Ser(UCN) and in the GJB2 (Connexin 26) Gene Are Not Modifiers of the Age at Onset or Severity of Hearing Loss in Spanish Patients with the 12S rRNA A1555G Mutation. American Journal of Human Genetics, 2000, 66, 1465-1467.	6.2	16
82	Mutations in GJB6 cause nonsyndromic autosomal dominant deafness at DFNA3 locus. Nature Genetics, 1999, 23, 16-18.	21.4	345
83	Splice-site mutation in thePDS gene may result in intrafamilial variability for deafness in Pendred syndrome. , 1999, 14, 520-526.		37
84	The A1555G Mutation in the 12S rRNA Gene of Human mtDNA: Recurrent Origins and Founder Events in Families Affected by Sensorineural Deafness. American Journal of Human Genetics, 1999, 65, 1349-1358.	6.2	111
85	Chapter 22: Molecular Basis of Deafness due to Mutations in the Connexin26 Gene (GJB2). Current Topics in Membranes, 1999, , 483-508.	0.9	0
86	Connexin-26 mutations in sporadic and inherited sensorineural deafness. Lancet, The, 1998, 351, 394-398.	13.7	610
87	A Sequence of the CIS Gene Promoter Interacts Preferentially with Two Associated STAT5A Dimers: a Distinct Biochemical Difference between STAT5A and STAT5B. Molecular and Cellular Biology, 1998, 18, 5852-5860.	2.3	148