John B Vincent

List of Publications by Year in descending order

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63 papers	10,732 citations	230014 27 h-index	134545 62 g-index
72 all docs	72 docs citations	72 times ranked	23218 citing authors

#	Article	IF	Citations
1	PTCHD1: Identification and Neurodevelopmental Contributions of an Autism Spectrum Disorder and Intellectual Disability Susceptibility Gene. Genes, 2022, 13, 527.	1.0	7
2	Heterozygous De Novo Truncating Mutation of Nucleolin in an ASD Individual Disrupts Its Nucleolar Localization. Genes, 2022, 13, 51.	1.0	1
3	MeCP2: The Genetic Driver of Rett Syndrome Epigenetics. Frontiers in Genetics, 2021, 12, 620859.	1.1	67
4	Biallelic mutations in the death domain of PIDD1 impair caspase-2 activation and are associated with intellectual disability. Translational Psychiatry, 2021, 11, 1.	2.4	334
5	MeCP2: latest insights fundamentally change our understanding of its interactions with chromatin and its functional attributes. BioEssays, 2021, 43, e2000281.	1.2	4
6	Genome-wide association study of suicidal behaviour severity in mood disorders. World Journal of Biological Psychiatry, 2021, 22, 1-19.	1.3	3
7	Pathogenic variants in PIDD1 lead to an autosomal recessive neurodevelopmental disorder with pachygyria and psychiatric features. European Journal of Human Genetics, 2021, 29, 1226-1234.	1.4	8
8	Characterisation of age and polarity at onset in bipolar disorder. British Journal of Psychiatry, 2021, 219, 659-669.	1.7	20
9	Exome sequencing identifies novel and known mutations in families with intellectual disability. BMC Medical Genomics, 2021, 14, 211.	0.7	5
10	Homozygosity mapping coupled with whole-exome sequencing and protein modelling identified a novel missense mutation in GUCY2D in a consanguineous Pakistani family with Leber congenital amaurosis. Journal of Genetics, 2021, 100, 1.	0.4	1
11	Investigating rare pathogenic/likely pathogenic exonic variation in bipolar disorder. Molecular Psychiatry, 2021, 26, 5239-5250.	4.1	15
12	Biallelic inheritance in a single Pakistani family with intellectual disability implicates new candidate gene RDH14. Scientific Reports, 2021, 11, 23113.	1.6	2
13	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	1.1	24
14	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	9.4	96
15	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	2.6	32
16	A novel biallelic single base insertion in WNK1 in a Pakistani family with congenital insensitivity to pain. Journal of Human Genetics, 2020, 65, 493-496.	1.1	0
17	MeCP2-E1 isoform is a dynamically expressed, weakly DNA-bound protein with different protein and DNA interactions compared to MeCP2-E2. Epigenetics and Chromatin, 2019, 12, 63.	1.8	50
18	GPT2 mutations in autosomal recessive developmental disability: extending the clinical phenotype and population prevalence estimates. Human Genetics, 2019, 138, 1183-1200.	1.8	6

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19	Ptchd1 exon3 truncating mutations recapitulate more clinically relevant autistic-like traits in mice. IBRO Reports, 2019, 6, S507.	0.3	1
20	Genome-wide association study identifies 30 loci associated with bipolar disorder. Nature Genetics, 2019, 51, 793-803.	9.4	1,191
21	Genetic studies of multiple consanguineous Pakistani families segregating oculocutaneous albinism identified novel and reported mutations. Annals of Human Genetics, 2019, 83, 278-284.	0.3	8
22	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. Cell, 2019, 179, 1469-1482.e11.	13.5	935
23	MeCP2 AT-Hook1 mutations in patients with intellectual disability and/or schizophrenia disrupt DNA binding and chromatin compaction in vitro. Human Mutation, 2018, 39, 717-728.	1.1	16
24	Three Mutations in the Bilateral Frontoparietal Polymicrogyria Gene GPR56 in Pakistani Intellectual Disability Families. Journal of Pediatric Genetics, 2018, 07, 060-066.	0.3	12
25	Sequence Analysis of Drug Target Genes with Suicidal Behavior in Bipolar Disorder Patients. Molecular Neuropsychiatry, 2018, 4, 1-6.	3.0	3
26	Biallelic missense variants in ZBTB11 can cause intellectual disability in humans. Human Molecular Genetics, 2018, 27, 3177-3188.	1.4	19
27	Investigation of correlations between DNA methylation, suicidal behavior and aging. Bipolar Disorders, 2017, 19, 32-40.	1.1	27
28	The Use of Next-Generation Sequencing for Research and Diagnostics for Intellectual Disability. Cold Spring Harbor Perspectives in Medicine, 2017, 7, a026864.	2.9	53
29	MeCP2_E1 N-terminal modifications affect its degradation rate and are disrupted by the Ala2Val Rett mutation. Human Molecular Genetics, 2017, 26, 4132-4141.	1.4	23
30	Mutations in the genes for thyroglobulin and thyroid peroxidase cause thyroid dyshormonogenesis and autosomal-recessive intellectual disability. Journal of Human Genetics, 2016, 61, 867-872.	1.1	15
31	From Function to Phenotype: Impaired DNA Binding and Clustering Correlates with Clinical Severity in Males with Missense Mutations in MECP2. Scientific Reports, 2016, 6, 38590.	1.6	21
32	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	9.4	2,421
33	Unstable repeat expansion in major psychiatric disorders. Psychiatric Genetics, 2016, 26, 156-165.	0.6	8
34	15q11.2 Duplication Encompassing Only theUBE3AGene Is Associated with Developmental Delay and Neuropsychiatric Phenotypes. Human Mutation, 2015, 36, 689-693.	1.1	67
35	New recessive truncating mutation in LTBP3 in a family with oligodontia, short stature, and mitral valve prolapse. American Journal of Medical Genetics, Part A, 2015, 167, 1396-1399.	0.7	23
36	Homozygosity mapping of autosomal recessive intellectual disability loci in 11 consanguineous Pakistani families. Acta Neuropsychiatrica, 2015, 27, 38-47.	1.0	2

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37	Novel VPS13B Mutations in Three Large Pakistani Cohen Syndrome Families Suggests a Baloch Variant with Autistic-Like Features. BMC Medical Genetics, 2015, 16, 41.	2.1	23
38	Mutations in DCPS and EDC3 in autosomal recessive intellectual disability indicate a crucial role for mRNA decapping in neurodevelopment. Human Molecular Genetics, 2015, 24, 3172-3180.	1.4	40
39	Mutations in the histamineN-methyltransferase gene,HNMT, are associated with nonsyndromic autosomal recessive intellectual disability. Human Molecular Genetics, 2015, 24, 5697-5710.	1.4	27
40	A genome-wide association study of suicide severity scores in bipolar disorder. Journal of Psychiatric Research, 2015, 65, 23-29.	1.5	36
41	ISDN2014_0146: REMOVED: MeCP2â€chromatin binding dynamics shows a direct genotypeâ€“phenotype correlation in males with single amino acid substitutions. International Journal of Developmental Neuroscience, 2015, 47, 42-42.	0.7	0
42	Investigation of the genetic interaction between BDNF and DRD3 genes in suicidal behaviour in psychiatric disorders. World Journal of Biological Psychiatry, 2015, 16, 171-179.	1.3	14
43	Over-Expression of Either MECP2_e1 or MECP2_e2 in Neuronally Differentiated Cells Results in Different Patterns of Gene Expression. PLoS ONE, 2014, 9, e91742.	1.1	16
44	Biallelic Truncating Mutations in FMN2, Encoding the Actin-Regulatory Protein Formin 2, Cause Nonsyndromic Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2014, 95, 721-728.	2.6	62
45	Disruption of the methyltransferase-like 23 gene METTL23 causes mild autosomal recessive intellectual disability. Human Molecular Genetics, 2014, 23, 4015-4023.	1.4	32
46	Copy number variant study of bipolar disorder in Canadian and UK populations implicates synaptic genes. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 303-313.	1.1	76
47	Genome-wide association study of bipolar disorder in Canadian and UK populations corroborates disease loci including SYNE1 and CSMD1. BMC Medical Genetics, 2014, 15, 2.	2.1	106
48	Truncation of the E3 ubiquitin ligase component FBXO31 causes non-syndromic autosomal recessive intellectual disability in a Pakistani family. Human Genetics, 2014, 133, 975-984.	1.8	24
49	Mice with an isoform-ablating Mecp2 exon 1 mutation recapitulate the neurologic deficits of Rett syndrome. Human Molecular Genetics, 2014, 23, 2447-2458.	1.4	63
50	A missense mutation in the PISA domain of HsSAS-6 causes autosomal recessive primary microcephaly in a large consanguineous Pakistani family. Human Molecular Genetics, 2014, 23, 5940-5949.	1.4	63
51	A synonymous change, p.Gly16Gly in MECP2 Exon 1, causes a cryptic splice event in a Rett syndrome patient. Orphanet Journal of Rare Diseases, 2013, 8, 108.	1.2	23
52	Admixture analysis of age at onset in bipolar disorder. Psychiatry Research, 2011, 185, 27-32.	1.7	51
53	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	13.7	1,803
54	Meta-analysis of genome-wide association data identifies a risk locus for major mood disorders on 3p21.1. Nature Genetics, 2010, 42, 128-131.	9.4	152

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55	Association analysis of DAOA and DAO in bipolar disorder: results from two independent caseâ€control studies. Bipolar Disorders, 2010, 12, 579-581.	1.1	9
56	Disruption at the PTCHD1 Locus on Xp22.11 in Autism Spectrum Disorder and Intellectual Disability. Science Translational Medicine, 2010, 2, 49ra68.	5.8	178
57	Genome-wide association and meta-analysis of bipolar disorder in individuals of European ancestry. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 7501-7506.	3.3	274
58	Novel exon 1 mutations in MECP2 implicate isoform MeCP2_e1 in classical Rett syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 1019-1023.	0.7	44
59	Oligodontia Is Caused by Mutation in LTBP3, the Gene Encoding Latent TGF-Î² Binding Protein 3. American Journal of Human Genetics, 2009, 84, 519-523.	2.6	79
60	Structural Variation of Chromosomes in Autism Spectrum Disorder. American Journal of Human Genetics, 2008, 82, 477-488.	2.6	1,641
61	Sequence variants within exon 1 ofMECP2 occur in females with mental retardation. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 355-360.	1.1	21
62	A previously unidentified MECP2 open reading frame defines a new protein isoform relevant to Rett syndrome. Nature Genetics, 2004, 36, 339-341.	9.4	290
63	Long repeat tracts atSCA8 in major psychosis. American Journal of Medical Genetics Part A, 2000, 96, 873-876.	2.4	55