Rami Abou Jamra

List of Publications by Year
in descending order

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115 papers	5,454 citations	87888 38 h-index	98798 67 g-index
130 all docs	130 docs citations	130 times ranked	11044 citing authors

#	Article	IF	Citations
1	Clinical exome sequencing: results from 2819 samples reflecting 1000 families. European Journal of Human Genetics, 2017, 25, 176-182.	2.8	291
2	Evidence for a Relationship Between Genetic Variants at the Brain-Derived Neurotrophic Factor (BDNF) Locus and Major Depression. Biological Psychiatry, 2005, 58, 307-314.	1.3	284
3	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	21.4	230
4	Combined Analysis from Eleven Linkage Studies of Bipolar Disorder Provides Strong Evidence of Susceptibility Loci on Chromosomes 6q and 8q. American Journal of Human Genetics, 2005, 77, 582-595.	6.2	218
5	Adaptor Protein Complex 4 Deficiency Causes Severe Autosomal-Recessive Intellectual Disability, Progressive Spastic Paraplegia, Shy Character, and Short Stature. American Journal of Human Genetics, 2011, 88, 788-795.	6.2	206
6	Genetic Variation in the Human Androgen Receptor Gene Is the Major Determinant of Common Early-Onset Androgenetic Alopecia. American Journal of Human Genetics, 2005, 77, 140-148.	6.2	198
7	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	3.2	190
8	Diagnostic Yield and Novel Candidate Genes by Exome Sequencing in 152 Consanguineous Families With Neurodevelopmental Disorders. JAMA Psychiatry, 2017, 74, 293.	11.0	186
9	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. American Journal of Human Genetics, 2015, 97, 886-893.	6.2	171
10	NEK1 Mutations Cause Short-Rib Polydactyly Syndrome Type Majewski. American Journal of Human Genetics, 2011, 88, 106-114.	6.2	151
11	GRIN2A-related disorders: genotype and functional consequence predict phenotype. Brain, 2019, 142, 80-92.	7.6	143
12	Genotype-Phenotype Studies in Bipolar Disorder Showing Association Between the DAOA/G30 Locus and Persecutory Delusions: A First Step Toward a Molecular Genetic Classification of Psychiatric Phenotypes. American Journal of Psychiatry, 2005, 162, 2101-2108.	7.2	123
13	Brain-specific tryptophan hydroxylase 2 (TPH2): a functional Pro206Ser substitution and variation in the 5'-region are associated with bipolar affective disorder. Human Molecular Genetics, 2007, 17, 87-97.	2.9	109
14	A Peroxisomal Disorder of Severe Intellectual Disability, Epilepsy, and Cataracts Due to Fatty Acyl-CoA Reductase 1 Deficiency. American Journal of Human Genetics, 2014, 95, 602-610.	6.2	106
15	Loss of Function of GALNT2 Lowers High-Density Lipoproteins in Humans, Nonhuman Primates, and Rodents. Cell Metabolism, 2016, 24, 234-245.	16.2	103
16	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. Genetics in Medicine, 2018, 20, 630-638.	2.4	101
17	Variants in PUS7 Cause Intellectual Disability with Speech Delay, Microcephaly, Short Stature, and Aggressive Behavior. American Journal of Human Genetics, 2018, 103, 1045-1052.	6.2	89
18	Hypomorphic Mutations in PGAP2, Encoding a GPI-Anchor-Remodeling Protein, Cause Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2013, 92, 575-583.	6.2	87

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19	The DISC locus and schizophrenia: evidence from an association study in a central European sample and from a meta-analysis across different European populations. Human Molecular Genetics, 2009, 18, 2719-2727.	2.9	78
20	AMPA-receptor specific biogenesis complexes control synaptic transmission and intellectual ability. Nature Communications, 2017, 8, 15910.	12.8	77
21	FOXG1 syndrome: genotypeâ€“phenotype association in 83 patients with FOXG1 variants. Genetics in Medicine, 2018, 20, 98-108.	2.4	77
22	Null Mutation in PGAP1 Impairing Gpi-Anchor Maturation in Patients with Intellectual Disability and Encephalopathy. PLoS Genetics, 2014, 10, e1004320.	3.5	72
23	Biallelic loss of human CTNNA2, encoding Î±N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. Nature Genetics, 2018, 50, 1093-1101.	21.4	70
24	Mutations in MBOAT7 , Encoding Lysophosphatidylinositol Acyltransferase I, Lead to Intellectual Disability Accompanied by Epilepsy and Autistic Features. American Journal of Human Genetics, 2016, 99, 912-916.	6.2	69
25	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. American Journal of Human Genetics, 2019, 104, 815-834.	6.2	59
26	Bi-allelic Variants in METTL5 Cause Autosomal-Recessive Intellectual Disability and Microcephaly. American Journal of Human Genetics, 2019, 105, 869-878.	6.2	58
27	A comprehensive global genotypeâ€“phenotype database for rare diseases. Molecular Genetics & Genomic Medicine, 2017, 5, 66-75.	1.2	57
28	Genomewide Scan and Fine-Mapping Linkage Studies in Four European Samples with Bipolar Affective Disorder Suggest a New Susceptibility Locus on Chromosome 1p35-p36 and Provides Further Evidence of Loci on Chromosome 4q31 and 6q24. American Journal of Human Genetics, 2005, 77, 1102-1111.	6.2	56
29	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
30	Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. Scientific Reports, 2017, 7, 12225.	3.3	53
31	De Novo Variants in GRIA4 Lead to Intellectual Disability with or without Seizures and Gait Abnormalities. American Journal of Human Genetics, 2017, 101, 1013-1020.	6.2	53
32	TALPID3 controls centrosome and cell polarity and the human ortholog KIAA0586 is mutated in Joubert syndrome (JBTS23). ELife, 2015, 4, .	6.0	51
33	G72and Its Association With Major Depression and Neuroticism in Large Population-Based Groups From Germany. American Journal of Psychiatry, 2008, 165, 753-762.	7.2	50
34	Variation in P2RX7 candidate gene (rs2230912) is not associated with bipolar I disorder and unipolar major depression in four European samples. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 1017-1021.	1.7	50
35	The First Genomewide Interaction and Locus-Heterogeneity Linkage Scan in Bipolar Affective Disorder: Strong Evidence of Epistatic Effects between Loci on Chromosomes 2q and 6q. American Journal of Human Genetics, 2007, 81, 974-986.	6.2	49
36	Value of renal gene panel diagnostics in adults waiting for kidney transplantation due to undetermined end-stage renal disease. Kidney International, 2019, 96, 222-230.	5.2	47

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37	Genes and Schizophrenia: The G72/G30 Gene Locus in Psychiatric Disorders: A Challenge to Diagnostic Boundaries?. Schizophrenia Bulletin, 2005, 32, 599-608.	4.3	46
38	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. Brain, 2020, 143, 1114-1126.	7.6	46
39	Autosomal-Recessive Mutations in the tRNA Splicing Endonuclease Subunit TSEN15 Cause Pontocerebellar Hypoplasia and Progressive Microcephaly. American Journal of Human Genetics, 2016, 99, 228-235.	6.2	44
40	De Novo Variants in MAPK8IP3 Cause Intellectual Disability with Variable Brain Anomalies. American Journal of Human Genetics, 2019, 104, 203-212.	6.2	44
41	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
42	Germline AGO2 mutations impair RNA interference and human neurological development. Nature Communications, 2020, 11, 5797.	12.8	43
43	PUF60 variants cause a syndrome of ID, short stature, microcephaly, coloboma, craniofacial, cardiac, renal and spinal features. European Journal of Human Genetics, 2017, 25, 552-559.	2.8	42
44	No Association Between the Putative Functional ZDHHC8 Single Nucleotide Polymorphism rs175174 and Schizophrenia in Large European Samples. Biological Psychiatry, 2005, 58, 78-80.	1.3	41
45	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.	2.9	40
46	Possible association between genetic variants at the GRIN1 gene and schizophrenia with lifetime history of depressive symptoms in a German sample. Psychiatric Genetics, 2007, 17, 308-310.	1.1	36
47	NDST1 missense mutations in autosomal recessive intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 2753-2763.	1.2	34
48	Identification of a Loss-of-Function Mutation in the Context of Glutaminase Deficiency and Neonatal Epileptic Encephalopathy. JAMA Neurology, 2019, 76, 342.	9.0	33
49	Monoamine related functional gene variants and relationships to monoamine metabolite concentrations in CSF of healthy volunteers. BMC Psychiatry, 2004, 4, 4.	2.6	32
50	Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. Brain, 2019, 142, 2617-2630.	7.6	31
51	The genetic landscape of intellectual disability and epilepsy in adults and the elderly: a systematic genetic work-up of 150 individuals. Genetics in Medicine, 2021, 23, 1492-1497.	2.4	31
52	Biallelic Mutations in TMTC3, Encoding a Transmembrane and TPR-Containing Protein, Lead to Cobblestone Lissencephaly. American Journal of Human Genetics, 2016, 99, 1181-1189.	6.2	30
53	De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. Brain, 2019, 142, 3351-3359.	7.6	29
54	Genetics of autosomal recessive intellectual disability. Medizinische Genetik, 2018, 30, 323-327.	0.2	28

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55	The Role of Periodontal Ligament Cells in Delayed Tooth Eruption in Patients with Cleidocranial Dysostosis*. Journal of Orofacial Orthopedics, 2009, 70, 495-510.	1.3	27
56	Hypomorphic Pathogenic Variants in TAF13 Are Associated with Autosomal-Recessive Intellectual Disability and Microcephaly. American Journal of Human Genetics, 2017, 100, 555-561.	6.2	26
57	Next-generation sequencing of 32 genes associated with hereditary aortopathies and related disorders of connective tissue in a cohort of 199 patients. Genetics in Medicine, 2019, 21, 1832-1841.	2.4	26
58	Biallelic COL3A1 mutations result in a clinical spectrum of specific structural brain anomalies and connective tissue abnormalities. American Journal of Medical Genetics, Part A, 2017, 173, 2534-2538.	1.2	25
59	Mutations in the mitochondrial gene C12ORF65 lead to syndromic autosomal recessive intellectual disability and show genotype phenotype correlation. European Journal of Medical Genetics, 2013, 56, 599-602.	1.3	24
60	Moodâ€incongruent psychosis in bipolar disorder: conditional linkage analysis shows genomeâ€wide suggestive linkage at 1q32.3, 7p13 and 20q13.31. Bipolar Disorders, 2009, 11, 610-620.	1.9	23
61	Broadening the phenotypic spectrum of pathogenic LARP7 variants: two cases with intellectual disability, variable growth retardation and distinct facial features. Journal of Human Genetics, 2016, 61, 229-233.	2.3	23
62	Haploinsufficiency of CUX1 Causes Nonsyndromic Global Developmental Delay With Possible Catchâ€up Development. Annals of Neurology, 2018, 84, 200-207.	5.3	23
63	Loss of function of SVBP leads to autosomal recessive intellectual disability, microcephaly, ataxia, and hypotonia. Genetics in Medicine, 2019, 21, 1790-1796.	2.4	23
64	Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. European Journal of Human Genetics, 2018, 26, 695-708.	2.8	22
65	Exome first approach to reduce diagnostic costs and time â€“ retrospective analysis of 111 individuals with rare neurodevelopmental disorders. European Journal of Human Genetics, 2022, 30, 117-125.	2.8	22
66	The catechol-O-methyl transferase (COMT) gene and its potential association with schizophrenia: Findings from a large German case-control and family-based sample. Schizophrenia Research, 2010, 122, 24-30.	2.0	21
67	SPATA5 mutations cause a distinct autosomal recessive phenotype of intellectual disability, hypotonia and hearing loss. Orphanet Journal of Rare Diseases, 2016, 11, 130.	2.7	19
68	Genetic basis of neurodevelopmental disorders in 103 Jordanian families. Clinical Genetics, 2020, 97, 621-627.	2.0	19
69	Inhibition of RAS Activation Due to a Homozygous Ezrin Variant in Patients with Profound Intellectual Disability. Human Mutation, 2015, 36, 270-278.	2.5	18
70	Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776.	2.5	18
71	A reappraisal of the association between Dysbindin (DTNBP1) and schizophrenia in a large combined caseâ€control and family-based sample of German ancestry. Schizophrenia Research, 2010, 118, 98-105.	2.0	17
72	Identification of a novel leptin receptor (LEPR) variant and proof of functional relevance directing treatment decisions in patients with morbid obesity. Metabolism: Clinical and Experimental, 2021, 116, 154438.	3.4	17

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73	No evidence for DUP25 in patients with panic disorder using a quantitative real-time PCR approach. Human Genetics, 2003, 114, 115-117.	3.8	16
74	European collaborative study of earlyâ€onset bipolar disorder: Evidence for genetic heterogeneity on 2q14 according to age at onset. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 1425-1433.	1.7	16
75	A new p.(Ile66Serfs*93) IGF2 variant is associated with pre- and postnatal growth retardation. European Journal of Endocrinology, 2019, 180, K1-K13.	3.7	16
76	Biallelic GRM7 variants cause epilepsy, microcephaly, and cerebral atrophy. Annals of Clinical and Translational Neurology, 2020, 7, 610-627.	3.7	15
77	A new susceptibility locus for bipolar affective disorder in PAR1 on Xp22.3/Yp11.3. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 1110-1114.	1.7	14
78	Association study of a functional promoter polymorphism in theXBP1 gene and schizophrenia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 71-75.	1.7	13
79	Recurrent null mutation in SPG20 leads to Troyer syndrome. Molecular and Cellular Probes, 2015, 29, 315-318.	2.1	13
80	Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. European Journal of Human Genetics, 2021, 29, 411-421.	2.8	13
81	MAN1B1 Mutation Leads to a Recognizable Phenotype: A Case Report and Future Prospects. Molecular Syndromology, 2015, 6, 58-62.	0.8	12
82	Association study between genetic variants at the PIP5K2A gene locus and schizophrenia and bipolar affective disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 663-665.	1.7	11
83	Evolutionary conserved networks of human height identify multiple Mendelian causes of short stature. European Journal of Human Genetics, 2019, 27, 1061-1071.	2.8	11
84	De novo mutations in the X-linked TFE3 gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. Journal of Medical Genetics, 2020, 57, 808-819.	3.2	11
85	De novo variants in ATP2B1 lead to neurodevelopmental delay. American Journal of Human Genetics, 2022, 109, 944-952.	6.2	11
86	Association study between genetic variants at the VAMP2 and VAMP3 loci and bipolar affective disorder. Psychiatric Genetics, 2008, 18, 199-203.	1.1	10
87	Possible association of different G72/G30 SNPs with mood episodes and persecutory delusions in bipolar I Romanian patients. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2010, 34, 657-663.	4.8	10
88	No association between the serine racemase gene (SRR) and schizophrenia in a German caseâ€“control sample. Psychiatric Genetics, 2007, 17, 125.	1.1	9
89	Bi-allelic loss of function variants in SLC30A5 as cause of perinatal lethal cardiomyopathy. European Journal of Human Genetics, 2021, 29, 808-815.	2.8	9
90	No evidence for an association between variants at the proline dehydrogenase locus and schizophrenia or bipolar affective disorder. Psychiatric Genetics, 2005, 15, 195-198.	1.1	8

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91	Novel EXOSC3 pathogenic variant results in a mild course of neurologic disease with cerebellum involvement. European Journal of Medical Genetics, 2020, 63, 103649.	1.3	7
92	No association between genetic variants at the ASCT1 gene and schizophrenia or bipolar disorder in a German sample. Psychiatric Genetics, 2006, 16, 233-234.	1.1	6
93	No evidence for an association between variants at the Î³-amino-n-butyric acid type A receptor Î²2 locus and schizophrenia. Psychiatric Genetics, 2007, 17, 43-45.	1.1	6
94	A recessive form of extreme macrocephaly and mild intellectual disability complements the spectrum of PTEN hamartoma tumour syndrome. European Journal of Human Genetics, 2016, 24, 889-894.	2.8	6
95	Family-based association studies of Î±-adrenergic receptor genes in chromosomal regions with linkage to bipolar affective disorder. , 2004, 126B, 79-81.		5
96	No association between genetic variants at the GLYT2 gene and bipolar affective disorder and schizophrenia. Psychiatric Genetics, 2006, 16, 91.	1.1	5
97	A systematic association mapping on chromosome 6q in bipolar affective disorderâ€"evidence for the melaninâ€concentratingâ€hormoneâ€receptorâ€2 gene as a risk factor for bipolar affective disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 878-884.	1.7	5
98	No association between genetic variants at the DGCR2 gene and schizophrenia in a German sample. Psychiatric Genetics, 2009, 19, 104.	1.1	5
99	Association study of the GRIA1 and CLINT1 (Epsin 4) genes in a German schizophrenia sample. Psychiatric Genetics, 2011, 21, 114.	1.1	5
100	Integrative bioinformatics analysis characterizing the role of EDC3 in mRNA decay and its association to intellectual disability. BMC Medical Genomics, 2018, 11, 41.	1.5	5
101	Defining and expanding the phenotype of QARS-associated developmental epileptic encephalopathy. Neurology: Genetics, 2019, 5, e373.	1.9	5
102	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. Orphanet Journal of Rare Diseases, 2021, 16, 136.	2.7	5
103	A new missense mutation in PLA2G6 gene among a family with infantile neuroaxonal dystrophy INAD. The Gazette of the Egyptian Paediatric Association, 2016, 64, 171-176.	0.4	4
104	Mutations of familial Mediterranean fever in Syrian patients and controls: Evidence for high carrier rate. Gene Reports, 2017, 6, 87-92.	0.8	4
105	Congenital cervical spine malformation due to biâ€allelic <scp>RIPPLY2</scp> variants in spondylocostal dysostosis type 6. Clinical Genetics, 2021, 99, 565-571.	2.0	4
106	De novo variants in the PABP domain of PABPC1 lead to developmental delay. Genetics in Medicine, 2022, , .	2.4	4
107	Prenatal phenotype of PNKP-related primary microcephaly associated with variants affecting both the FHA and phosphatase domain. European Journal of Human Genetics, 2022, 30, 101-110.	2.8	3
108	Bi-allelic variants inCHKAcause a neurodevelopmental disorder with epilepsy and microcephaly. Brain, 2022, 145, 1916-1923.	7.6	3

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109	Hypochondroplasia and temporal lobe epilepsy â€“ A series of 4 cases. Epilepsy and Behavior, 2022, 126, 108479.	1.7	2
110	Brief Report: No Association Between Premorbid Adjustment in Adult-Onset Schizophrenia and Genetic Variation in Dysbindin. Journal of Autism and Developmental Disorders, 2008, 38, 1977-1981.	2.7	1
111	No association between the D-aspartate oxidase locus and schizophrenia. Psychiatric Genetics, 2009, 19, 56.	1.1	1
112	Association study of 20 genetic variants at the D-amino acid oxidase gene in schizophrenia. Psychiatric Genetics, 2010, 20, 82-83.	1.1	1
113	No association between the serine racemase gene (SRR) and bipolar disorder in a German caseâ€“control sample. Psychiatric Genetics, 2007, 17, 127.	1.1	0
114	Smooth velvety hyperextensible skin in a young patient. JDDG - Journal of the German Society of Dermatology, 2018, 16, 504-507.	0.8	0
115	In cis TP53 and RAD51C pathogenic variants may predispose to sebaceous gland carcinomas. European Journal of Human Genetics, 2021, 29, 489-494.	2.8	0