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	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
2	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
3	Hypochondroplasia and temporal lobe epilepsy â€“ A series of 4 cases. Epilepsy and Behavior, 2022, 126, 108479.	1.7	2
4	Bi-allelic variants inCHKAcause a neurodevelopmental disorder with epilepsy and microcephaly. Brain, 2022, 145, 1916-1923.	7.6	3
5	De novo variants in ATP2B1 lead to neurodevelopmental delay. American Journal of Human Genetics, 2022, 109, 944-952.	6.2	11
6	De novo variants in the PABP domain of PABPC1 lead to developmental delay. Genetics in Medicine, 2022, , .	2.4	4
7	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
8	Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. European Journal of Human Genetics, 2021, 29, 411-421.	2.8	13
9	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
10	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
11	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. Orphanet Journal of Rare Diseases, 2021, 16, 136.	2.7	5
12	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
13	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
14	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
15	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
16	Germline AGO2 mutations impair RNA interference and human neurological development. Nature Communications, 2020, 11, 5797.	12.8	43
17	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
18	Genetic basis of neurodevelopmental disorders in 103 Jordanian families. Clinical Genetics, 2020, 97, 621-627.	2.0	19

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19	Biallelic GRM7 variants cause epilepsy, microcephaly, and cerebral atrophy. Annals of Clinical and Translational Neurology, 2020, 7, 610-627.	3.7	15
20	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. Brain, 2020, 143, 1114-1126.	7.6	46
21	Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. Brain, 2019, 142, 2617-2630.	7.6	31
22	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
23	De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. Brain, 2019, 142, 3351-3359.	7.6	29
24	Bi-allelic Variants in METTL5 Cause Autosomal-Recessive Intellectual Disability and Microcephaly. American Journal of Human Genetics, 2019, 105, 869-878.	6.2	58
25	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
26	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
27	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
28	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
29	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
30	Defining and expanding the phenotype of QARS-associated developmental epileptic encephalopathy. Neurology: Genetics, 2019, 5, e373.	1.9	5
31	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
32	GRIN2A-related disorders: genotype and functional consequence predict phenotype. Brain, 2019, 142, 80-92.	7.6	143
33	De Novo Variants in MAPK8IP3 Cause Intellectual Disability with Variable Brain Anomalies. American Journal of Human Genetics, 2019, 104, 203-212.	6.2	44
34	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
35	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
36	Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. European Journal of Human Genetics, 2018, 26, 695-708.	2.8	22

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37	Smooth velvety hyperextensible skin in a young patient. JDDG - Journal of the German Society of Dermatology, 2018, 16, 504-507.	0.8	0
38	FOXG1 syndrome: genotypeâ€“phenotype association in 83 patients with FOXG1 variants. Genetics in Medicine, 2018, 20, 98-108.	2.4	77
39	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. Genetics in Medicine, 2018, 20, 630-638.	2.4	101
40	Genetics of autosomal recessive intellectual disability. Medizinische Genetik, 2018, 30, 323-327.	0.2	28
41	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
42	De novo variants in neurodevelopmental disorders with epilepsy. Nature Genetics, 2018, 50, 1048-1053.	21.4	230
43	Haploinsufficiency of CUX1 Causes Nonsyndromic Global Developmental Delay With Possible Catchâ€up Development. Annals of Neurology, 2018, 84, 200-207.	5.3	23
44	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
45	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
46	Diagnostic Yield and Novel Candidate Genes by Exome Sequencing in 152 Consanguineous Families With Neurodevelopmental Disorders. JAMA Psychiatry, 2017, 74, 293.	11.0	186
47	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
48	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
49	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	3.2	190
50	A comprehensive global genotypeâ€“phenotype database for rare diseases. Molecular Genetics & Genomic Medicine, 2017, 5, 66-75.	1.2	57
51	Mutations of familial Mediterranean fever in Syrian patients and controls: Evidence for high carrier rate. Gene Reports, 2017, 6, 87-92.	0.8	4
52	Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. Scientific Reports, 2017, 7, 12225.	3.3	53
53	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
54	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

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55	AMPA-receptor specific biogenesis complexes control synaptic transmission and intellectual ability. Nature Communications, 2017, 8, 15910.	12.8	77
56	Clinical exome sequencing: results from 2819 samples reflecting 1000 families. European Journal of Human Genetics, 2017, 25, 176-182.	2.8	291
57	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
58	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
59	Loss of Function of GALNT2 Lowers High-Density Lipoproteins in Humans, Nonhuman Primates, and Rodents. Cell Metabolism, 2016, 24, 234-245.	16.2	103
60	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
61	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
62	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
63	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
64	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
65	MAN1B1 Mutation Leads to a Recognizable Phenotype: A Case Report and Future Prospects. Molecular Syndromology, 2015, 6, 58-62.	0.8	12
66	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
67	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
68	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
69	Recurrent null mutation in SPG20 leads to Troyer syndrome. Molecular and Cellular Probes, 2015, 29, 315-318.	2.1	13
70	TALPID3 controls centrosome and cell polarity and the human ortholog KIAA0586 is mutated in Joubert syndrome (JBTS23). ELife, 2015, 4, .	6.0	51
71	Null Mutation in PGAP1 Impairing Gpi-Anchor Maturation in Patients with Intellectual Disability and Encephalopathy. PLoS Genetics, 2014, 10, e1004320.	3.5	72
72	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

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73	NDST1 missense mutations in autosomal recessive intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 2753-2763.	1.2	34
74	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
75	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
76	Association study of the GRIA1 and CLINT1 (Epsin 4) genes in a German schizophrenia sample. Psychiatric Genetics, 2011, 21, 114.	1.1	5
77	NEK1 Mutations Cause Short-Rib Polydactyly Syndrome Type Majewski. American Journal of Human Genetics, 2011, 88, 106-114.	6.2	151
78	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
79	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
80	Association study of 20 genetic variants at the D-amino acid oxidase gene in schizophrenia. Psychiatric Genetics, 2010, 20, 82-83.	1.1	1
81	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
82	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
83	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
84	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
85	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
86	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
87	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
88	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
89	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
90	No association between genetic variants at the DGCR2 gene and schizophrenia in a German sample. Psychiatric Genetics, 2009, 19, 104.	1.1	5

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91	No association between the D-aspartate oxidase locus and schizophrenia. Psychiatric Genetics, 2009, 19, 56.	1.1	1
92	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
93	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
94	Association study between genetic variants at the VAMP2 and VAMP3 loci and bipolar affective disorder. Psychiatric Genetics, 2008, 18, 199-203.	1.1	10
95	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
96	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
97	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
98	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
99	No association between the serine racemase gene (SRR) and schizophrenia in a German caseâ€“control sample. Psychiatric Genetics, 2007, 17, 125.	1.1	9
100	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
101	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
102	No association between genetic variants at the GLYT2 gene and bipolar affective disorder and schizophrenia. Psychiatric Genetics, 2006, 16, 91.	1.1	5
103	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
104	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
105	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
106	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
107	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
108	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

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109	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
110	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
111	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
112	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
113	Monoamine related functional gene variants and relationships to monoamine metabolite concentrations in CSF of healthy volunteers. BMC Psychiatry, 2004, 4, 4.	2.6	32
114	Family-based association studies of Î±-adrenergic receptor genes in chromosomal regions with linkage to bipolar affective disorder. , 2004, 126B, 79-81.		5
115	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