Jamel Chelly

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2845080/publications.pdf

Version: 2024-02-01

245 papers	21,535 citations	8159 76 h-index	11030 137 g-index
252 all docs	252 docs citations	252 times ranked	19803 citing authors

#	Article	IF	Citations
1	Doublecortin Is a Developmentally Regulated, Microtubule-Associated Protein Expressed in Migrating and Differentiating Neurons. Neuron, 1999, 23, 247-256.	3.8	936
2	Transcription of the dystrophin gene in human muscle and non-muscle tissues. Nature, 1988, 333, 858-860.	13.7	760
3	A Novel CNS Gene Required for Neuronal Migration and Involved in X-Linked Subcortical Laminar Heterotopia and Lissencephaly Syndrome. Cell, 1998, 92, 51-61.	13.5	729
4	Isolation of a candidate gene for Menkes disease that encodes a potential heavy metal binding protein. Nature Genetics, 1993, 3, 14-19.	9.4	708
5	X-Linked Mental Retardation and Autism Are Associated with a Mutation in the NLGN4 Gene, a Member of the Neuroligin Family. American Journal of Human Genetics, 2004, 74, 552-557.	2.6	686
6	A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. Nature Genetics, 2009, 41, 535-543.	9.4	528
7	Oligophrenin-1 encodes a rhoGAP protein involved in X-linked mental retardation. Nature, 1998, 392, 923-926.	13.7	412
8	Mutations in TUBG1, DYNC1H1, KIF5C and KIF2A cause malformations of cortical development and microcephaly. Nature Genetics, 2013, 45, 639-647.	9.4	399
9	Mutations in Î±-Tubulin Cause Abnormal Neuronal Migration in Mice and Lissencephaly in Humans. Cell, 2007, 128, 45-57.	13.5	397
10	Mutations in the JARID1C Gene, Which Is Involved in Transcriptional Regulation and Chromatin Remodeling, Cause X-Linked Mental Retardation. American Journal of Human Genetics, 2005, 76, 227-236.	2.6	349
11	Differentially Activated Macrophages Orchestrate Myogenic Precursor Cell Fate During Human Skeletal Muscle Regeneration. Stem Cells, 2013, 31, 384-396.	1.4	343
12	Mutations in the Î²-tubulin gene TUBB2B result in asymmetrical polymicrogyria. Nature Genetics, 2009, 41, 746-752.	9.4	330
13	Mutations in ARHGEF6, encoding a guanine nucleotide exchange factor for Rho GTPases, in patients with X-linked mental retardation. Nature Genetics, 2000, 26, 247-250.	9.4	329
14	Genotype-phenotype analysis in 2,405 patients with a dystrophinopathy using the UMD-DMD database: a model of nationwide knowledgebase. Human Mutation, 2009, 30, 934-945.	1.1	309
15	Mutations in GDI1 are responsible for X-linked non-specific mental retardation. Nature Genetics, 1998, 19, 134-139.	9.4	304
16	Molecular genetics of Rett syndrome: when DNA methylation goes unrecognized. Nature Reviews Genetics, 2006, 7, 415-426.	7.7	266
17	Transcription Factor SOX3 Is Involved in X-Linked Mental Retardation with Growth Hormone Deficiency. American Journal of Human Genetics, 2002, 71, 1450-1455.	2.6	265
18	A new member of the IL-1 receptor family highly expressed in hippocampus and involved in X-linked mental retardation. Nature Genetics, 1999, 23, 25-31.	9.4	256

#	Article	IF	Citations
19	The wide spectrum of tubulinopathies: what are the key features for the diagnosis?. Brain, 2014, 137, 1676-1700.	3.7	252
20	ARX, a novel Prd-class-homeobox gene highly expressed in the telencephalon, is mutated in X-linked mental retardation. Human Molecular Genetics, 2002, 11, 981-991.	1.4	248
21	Immunolocalization and developmental expression of dystrophin related protein in skeletal muscle. Neuromuscular Disorders, 1991, 1, 185-194.	0.3	242
22	Key clinical features to identify girls with CDKL5 mutations. Brain, 2008, 131, 2647-2661.	3.7	242
23	Monogenic causes of X-linked mental retardation. Nature Reviews Genetics, 2001, 2, 669-680.	7.7	240
24	MECP2 is highly mutated in X-linked mental retardation. Human Molecular Genetics, 2001, 10, 941-946.	1.4	238
25	Mutations in the neuronal Î²-tubulin subunit TUBB3 result in malformation of cortical development and neuronal migration defects. Human Molecular Genetics, 2010, 19, 4462-4473.	1.4	231
26	Genetics and pathophysiology of mental retardation. European Journal of Human Genetics, 2006, 14, 701-713.	1.4	224
27	Mechanism of Microtubule Stabilization by Doublecortin. Molecular Cell, 2004, 14, 833-839.	4.5	220
28	Mutations in the Small GTPase Gene RAB39B Are Responsible for X-linked Mental Retardation Associated with Autism, Epilepsy, and Macrocephaly. American Journal of Human Genetics, 2010, 86, 185-195.	2.6	220
29	A new gene involved in X-linked mental retardation identified by analysis of an X;2 balanced translocation. Nature Genetics, 2000, 24, 167-170.	9.4	215
30	Large spectrum of lissencephaly and pachygyria phenotypes resulting from de novo missense mutations in tubulin alpha 1A (TUBA1A). Human Mutation, 2007, 28, 1055-1064.	1.1	213
31	Mutations in the connexin 32 gene in X-linked dominant Charcot- Marie - Tooth disease (CMTX1). Human Molecular Genetics, 1994, 3, 29-34.	1.4	191
32	Submicroscopic Duplications of the Hydroxysteroid Dehydrogenase HSD17B10 and the E3 Ubiquitin Ligase HUWE1 Are Associated with Mental Retardation. American Journal of Human Genetics, 2008, 82, 432-443.	2.6	187
33	Quantitative estimation of minor mRNAs by cDNA-polymerase chain reaction. Application to dystrophin mRNA in cultured myogenic and brain cells. FEBS Journal, 1990, 187, 691-698.	0.2	178
34	Effect of dystrophin gene deletions on mRNA levels and processing in Duchenne and Becker muscular dystrophies. Cell, 1990, 63, 1239-1248.	13.5	165
35	Doublecortin Functions at the Extremities of Growing Neuronal Processes. Cerebral Cortex, 2003, 13, 620-626.	1.6	163
36	Tubulin-related cortical dysgeneses: microtubule dysfunction underlying neuronal migration defects. Trends in Genetics, 2009, 25, 555-566.	2.9	162

#	Article	IF	Citations
37	Mutations in the Î²-Tubulin Gene TUBB5 Cause Microcephaly with Structural Brain Abnormalities. Cell Reports, 2012, 2, 1554-1562.	2.9	162
38	Dystrophin gene transcribed from different promoters in neuronal and glial cells. Nature, 1990, 344, 64-65.	13.7	159
39	Branching and nucleokinesis defects in migrating interneurons derived from doublecortin knockout mice. Human Molecular Genetics, 2006, 15, 1387-1400.	1.4	145
40	Clinical Heterogeneity of Duchenne Muscular Dystrophy (DMD): Definition of Sub-Phenotypes and Predictive Criteria by Long-Term Follow-Up. PLoS ONE, 2009, 4, e4347.	1.1	141
41	Mutations in the polyglutamine binding protein 1 gene cause X-linked mental retardation. Nature Genetics, 2003, 35, 313-315.	9.4	139
42	Human disorders of cortical development: from past to present. European Journal of Neuroscience, 2006, 23, 877-893.	1.2	138
43	Mutations in Eml1 lead to ectopic progenitors and neuronal heterotopia in mouse and human. Nature Neuroscience, 2014, 17, 923-933.	7.1	137
44	FACL4, encoding fatty acid-CoA ligase 4, is mutated in nonspecific X-linked mental retardation. Nature Genetics, 2002, 30, 436-440.	9.4	135
45	Structural variation in Xq28: MECP2 duplications in 1% of patients with unexplained XLMR and in 2% of male patients with severe encephalopathy. European Journal of Human Genetics, 2009, 17, 444-453.	1.4	130
46	Distinct roles of doublecortin modulating the microtubule cytoskeleton. EMBO Journal, 2006, 25, 4448-4457.	3.5	126
47	Loss of X-Linked Mental Retardation Gene Oligophrenin1 in Mice Impairs Spatial Memory and Leads to Ventricular Enlargement and Dendritic Spine Immaturity. Journal of Neuroscience, 2007, 27, 9439-9450.	1.7	125
48	GPR56-related bilateral frontoparietal polymicrogyria: further evidence for an overlap with the cobblestone complex. Brain, 2010, 133, 3194-3209.	3.7	125
49	Missense mutation in PAK3, R67C, causes X-linked nonspecific mental retardation. American Journal of Medical Genetics Part A, 2000, 93, 294-298.	2.4	122
50	Analysis of Dp71 contribution in the severity of mental retardation through comparison of Duchenne and Becker patients differing by mutation consequences on Dp71 expression. Human Molecular Genetics, 2009, 18, 3779-3794.	1.4	122
51	NovelJARID1C/SMCX mutations in patients with X-linked mental retardation. Human Mutation, 2006, 27, 389-389.	1.1	120
52	Cell-Autonomous Roles of ARX in Cell Proliferation and Neuronal Migration during Corticogenesis. Journal of Neuroscience, 2008, 28, 5794-5805.	1.7	118
53	Mutations in the FTSJ1 Gene Coding for a Novel S-Adenosylmethionineâ€“Binding Protein Cause Nonsyndromic X-Linked Mental Retardation. American Journal of Human Genetics, 2004, 75, 305-309.	2.6	117
54	A Novel Ribosomal S6-Kinase (RSK4; RPS6KA6) Is Commonly Deleted in Patients with Complex X-Linked Mental Retardation. Genomics, 1999, 62, 332-343.	1.3	113

#	Article	IF	Citations
55	Mutation of plasma membrane Ca ²⁺ ATPase isoform 3 in a family with X-linked congenital cerebellar ataxia impairs Ca ²⁺ homeostasis. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 14514-14519.	3.3	113
56	Parental origin of de novo MECP2 mutations in Rett syndrome. European Journal of Human Genetics, 2001, 9, 231-236.	1.4	109
57	Protein- and mRNA-based phenotype-genotype correlations in DMD/BMD with point mutations and molecular basis for BMD with nonsense and frameshift mutations in the DMD gene. Human Mutation, 2007, 28, 183-195.	1.1	107
58	A Postsynaptic Signaling Pathway that May Account for the Cognitive Defect Due to IL1RAPL1 Mutation. Current Biology, 2010, 20, 103-115.	1.8	106
59	Mutations in tubulin genes are frequent causes of various foetal malformations of cortical development including microlissencephaly. Acta Neuropathologica Communications, 2014, 2, 69.	2.4	106
60	Mutation frequencies of X-linked mental retardation genes in families from the EuroMRX consortium. Human Mutation, 2007, 28, 207-208.	1.1	103
61	The role of ARX in cortical development. European Journal of Neuroscience, 2006, 23, 869-876.	1.2	101
62	Mutations in the HECT domain of NEDD4L lead to AKTâ€“mTOR pathway deregulation and cause periventricular nodular heterotopia. Nature Genetics, 2016, 48, 1349-1358.	9.4	101
63	New insights into genotypeâ€“phenotype correlations for the doublecortin-related lissencephaly spectrum. Brain, 2013, 136, 223-244.	3.7	99
64	Seven novel Tay-Sachs mutations detected by chemical mismatch cleavage of PCR-amplified cDNA fragments. Genomics, 1991, 11, 124-134.	1.3	98
65	Monogenic X-linked mental retardation: Is it as frequent as currently estimated? The paradox of the ARX (Aristaless X) mutations. European Journal of Human Genetics, 2004, 12, 689-693.	1.4	98
66	Neuropathological phenotype of a distinct form of lissencephaly associated with mutations in TUBA1A. Brain, 2008, 131, 2304-2320.	3.7	98
67	IL1 receptor accessory protein like, a protein involved in X-linked mental retardation, interacts with Neuronal Calcium Sensor-1 and regulates exocytosis. Human Molecular Genetics, 2003, 12, 1415-1425.	1.4	96
68	The RhoGAP activity of OPHN1, a new F-actin-binding protein, is negatively controlled by its amino-terminal domain. Molecular and Cellular Neurosciences, 2003, 23, 574-586.	1.0	94
69	Role of Mental Retardation-Associated Dystrophin-Gene Product Dp71 in Excitatory Synapse Organization, Synaptic Plasticity and Behavioral Functions. PLoS ONE, 2009, 4, e6574.	1.1	94
70	In vitro follicular growth affects oocyte imprinting establishment in mice. European Journal of Human Genetics, 2003, 11, 493-496.	1.4	91
71	Neuroanatomical distribution of ARX in brain and its localisation in GABAergic neurons. Molecular Brain Research, 2004, 122, 35-46.	2.5	85
72	Mutations in the ZNF41 Gene Are Associated with Cognitive Deficits: Identification of a New Candidate for X-Linked Mental Retardation. American Journal of Human Genetics, 2003, 73, 1341-1354.	2.6	83

#	Article	IF	Citations
73	LIS1-Related Isolated Lissencephaly. Archives of Neurology, 2009, 66, 1007-15.	4.9	82
74	Illegitimate transcription: Its use in the study of inherited disease. Human Mutation, 1992, 1, 357-360.	1.1	80
75	Inhibition of RhoA pathway rescues the endocytosis defects in Oligophrenin1 mouse model of mental retardation. Human Molecular Genetics, 2009, 18, 2575-2583.	1.4	80
76	Next generation sequencing for molecular diagnosis of neuromuscular diseases. Acta Neuropathologica, 2012, 124, 273-283.	3.9	80
77	IL1-receptor accessory protein-like 1 (IL1RAPL1), a protein involved in cognitive functions, regulates N-type Ca2+-channel and neurite elongation. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 9063-9068.	3.3	78
78	ZNF674: A New KrÃ¼ppel-Associated Boxâ€“Containing Zinc-Finger Gene Involved in Nonsyndromic X-Linked Mental Retardation. American Journal of Human Genetics, 2006, 78, 265-278.	2.6	75
79	Revised spectrum of mutations in sarcoglycanopathies. European Journal of Human Genetics, 2008, 16, 793-803.	1.4	75
80	Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study. Lancet Neurology, The, 2015, 14, 1182-1195.	4.9	74
81	Revisiting the phenotype associated with FOXG1 mutations: two novel cases of congenital Rett variant. Neurogenetics, 2010, 11, 241-249.	0.7	72
82	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.	9.4	70
83	ZC4H2 Mutations Are Associated with Arthrogryposis Multiplex Congenita and Intellectual Disability through Impairment of Central and Peripheral Synaptic Plasticity. American Journal of Human Genetics, 2013, 92, 681-695.	2.6	68
84	Nonsyndromic X-linked mental retardation: where are the missing mutations?. Trends in Genetics, 2003, 19, 316-320.	2.9	65
85	Magnetic resonance imaging and histological studies of corpus callosal and hippocampal abnormalities linked todoublecortindeficiency. Journal of Comparative Neurology, 2007, 500, 239-254.	0.9	64
86	Disease-associated mutations in TUBA1A result in a spectrum of defects in the tubulin folding and heterodimer assembly pathway. Human Molecular Genetics, 2010, 19, 3599-3613.	1.4	63
87	Expanding the spectrum of TUBA1A-related cortical dysgenesis to Polymicrogyria. European Journal of Human Genetics, 2013, 21, 381-385.	1.4	63
88	Epilepsy in Dcx Knockout Mice Associated with Discrete Lamination Defects and Enhanced Excitability in the Hippocampus. PLoS ONE, 2008, 3, e2473.	1.1	63
89	Refining the phenotype associated with MEF2C point mutations. Neurogenetics, 2013, 14, 71-75.	0.7	60
90	Cloning the Wilson disease gene. Nature Genetics, 1993, 5, 317-318.	9.4	58

#	Article	IF	Citations
91	2.6 Mb YAC contig of the human X inactivation center region in Xq13: physical linkage of the RPS4X, PHKA1, XIST and DXS128E genes. Human Molecular Genetics, 1993, 2, 1105-1115.	1.4	57
92	Doublecortin Interacts with Î¼ Subunits of Clathrin Adaptor Complexes in the Developing Nervous System. Molecular and Cellular Neurosciences, 2001, 18, 307-319.	1.0	57
93	A circadian clock in hippocampus is regulated by interaction between oligophrenin-1 and Rev-erbÎ±. Nature Neuroscience, 2011, 14, 1293-1301.	7.1	57
94	In-frame deletion inMECP2 causes mild nonspecific mental retardation. American Journal of Medical Genetics Part A, 2002, 107, 81-83.	2.4	56
95	MCT8 mutation analysis and identification of the first female with Allanâ€“Herndonâ€“Dudley syndrome due to loss of MCT8 expression. European Journal of Human Genetics, 2008, 16, 1029-1037.	1.4	56
96	Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. Human Genetics, 2006, 118, 578-590.	1.8	55
97	Specific clinical and brain MRI features in mentally retarded patients with mutations in theOligophrenin-1 gene. American Journal of Medical Genetics Part A, 2004, 124A, 364-371.	2.4	52
98	Genetic and clinical specificity of 26 symptomatic carriers for dystrophinopathies at pediatric age. European Journal of Human Genetics, 2013, 21, 855-863.	1.4	52
99	Detection of exonic copy-number changes using a highly efficient oligonucleotide-based comparative genomic hybridization-array method. Human Mutation, 2008, 29, 1083-1090.	1.1	51
100	Xâ€linked mental retardation: focus on synaptic function and plasticity. Journal of Neurochemistry, 2009, 109, 1-14.	2.1	51
101	A shape analysis framework for neuromorphometry. Network: Computation in Neural Systems, 2002, 13, 283-310.	2.2	50
102	Rare De Novo Missense Variants in RNA Helicase DDX6 Cause Intellectual Disability and Dysmorphic Features and Lead to P-Body Defects and RNA Dysregulation. American Journal of Human Genetics, 2019, 105, 509-525.	2.6	50
103	Human lissencephaly with cerebellar hypoplasia due to mutations in TUBA1A: expansion of the foetal neuropathological phenotype. Acta Neuropathologica, 2010, 119, 779-789.	3.9	49
104	A novel mutation in the DLG3 gene encoding the synapse-associated protein 102 (SAP102) causes non-syndromic mental retardation. Neurogenetics, 2010, 11, 251-255.	0.7	49
105	A Pachygyria-causing Î±-Tubulin Mutation Results in Inefficient Cycling with CCT and a Deficient Interaction with TBCB. Molecular Biology of the Cell, 2008, 19, 1152-1161.	0.9	48
106	Mutation screening in 86 known X-linked mental retardation genes by droplet-based multiplex PCR and massive parallel sequencing. The HUGO Journal, 2009, 3, 41-49.	4.1	48
107	No mutations in the coding region of the Rett syndrome gene MECP2 in 59 autistic patients. European Journal of Human Genetics, 2001, 9, 556-558.	1.4	47
108	Mutation of the Diamond-Blackfan Anemia Gene Rps7 in Mouse Results in Morphological and Neuroanatomical Phenotypes. PLoS Genetics, 2013, 9, e1003094.	1.5	47

#	Article	IF	Citations
109	De novo mutations ofKIAA2022in females cause intellectual disability and intractable epilepsy. Journal of Medical Genetics, 2016, 53, 850-858.	1.5	47
110	Dystrophinopathy caused by mid-intronic substitutions activating cryptic exons in the DMD gene. Neuromuscular Disorders, 2004, 14, 10-18.	0.3	46
111	Loss of SLC38A5 and FTSJ1 at Xp11.23 in three brothers with non-syndromic mental retardation due to a microdeletion in an unstable genomic region. Human Genetics, 2007, 121, 539-547.	1.8	46
112	Beta tubulin isoforms are not interchangeable for rescuing impaired radial migration due to Tubb3 knockdown. Human Molecular Genetics, 2014, 23, 1516-1526.	1.4	46
113	Assessment of the structural and functional impact of in-frame mutations of the DMD gene, using the tools included in the eDystrophin online database. Orphanet Journal of Rare Diseases, 2012, 7, 45.	1.2	45
114	The Incidence of Rett Syndrome in France. Pediatric Neurology, 2006, 34, 372-375.	1.0	44
115	Doublecortin interacts with the ubiquitin protease DFFRX, which associates with microtubules in neuronal processes. Molecular and Cellular Neurosciences, 2005, 28, 153-164.	1.0	43
116	Novel mutation of IL1RAPL1 gene in a nonspecific Xâ€linked mental retardation (MRX) family. American Journal of Medical Genetics, Part A, 2008, 146A, 3167-3172.	0.7	43
117	Ade novoX;8 translocation creates aPTK2-THOC2gene fusion withTHOC2expression knockdown in a patient with psychomotor retardation and congenital cerebellar hypoplasia. Journal of Medical Genetics, 2013, 50, 543-551.	1.5	42
118	Novel mutations in NLGN3 causing autism spectrum disorder and cognitive impairment. Human Mutation, 2019, 40, 2021-2032.	1.1	42
119	A FOXG1 mutation in a boy with congenital variant of Rett syndrome. Neurogenetics, 2011, 12, 1-8.	0.7	40
120	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. PLoS Genetics, 2018, 14, e1007281.	1.5	40
121	Linkage of X-linked myopathy with excessive autophagy (XMEA) to Xq28. European Journal of Human Genetics, 2000, 8, 125-129.	1.4	39
122	DXS106 and DXS559 Flank the X-Linked Dystonia-Parkinsonism Syndrome Locus (DYT3). Genomics, 1994, 23, 114-117.	1.3	38
123	X-linked congenital ataxia: A clinical and genetic study. , 2000, 92, 53-56.		38
124	Myogenic Progenitor Cells Exhibit Type I Interferonâ€“Driven Proangiogenic Properties and Molecular Signature During Juvenile Dermatomyositis. Arthritis and Rheumatology, 2018, 70, 134-145.	2.9	38
125	A new model of experimental fibrosis in hindlimb skeletal muscle of adult mdx mouse mimicking muscular dystrophy. Muscle and Nerve, 2012, 45, 803-814.	1.0	37
126	Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117.	1.1	37

#	Article	IF	Citations
127	A novel splice mutation in PAK3 gene underlying mental retardation with neuropsychiatric features. European Journal of Human Genetics, 2008, 16, 1358-1363.	1.4	35
128	A distinctive gene expression fingerprint in mentally retarded male patients reflects disease-causing defects in the histone demethylase KDM5C. PathoGenetics, 2010, 3, 2.	5.7	35
129	Rapid reversal of impaired inhibitory and excitatory transmission but not spine dysgenesis in a mouse model of mental retardation. Journal of Physiology, 2012, 590, 763-776.	1.3	35
130	Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. Neuron, 2018, 100, 1354-1368.e5.	3.8	35
131	Characterization of a 1.0 Mb YAC contig spannning two chromosome breakpoints related to Menkes disease. Human Molecular Genetics, 1992, 1, 483-489.	1.4	34
132	CFTR illegitimate transcription in lymphoid cells: quantification and applications to the investigation of pathological transcripts. Human Genetics, 1992, 88, 508-512.	1.8	34
133	Gene for nonspecific X-linked mental retardation (MRX 47) is located in Xq22.3-q24. American Journal of Medical Genetics Part A, 1997, 72, 324-328.	2.4	34
134	Ciliogenesis and cell cycle alterations contribute to KIF2A-related malformations of cortical development. Human Molecular Genetics, 2018, 27, 224-238.	1.4	34
135	Increased diagnostic yield in complex dystonia through exome sequencing. Parkinsonism and Related Disorders, 2020, 74, 50-56.	1.1	34
136	A YAC contig in Xp21 containing the adrenal hypoplasia congenita and glycerol kinase deficiency genes. Human Molecular Genetics, 1992, 1, 579-585.	1.4	33
137	MECP2 gene mutations in non-syndromic X-linked mental retardation: Phenotype-genotype correlation. American Journal of Medical Genetics Part A, 2003, 123A, 129-139.	2.4	33
138	The location of DCX mutations predicts malformation severity in X-linked lissencephaly. Neurogenetics, 2008, 9, 277-285.	0.7	33
139	Combination of infantile spasms, non-epileptic seizures and complex movement disorder: A new case of ARX-related epilepsy. Epilepsy Research, 2008, 80, 224-228.	0.8	33
140	Mosaic parental germline mutations causing recurrent forms of malformations of cortical development. European Journal of Human Genetics, 2016, 24, 611-614.	1.4	33
141	RT-PCR and mRNA Quantitation. , 1994, , 97-109.		33
142	The first missense mutation causing Rett syndrome specifically affecting the MeCP2_e1 isoform. Neurogenetics, 2009, 10, 127-133.	0.7	32
143	IL1RAPL1 controls inhibitory networks during cerebellar development in mice. European Journal of Neuroscience, 2009, 30, 1476-1486.	1.2	32
144	Neuronal JNK pathway activation by IL-1 is mediated through IL1RAPL1, a protein required for development of cognitive functions. Communicative and Integrative Biology, 2010, 3, 245-247.	0.6	32

#	Article	IF	Citations
145	Fasudil treatment in adult reverses behavioural changes and brain ventricular enlargement in Oligophrenin-1 mouse model of intellectual disability. Human Molecular Genetics, 2016, 25, 2314-2323.	1.4	32
146	Calpain 10 and development of diabetes mellitus in cystic fibrosis. Journal of Cystic Fibrosis, 2006, 5, 47-51.	0.3	31
147	Somatic mosaicism for a CDKL5 mutation as an epileptic encephalopathy in males. American Journal of Medical Genetics, Part A, 2010, 152A, 2110-2111.	0.7	31
148	Astrocyte Transcriptome from the Mecp2308-Truncated Mouse Model of Rett Syndrome. NeuroMolecular Medicine, 2015, 17, 353-363.	1.8	31
149	Novel IL1RAPL1 mutations associated with intellectual disability impair synaptogenesis. Human Molecular Genetics, 2015, 24, 1106-1118.	1.4	31
150	Homozygous truncating mutation of the KBP gene, encoding a KIF1B-binding protein, in a familial case of fetal polymicrogyria. Neurogenetics, 2013, 14, 215-224.	0.7	30
151	A 3-base pair deletion, c.9711_9713del, in DMD results in intellectual disability without muscular dystrophy. European Journal of Human Genetics, 2014, 22, 480-485.	1.4	30
152	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	2.6	30
153	Mapping of the X-Breakpoint Involved in a Balanced X;12 Translocation in a Female with Mild Mental Retardation. European Journal of Human Genetics, 1997, 5, 105-109.	1.4	30
154	TheARX mutations: A frequent cause of X-linked mental retardation. American Journal of Medical Genetics, Part A, 2006, 140A, 727-732.	0.7	29
155	Use of SNP array analysis to identify a novel TRIM32 mutation in limb-girdle muscular dystrophy type 2H. Neuromuscular Disorders, 2009, 19, 255-260.	0.3	29
156	Target-Specific Vulnerability of Excitatory Synapses Leads to Deficits in Associative Memory in a Model of Intellectual Disorder. Journal of Neuroscience, 2013, 33, 13805-13819.	1.7	29
157	Expanding the clinical phenotype of patients with a ZDHHC9 mutation. American Journal of Medical Genetics, Part A, 2014, 164, 789-795.	0.7	29
158	Loss of parental-specific methylation at theIGF2locus in human hepatocellular carcinoma. Journal of Pathology, 2003, 201, 473-479.	2.1	28
159	Lack of the presynaptic RhoGAP protein oligophrenin1 leads to cognitive disabilities through dysregulation of the cAMP/PKA signalling pathway. Philosophical Transactions of the Royal Society B: Biological Sciences, 2014, 369, 20130160.	1.8	28
160	Dp412e: a novel human embryonic dystrophin isoform induced by BMP4 in early differentiated cells. Skeletal Muscle, 2015, 5, 40.	1.9	28
161	WDR81 mutations cause extreme microcephaly and impair mitotic progression in human fibroblasts and Drosophila neural stem cells. Brain, 2017, 140, 2597-2609.	3.7	28
162	Analysis of Mnk, the Murine Homologue of the Locus for Menkes Disease, in Normal and Mottled (Mo) Mice. Genomics, 1994, 22, 27-35.	1.3	27

#	Article	IF	Citations
163	An isoform of the severe encephalopathy-related CDKL5 gene, including a novel exon with extremely high sequence conservation, is specifically expressed in brain. Journal of Human Genetics, 2011, 56, 52-57.	1.1	27
164	Altered microtubule dynamics in Mecp2â€deficient astrocytes. Journal of Neuroscience Research, 2012, 90, 990-998.	1.3	27
165	When a mid-intronic variation of DMD gene creates an ESE site. Neuromuscular Disorders, 2014, 24, 1111-1117.	0.3	27
166	From Fragile X Mental Retardation Protein to Rac1 GTPase. Neuron, 2003, 38, 843-845.	3.8	26
167	Netrin G1 Mutations Are an Uncommon Cause of Atypical Rett Syndrome With or Without Epilepsy. Pediatric Neurology, 2007, 37, 270-274.	1.0	26
168	Rare ACTG1 variants in fetal microlissencephaly. European Journal of Medical Genetics, 2015, 58, 416-418.	0.7	26
169	Mutation screening of ASMT, the last enzyme of the melatonin pathway, in a large sample of patients with Intellectual Disability. BMC Medical Genetics, 2011, 12, 17.	2.1	25
170	MRX review. American Journal of Medical Genetics Part A, 2000, 94, 364-366.	2.4	23
171	Mutation screening of brain-expressed X-chromosomal miRNA genes in 464 patients with nonsyndromic X-linked mental retardation. European Journal of Human Genetics, 2007, 15, 375-378.	1.4	23
172	ARX polyalanine expansions are highly implicated in familial cases of mental retardation with infantile epilepsy and/or hand dystonia. American Journal of Medical Genetics, Part A, 2011, 155, 98-105.	0.7	23
173	MeCP2 deficiency is associated with impaired microtubule stability. FEBS Letters, 2013, 587, 245-253.	1.3	23
174	De novo TUBB2B mutation causes fetal akinesia deformation sequence with microlissencephaly: An unusual presentation of tubulinopathy. European Journal of Medical Genetics, 2016, 59, 249-256.	0.7	23
175	Synaptic Maturation at Cortical Projections to the Lateral Amygdala in a Mouse Model of Rett Syndrome. PLoS ONE, 2010, 5, e11399.	1.1	23
176	Molecular cytogenetic analysis of a duplication Xp in a female with an abnormal phenotype and random X inactivation. Clinical Genetics, 2001, 58, 116-122.	1.0	22
177	Inv(X)(p21.1;q22.1) in a man with mental retardation, short stature, general muscle wasting, and facial dysmorphism: Clinical study and mutation analysis of theNXF5 Gene. American Journal of Medical Genetics Part A, 2003, 119A, 367-374.	2.4	21
178	Hybridisation-based resequencing of 17 X-linked intellectual disability genes in 135 patients reveals novel mutations in ATRX, SLC6A8 and PQBP1. European Journal of Human Genetics, 2011, 19, 717-720.	1.4	21
179	Detection of TRIM32 deletions in LGMD patients analyzed by a combined strategy of CGH array and massively parallel sequencing. European Journal of Human Genetics, 2015, 23, 929-934.	1.4	21
180	Liver insulin-like growth factor 2 methylation in hepatitis C virus cirrhosis and further occurrence of hepatocellular carcinoma. World Journal of Gastroenterology, 2008, 14, 5419.	1.4	21

#	Article	IF	Citations
181	Fine mapping of the human SCIDX1 locus at Xq12â€“13.1. Human Molecular Genetics, 1993, 2, 651-654.	1.4	20
182	Custom oligonucleotide array-based CGH: a reliable diagnostic tool for detection of exonic copy-number changes in multiple targeted genes. European Journal of Human Genetics, 2013, 21, 977-987.	1.4	20
183	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. Human Molecular Genetics, 2018, 27, 589-600.	1.4	20
184	Illegitimate (or ectopic) transcription proceeds through the usual promoters. Biochemical and Biophysical Research Communications, 1991, 178, 553-557.	1.0	19
185	X-linked mental retardation: a comprehensive molecular screen of 47 candidate genes from a 7.4 Mb interval in Xp11. European Journal of Human Genetics, 2007, 15, 68-75.	1.4	19
186	Alternative Transcripts of <i>Dclk</i>1 and <i>Dclk</i>2 and Their Expression in <i>Doublecortin</i> Knockout Mice. Developmental Neuroscience, 2008, 30, 171-186.	1.0	19
187	Progressive muscular dystrophies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1343-1366.	1.0	19
188	Conditional switching of KIF2A mutation provides new insights into cortical malformation pathogeny. Human Molecular Genetics, 2020, 29, 766-784.	1.4	19
189	Construction of a YAC contig spanning the Xq13.3 subband. Genomics, 1995, 26, 115-122.	1.3	18
190	Epileptic encephalopathy in a girl with an interstitial deletion of Xp22 comprising promoter and exon 1 of the CDKL5 gene. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 202-207.	1.1	18
191	Mutations of TSEN and CASK genes are prevalent in pontocerebellar hypoplasias type 2 and 4. Brain, 2012, 135, e199-e199.	3.7	18
192	Spectrum of MECP2 Mutations in Rett Syndrome. Genetic Testing and Molecular Biomarkers, 2002, 6, 1-6.	1.7	17
193	A missense mutation within the fork-head domain of the forkhead box G1 Gene (FOXG1) affects its nuclear localization. Human Mutation, 2011, 32, E2026-E2035.	1.1	17
194	Mosaic DCX deletion causes subcortical band heterotopia in males. Neurogenetics, 2012, 13, 367-373.	0.7	17
195	Insulin-like growth factor 2 gene methylation in peripheral blood mononuclear cells of patients with hepatitis C related cirrhosis or hepatocellular carcinoma. Clinics and Research in Hepatology and Gastroenterology, 2012, 36, 345-351.	0.7	17
196	TUBG1 missense variants underlying cortical malformations disrupt neuronal locomotion and microtubule dynamics but not neurogenesis. Nature Communications, 2019, 10, 2129.	5.8	17
197	X-linked nonspecific mental retardation (MRX16) mapping to distal Xq28: Linkage study and neuropsychological data in a large family. American Journal of Medical Genetics Part A, 1999, 83, 411-418.	2.4	16
198	X-linked nonspecific mental retardation (MRX) linkage studies in 25 unrelated families: The European XLMR consortium. , 1999, 85, 263-265.		16

#	Article	IF	Citations
199	A novel frame shift mutation in the PQBP1 gene identified in a Tunisian family with X-linked mental retardation. European Journal of Medical Genetics, 2011, 54, 241-246.	0.7	16
200	First case report of Cohen syndrome in the Tunisian population caused by VPS13B mutations. BMC Medical Genetics, 2017, 18, 134.	2.1	16
201	A new mouse model of ARX dup24 recapitulates the patientsâ€™ behavioral and fine motor alterations. Human Molecular Genetics, 2018, 27, 2138-2153.	1.4	16
202	Neuropathological Hallmarks of Brain Malformations in Extreme Phenotypes Related to DYNC1H1 Mutations. Journal of Neuropathology and Experimental Neurology, 2017, 76, 195-205.	0.9	15
203	Mutation in the SLC9A6 gene is not a frequent cause of sporadic Angelman-like syndrome. European Journal of Human Genetics, 2009, 17, 1378-1380.	1.4	14
204	A null allele frequent in non-Jewish Tay-Sachs patients. Human Genetics, 1993, 90, 614-20.	1.8	13
205	7 Menkes and Wilson Diseases. Advances in Genetics, 1995, 33, 233-253.	0.8	13
206	Characterization of a Highly Complex Region in Xq13 and Mapping of Three Isodicentric Breakpoints Associated with Preleukemia. Genomics, 2000, 64, 221-229.	1.3	13
207	TM4SF2 gene involvement reconsidered in an XLMR family after neuropsychological assessment. American Journal of Medical Genetics Part A, 2002, 112, 400-404.	2.4	13
208	Isolation of DNTR polymorphisms from yeast artificial chromosomes encompassing X chromosomal loci PGK1 and DXS56. Human Genetics, 1992, 90, 270-4.	1.8	12
209	Determination of the gene structure of human oligophrenin-1 and identification of three novel polymorphisms by screening of DNA from 164 patients with non-specific X-linked mental retardation. Annales De GÃ©nÃ©tique, 2000, 43, 5-9.	0.4	12
210	Xâ€linked congenital ataxia: A new locus maps to Xq25â€q27.1. American Journal of Medical Genetics, Part A, 2008, 146A, 593-600.	0.7	12
211	The c.429_452 duplication of the ARX gene: a unique developmental-model of limb kinetic apraxia. Orphanet Journal of Rare Diseases, 2014, 9, 25.	1.2	12
212	Analysis of Alternative Splicing Patterns in the Cystic Fibrosis Transmembrane Conductance Regulator Gene Using mRNA Derived from Lymphoblastoid Cells of Cystic Fibrosis Patients. European Journal of Human Genetics, 1996, 4, 127-134.	1.4	12
213	Mutations in exon 1 of MECP2B are not a common cause of X-linked mental retardation in males. European Journal of Human Genetics, 2005, 13, 523-524.	1.4	11
214	Inherited microdeletion in Xp21.3â€“22.1 involved in nonâ€specific mental retardation. Clinical Genetics, 1998, 53, 136-141.	1.0	11
215	Cell cloning-based transcriptome analysis in cyclin-dependent kinase-like 5 mutation patients with severe epileptic encephalopathy. Journal of Molecular Medicine, 2011, 89, 193-202.	1.7	11
216	Non Random Distribution of DMD Deletion Breakpoints and Implication of Double Strand Breaks Repair and Replication Error Repair Mechanisms. Journal of Neuromuscular Diseases, 2016, 3, 227-245.	1.1	11

#	Article	IF	Citations
217	A Novel Analog Reasoning Paradigm: New Insights in Intellectually Disabled Patients. PLoS ONE, 2016, 11, e0149717.	1.1	10
218	Impairment of cerebello-thalamo-frontal pathway in Rab-GDI mutated patients with pure mental deficiency. European Journal of Medical Genetics, 2009, 52, 6-13.	0.7	9
219	A step toward essential tremor gene discovery: identification of extreme phenotype and screening of HTRA2 and ANO3. BMC Neurology, 2016, 16, 238.	0.8	9
220	Protein Kinase A Deregulation in the Medial Prefrontal Cortex Impairs Working Memory in Murine Oligophrenin-1 Deficiency. Journal of Neuroscience, 2017, 37, 11114-11126.	1.7	9
221	Dinucleotide repeat polymorphisms from DXS106 and DXS227 YACs using a two stage approach. Human Molecular Genetics, 1993, 2, 607-608.	1.4	8
222	Assignment of the gene (EPLG2) encoding a high-affinity binding protein for the receptor tyrosine kinase Elk to a 200-kilobasepair region in human chromosome Xq12. Genomics, 1995, 25, 334-335.	1.3	8
223	Screening of 20 patients with X-linked mental retardation using chromosome X-specific array-MAPH. European Journal of Medical Genetics, 2007, 50, 399-410.	0.7	8
224	A New Chromosome X Exon-Specific Microarray Platform for Screening of Patients with X-Linked Disorders. Journal of Molecular Diagnostics, 2009, 11, 562-568.	1.2	8
225	Focal polymicrogyria are associated with submicroscopic chromosomal rearrangements detected by CGH microarray analysis. European Journal of Medical Genetics, 2012, 55, 527-530.	0.7	8
226	Refined 2.7 centimorgan locus in Xp21.3-22.1 for a nonspecific X-linked mental retardation gene (MRX54). , 1999, 85, 276-282.		7
227	X-linked mental retardation, short stature, microcephaly and hypogonadism maps to Xp22.1-p21.3 in a Belgian family. European Journal of Medical Genetics, 2005, 48, 145-152.	0.7	7
228	Deep intronic variation in splicing regulatory element of the ERCC8 gene associated with severe but long-term survival Cockayne syndrome. European Journal of Human Genetics, 2018, 26, 527-536.	1.4	7
229	Cosegregation of T108A Elk-1 with mental retardation. American Journal of Medical Genetics Part A, 2000, 95, 404-405.	2.4	5
230	Sequence variation in ultraconserved and highly conserved elements does not cause X-linked mental retardation. American Journal of Medical Genetics, Part A, 2007, 143A, 888-890.	0.7	5
231	Recurrent deletion of ZNF630 at Xp11.23 is not associated with mental retardation. American Journal of Medical Genetics, Part A, 2010, 152A, 638-645.	0.7	5
232	Mutations in the ERCC2 (XPD) gene associated with severe fetal ichthyosis and dysmorphic features. Prenatal Diagnosis, 2016, 36, 1276-1279.	1.1	5
233	RT-PCR and Gene Expression. , 1994, , 277-294.		5
234	Striking conservation of the brain-specific region of the dystrophin gene. Mammalian Genome, 1993, 4, 393-396.	1.0	4

#	Article	IF	Citations
235	CDKL5 and ARX mutations are not responsible for early onset severe myoclonic epilepsy in infancy. Epilepsy Research, 2009, 87, 25-30.	0.8	4
236	Novel role of the synaptic scaffold protein Dlgap4 in ventricular surface integrity and neuronal migration during cortical development. Nature Communications, 2022, 13, 2746.	5.8	4
237	A gene for nonsyndromic X-linked mental retardation (MRX77) maps to Xq12-Xq21.33. American Journal of Medical Genetics Part A, 2003, 122A, 46-50.	2.4	3
238	Five novel frameshift mutations in exon 3 and 4 of the MECP2 gene identified in Rett patients: Consequences for the molecular diagnosis strategy. Human Mutation, 2001, 18, 251-252.	1.1	2
239	Exclusion of the coding sequence of the doublecortin gene as a susceptibility locus in autistic disorder. American Journal of Medical Genetics Part A, 2002, 108, 164-167.	2.4	2
240	Nebulin seen in DMD males including one patient with a large DNA deletion encompassing the DMD gene. Human Genetics, 1988, 78, 285-285.	1.8	1
241	Construction of a highly annotated cosmid contig spanning 550Kb within the X-linked nonspecific mental retardation candidate region at Xp21.3-22.1. , 1999, 85, 252-254.		1
242	Tubulin-Related Malformations of Cortical Development. , 2012, , 315-341.		1
243	Genetic Causes of Intellectual Disability: TheÂGenes Controlling Cortical Development. , 2016, , 43-64.		0
244	Dystrophin Gene Expression in Neuronal Tissues. Methods in Neurosciences, 1992, , 47-63.	0.5	0
245	Genetics of X-Linked Mental Retardation. , 0, , 263-288.		0