Jozef Geczy

List of Publications by Year
in descending order

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334 papers	24,700 citations	4641 85 h-index	11030 137 g-index
359 all docs	359 docs citations	359 times ranked	25039 citing authors

#	Article	IF	Citations
1	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	9.4	583
2	Duplication of the MECP2 Region Is a Frequent Cause of Severe Mental Retardation and Progressive Neurological Symptoms in Males. American Journal of Human Genetics, 2005, 77, 442-453.	2.6	550
3	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	9.4	539
4	A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. Nature Genetics, 2009, 41, 535-543.	9.4	528
5	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	9.4	443
6	Mutations of CDKL5 Cause a Severe Neurodevelopmental Disorder with Infantile Spasms and Mental Retardation. American Journal of Human Genetics, 2004, 75, 1079-1093.	2.6	414
7	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. Nature Genetics, 2008, 40, 776-781.	9.4	397
8	Mutations in the human ortholog of Aristaless cause X-linked mental retardation and epilepsy. Nature Genetics, 2002, 30, 441-445.	9.4	396
9	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
10	Identification of the gene FMR2, associated with FRAXE mental retardation. Nature Genetics, 1996, 13, 105-108.	9.4	303
11	A novel X-linked trichothiodystrophy associated with a nonsense mutation in RNF113A. Journal of Medical Genetics, 2015, 52, 269-274.	1.5	302
12	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. Nature Genetics, 2013, 45, 546-551.	9.4	301
13	Cerebral palsy: causes, pathways, and the role of genetic variants. American Journal of Obstetrics and Gynecology, 2015, 213, 779-788.	0.7	290
14	PHF6 mutations in T-cell acute lymphoblastic leukemia. Nature Genetics, 2010, 42, 338-342.	9.4	282
15	Identification of a MicroRNA that Activates Gene Expression by Repressing Nonsense-Mediated RNA Decay. Molecular Cell, 2011, 42, 500-510.	4.5	267
16	Disruption of the Serine/Threonine Kinase 9 Gene Causes Severe X-Linked Infantile Spasms and Mental Retardation. American Journal of Human Genetics, 2003, 72, 1401-1411.	2.6	265
17	Mutations in the X-Linked Cyclin-Dependent Kinaseâ€“Like 5 (CDKL5/STK9) Gene Are Associated with Severe Neurodevelopmental Retardation. American Journal of Human Genetics, 2004, 75, 1149-1154.	2.6	264
18	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

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19	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. American Journal of Human Genetics, 2016, 99, 287-298.	2.6	247
20	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. Nature, 2018, 562, 268-271.	13.7	246
21	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. Molecular Psychiatry, 2016, 21, 133-148.	4.1	243
22	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. Genome Biology, 2016, 17, 243.	3.8	241
23	PRRT2 Mutations Cause Benign Familial Infantile Epilepsy and Infantile Convulsions with Choreoathetosis Syndrome. American Journal of Human Genetics, 2012, 90, 152-160.	2.6	234
24	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	2.6	230
25	Mutations in UPF3B, a member of the nonsense-mediated mRNA decay complex, cause syndromic and nonsyndromic mental retardation. Nature Genetics, 2007, 39, 1127-1133.	9.4	228
26	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
27	SLC9A6 Mutations Cause X-Linked Mental Retardation, Microcephaly, Epilepsy, and Ataxia, a Phenotype Mimicking Angelman Syndrome. American Journal of Human Genetics, 2008, 82, 1003-1010.	2.6	209
28	COMMD1 is linked to the WASH complex and regulates endosomal trafficking of the copper transporter ATP7A. Molecular Biology of the Cell, 2015, 26, 91-103.	0.9	200
29	Mutations in CUL4B, Which Encodes a Ubiquitin E3 Ligase Subunit, Cause an X-linked Mental Retardation Syndrome Associated with Aggressive Outbursts, Seizures, Relative Macrocephaly, Central Obesity, Hypogonadism, Pes Cavus, and Tremor. American Journal of Human Genetics, 2007, 80, 345-352.	2.6	197
30	Mutations in mammalian target of rapamycin regulator DEPDC5 cause focal epilepsy with brain malformations. Annals of Neurology, 2014, 75, 782-787.	2.8	193
31	Mutations in PHF6 are associated with BÃ¶rjesonâ€“ForssmanÂâ€“Lehmann syndrome. Nature Genetics, 2002, 32, 661-665.	9.4	192
32	The genetic landscape of intellectual disability arising from chromosome X. Trends in Genetics, 2009, 25, 308-316.	2.9	190
33	Identification of the gene (SEDL) causing X-linked spondyloepiphyseal dysplasia tarda. Nature Genetics, 1999, 22, 400-404.	9.4	188
34	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
35	Disruption at the PTCHD1 Locus on Xp22.11 in Autism Spectrum Disorder and Intellectual Disability. Science Translational Medicine, 2010, 2, 49ra68.	5.8	178
36	Whole-exome sequencing points to considerable genetic heterogeneity of cerebral palsy. Molecular Psychiatry, 2015, 20, 176-182.	4.1	178

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37	Epilepsy and mental retardation limited to females: an under-recognized disorder. Brain, 2008, 131, 918-927.	3.7	172
38	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	2.6	171
39	Infantile spasms, dystonia, and other X-linked phenotypes caused by mutations in Aristaless related homeobox gene, ARX. Brain and Development, 2002, 24, 266-268.	0.6	170
40	A Mutation in the Golgi Qb-SNARE Gene GOSR2 Causes Progressive Myoclonus Epilepsy with Early Ataxia. American Journal of Human Genetics, 2011, 88, 657-663.	2.6	166
41	Rett syndrome: clinical review and genetic update. Journal of Medical Genetics, 2005, 42, 1-7.	1.5	161
42	Targeted Nextâ€Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. Human Mutation, 2015, 36, 1197-1204.	1.1	161
43	Early onset seizures and Rett-like features associated with mutations in CDKL5. European Journal of Human Genetics, 2005, 13, 1113-1120.	1.4	160
44	Mutations in the DLG3 Gene Cause Nonsyndromic X-Linked Mental Retardation. American Journal of Human Genetics, 2004, 75, 318-324.	2.6	157
45	XLMR genes: update 2007. European Journal of Human Genetics, 2008, 16, 422-434.	1.4	155
46	The original Lujan syndrome family has a novel missense mutation (p.N1007S) in the MED12 gene. Journal of Medical Genetics, 2007, 44, 472-477.	1.5	153
47	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	7.1	152
48	ARX spectrum disorders: making inroads into the molecular pathology. Human Mutation, 2010, 31, 889-900.	1.1	151
49	Mutations in ZDHHC9, Which Encodes a Palmitoyltransferase of NRAS and HRAS, Cause X-Linked Mental Retardation Associated with a Marfanoid Habitus. American Journal of Human Genetics, 2007, 80, 982-987.	2.6	150
50	La FAM fatale: USP9X in development and disease. Cellular and Molecular Life Sciences, 2015, 72, 2075-2089.	2.4	145
51	A missense mutation in RPS6KA3 (RSK2) responsible for non-specific mental retardation. Nature Genetics, 1999, 22, 13-14.	9.4	142
52	Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. Molecular Psychiatry, 2016, 21, 126-132.	4.1	142
53	Mutations in the polyglutamine binding protein 1 gene cause X-linked mental retardation. Nature Genetics, 2003, 35, 313-315.	9.4	139
54	Mutations in the guanine nucleotide exchange factor gene IQSEC2 cause nonsyndromic intellectual disability. Nature Genetics, 2010, 42, 486-488.	9.4	134

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55	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	2.6	132
56	Oligosaccharyltransferase-Subunit Mutations in Nonsyndromic Mental Retardation. American Journal of Human Genetics, 2008, 82, 1150-1157.	2.6	130
57	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
58	Nonsense-mediated mRNA decay: Inter-individual variability and human disease. Neuroscience and Biobehavioral Reviews, 2014, 46, 175-186.	2.9	130
59	Functional characterization of GATA3 mutations causing the hypoparathyroidism-deafness-renal (HDR) dysplasia syndrome: insight into mechanisms of DNA binding by the GATA3 transcription factor. Human Molecular Genetics, 2006, 16, 265-275.	1.4	129
60	â€˜North Seaâ€™ progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. Brain, 2013, 136, 1146-1154.	3.7	129
61	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	2.6	125
62	The genetic basis of cerebral palsy. Developmental Medicine and Child Neurology, 2017, 59, 462-469.	1.1	125
63	ARX: a gene for all seasons. Current Opinion in Genetics and Development, 2006, 16, 308-316.	1.5	123
64	Defects in tRNA Anticodon Loop 2â€²-O-Methylation Are Implicated in Nonsyndromic X-Linked Intellectual Disability due to Mutations inFTSJ1. Human Mutation, 2015, 36, 1176-1187.	1.1	122
65	Contribution of copy number variants involving nonsense-mediated mRNA decay pathway genes to neuro-developmental disorders. Human Molecular Genetics, 2013, 22, 1816-1825.	1.4	120
66	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
67	Mutations in USP9X Are Associated with X-Linked Intellectual Disability and Disrupt Neuronal Cell Migration and Growth. American Journal of Human Genetics, 2014, 94, 470-478.	2.6	117
68	Mutations of the UPF3B gene, which encodes a protein widely expressed in neurons, are associated with nonspecific mental retardation with or without autism. Molecular Psychiatry, 2010, 15, 767-776.	4.1	113
69	Biallelic SUN5 Mutations Cause Autosomal-Recessive Acephalic Spermatozoa Syndrome. American Journal of Human Genetics, 2016, 99, 942-949.	2.6	113
70	Variable expression of mental retardation, autism, seizures, and dystonic hand movements in two families with an identicalARX gene mutation. American Journal of Medical Genetics Part A, 2002, 112, 405-411.	2.4	111
71	A Focal Epilepsy and Intellectual Disability Syndrome Is Due to a Mutation in TBC1D24. American Journal of Human Genetics, 2010, 87, 371-375.	2.6	111
72	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. Nature Communications, 2019, 10, 4919.	5.8	111

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73	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659.	1.4	108
74	Mutations in a Novel Gene, NHS, Cause the Pleiotropic Effects of Nance-Horan Syndrome, Including Severe Congenital Cataract, Dental Anomalies, and Mental Retardation. American Journal of Human Genetics, 2003, 73, 1120-1130.	2.6	107
75	Fine-Scale Survey of X Chromosome Copy Number Variants and Indels Underlying Intellectual Disability. American Journal of Human Genetics, 2010, 87, 173-188.	2.6	107
76	A UPF3-mediated regulatory switch that maintains RNA surveillance. Nature Structural and Molecular Biology, 2009, 16, 747-753.	3.6	106
77	CASK mutations are frequent in males and cause X-linked nystagmus and variable XLMR phenotypes. European Journal of Human Genetics, 2010, 18, 544-552.	1.4	105
78	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
79	Mutation frequencies of X-linked mental retardation genes in families from the EuroMRX consortium. Human Mutation, 2007, 28, 207-208.	1.1	103
80	Mutations in the Gene Encoding the Sigma 2 Subunit of the Adaptor Protein 1 Complex, AP1S2, Cause X-Linked Mental Retardation. American Journal of Human Genetics, 2006, 79, 1119-1124.	2.6	102
81	XNP mutation in a large family with Juberg-Marsidi syndrome. Nature Genetics, 1996, 12, 359-360.	9.4	101
82	The UPF3B gene, implicated in intellectual disability, autism, ADHD and childhood onset schizophrenia regulates neural progenitor cell behaviour and neuronal outgrowth. Human Molecular Genetics, 2013, 22, 4673-4687.	1.4	101
83	CCDC22 deficiency in humans blunts activation of proinflammatory NF-ÎºB signaling. Journal of Clinical Investigation, 2013, 123, 2244-2256.	3.9	101
84	Mutations in ionotropic AMPA receptor 3 alter channel properties and are associated with moderate cognitive impairment in humans. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 18163-18168.	3.3	100
85	Abnormal Cell Sorting Underlies the Unique X-Linked Inheritance of PCDH19 Epilepsy. Neuron, 2018, 97, 59-66.e5.	3.8	100
86	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	5.8	99
87	Transcriptome profiling of UPF3B/NMD-deficient lymphoblastoid cells from patients with various forms of intellectual disability. Molecular Psychiatry, 2012, 17, 1103-1115.	4.1	97
88	TBC1D24 genotypeâ€“phenotype correlation. Neurology, 2016, 87, 77-85.	1.5	97
89	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	9.4	96
90	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. American Journal of Human Genetics, 2016, 98, 373-381.	2.6	95

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91	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	1.4	93
92	The Molecular Basis of X-Linked Spondyloepiphyseal Dysplasia Tarda. American Journal of Human Genetics, 2001, 68, 1386-1397.	2.6	90
93	A Noncoding, Regulatory Mutation Implicates HCFC1 in Nonsyndromic Intellectual Disability. American Journal of Human Genetics, 2012, 91, 694-702.	2.6	89
94	X-linked myoclonic epilepsy with spasticity and intellectual disability. Neurology, 2002, 59, 348-356.	1.5	88
95	Identification and characterization of a missense mutation in the O-linked Î²-N-acetylglucosamine (O-GlcNAc) transferase gene that segregates with X-linked intellectual disability. Journal of Biological Chemistry, 2017, 292, 8948-8963.	1.6	87
96	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	2.6	87
97	A systematic review and meta-analysis of 271 PCDH19-variant individuals identifies psychiatric comorbidities, and association of seizure onset and disease severity. Molecular Psychiatry, 2019, 24, 241-251.	4.1	86
98	Mutations in the BRWD3 Gene Cause X-Linked Mental Retardation Associated with Macrocephaly. American Journal of Human Genetics, 2007, 81, 367-374.	2.6	85
99	Characterization of the Human Glutamate Receptor Subunit 3 Gene (GRIA3), a Candidate for Bipolar Disorder and Nonspecific X-Linked Mental Retardation. Genomics, 1999, 62, 356-368.	1.3	84
100	A ubiquitin-dependent signalling axis specific for ALKBH-mediated DNA dealkylation repair. Nature, 2017, 551, 389-393.	13.7	83
101	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. American Journal of Human Genetics, 2015, 97, 302-310.	2.6	82
102	Genetic or Other Causation Should Not Change the Clinical Diagnosis of Cerebral Palsy. Journal of Child Neurology, 2019, 34, 472-476.	0.7	82
103	FOXP1 mutations cause intellectual disability and a recognizable phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 3166-3175.	0.7	79
104	Loss of Usp9x Disrupts Cortical Architecture, Hippocampal Development and TGFÎ²-Mediated Axonogenesis. PLoS ONE, 2013, 8, e68287.	1.1	77
105	Mouse orthologue of ARX, a gene mutated in several X-linked forms of mental retardation and epilepsy, is a marker of adult neural stem cells and forebrain GABAergic neurons. Developmental Dynamics, 2004, 231, 631-639.	0.8	76
106	Two novel JAK2 exon 12 mutations in JAK2V617F-negative polycythaemia vera patients. Leukemia, 2008, 22, 870-873.	3.3	76
107	Fibroblast growth factor homologous factor 2 (FHF2): gene structure, expression and mapping to the BÃ¶rjeson-Forssman-Lehmann syndrome region in Xq26 delineated by a duplication breakpoint in a BFLS-like patient. Human Genetics, 1999, 104, 56-63.	1.8	75
108	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

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109	Epilepsy and mental retardation limited to females with PCDH19 mutations can present de novo or in single generation families. Journal of Medical Genetics, 2010, 47, 211-216.	1.5	74
110	Evolution of the human X â€“ a smart and sexy chromosome that controls speciation and development. Cytogenetic and Genome Research, 2002, 99, 141-145.	0.6	72
111	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. Neurology, 2016, 87, 1975-1984.	1.5	71
112	Mutations in the intellectual disability gene KDM5C reduce protein stability and demethylase activity. Human Molecular Genetics, 2015, 24, 2861-2872.	1.4	69
113	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
114	Severe childhood speech disorder. Neurology, 2020, 94, e2148-e2167.	1.5	68
115	Genes for Cognitive Function: Developments on the X. Genome Research, 2000, 10, 157-163.	2.4	67
116	FRAXE-associated mental retardation protein (FMR2) is an RNA-binding protein with high affinity for G-quartet RNA forming structure. Nucleic Acids Research, 2009, 37, 1269-1279.	6.5	67
117	Identification and characterization of two novel JARID1C mutations: suggestion of an emerging genotypeâ€“phenotype correlation. European Journal of Human Genetics, 2010, 18, 330-335.	1.4	66
118	Seizures Are Regulated by Ubiquitin-specific Peptidase 9 X-linked (USP9X), a De-Ubiquitinase. PLoS Genetics, 2015, 11, e1005022.	1.5	66
119	Nonsyndromic X-linked mental retardation: where are the missing mutations?. Trends in Genetics, 2003, 19, 316-320.	2.9	65
120	Rare copy number variation in cerebral palsy. European Journal of Human Genetics, 2014, 22, 40-45.	1.4	65
121	Gene Structure and Subcellular Localization of FMR2, a Member of a New Family of Putative Transcription Activators. Genomics, 1997, 44, 201-213.	1.3	64
122	Hypomorphic Temperature-Sensitive Alleles of NSDHL Cause CK Syndrome. American Journal of Human Genetics, 2010, 87, 905-914.	2.6	64
123	The clinical picture of the BÃ¶rjeson-Forssman-Lehmann syndrome in males and heterozygous females with PHF6 mutations. Clinical Genetics, 2004, 65, 226-232.	1.0	63
124	Functional characterization of the AFF (AF4/FMR2) family of RNA-binding proteins: insights into the molecular pathology of FRAXE intellectual disability. Human Molecular Genetics, 2011, 20, 1873-1885.	1.4	63
125	Aristaless-related homeobox gene, the gene responsible for West syndrome and related disorders, is a Groucho/transducin-like enhancer of split dependent transcriptional repressor. Neuroscience, 2007, 146, 236-247.	1.1	62
126	Mutation detection in FGFR2 craniosynostosis syndromes. Human Genetics, 1997, 99, 251-255.	1.8	60

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127	X-linked mild non-syndromic mental retardation with neuropsychiatric problems and the missense mutation A365E in PAK3. , 2003, 120A, 509-517.		60
128	Disruption of a new X linked gene highly expressed in brain in a family with two mentally retarded males. Journal of Medical Genetics, 2004, 41, 736-742.	1.5	60
129	Folate-sensitive fragile site FRA10A is due to an expansion of a CGG repeat in a novel gene, FRA10AC1, encoding a nuclear protein. Genomics, 2004, 84, 69-81.	1.3	60
130	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	2.6	59
131	Cloning and characterization of a new human Xq13 gene, encoding a putative helicase. Human Molecular Genetics, 1994, 3, 1957-1964.	1.4	58
132	Three new families with X-linked mental retardation caused by the 428-451dup(24bp) mutation in ARX. Clinical Genetics, 2004, 66, 39-45.	1.0	58
133	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
134	O-GlcNAc transferase missense mutations linked to X-linked intellectual disability deregulate genes involved in cell fate determination and signaling. Journal of Biological Chemistry, 2018, 293, 10810-10824.	1.6	56
135	Splicing mutation in the ATR-X gene can lead to a dysmorphic mental retardation phenotype without alpha-thalassemia. American Journal of Human Genetics, 1996, 58, 499-505.	2.6	56
136	Fragile XE-associated familial mental retardation protein 2 (FMR2) acts as a potent transcription activator. Journal of Human Genetics, 2001, 46, 251-259.	1.1	55
137	Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. Human Genetics, 2006, 118, 578-590.	1.8	55
138	A Upf3b-mutant mouse model with behavioral and neurogenesis defects. Molecular Psychiatry, 2018, 23, 1773-1786.	4.1	54
139	FMR2 Expression in Families with Fraxe Mental Retardation. Human Molecular Genetics, 1997, 6, 435-441.	1.4	53
140	â€˜Big issuesâ€™ in neurodevelopment for children and adults with congenital heart disease. Open Heart, 2019, 6, e000998.	0.9	53
141	Expanding the molecular basis and phenotypic spectrum of X-linked Joubert syndrome associated with OFD1 mutations. European Journal of Human Genetics, 2012, 20, 806-809.	1.4	52
142	Pcdh19 Loss-of-Function Increases Neuronal Migration In Vitro but is Dispensable for Brain Development in Mice. Scientific Reports, 2016, 6, 26765.	1.6	52
143	New insights into Brunner syndrome and potential for targeted therapy. Clinical Genetics, 2016, 89, 120-127.	1.0	52
144	CCDC22: a novel candidate gene for syndromic X-linked intellectual disability. Molecular Psychiatry, 2012, 17, 4-7.	4.1	50

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145	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
146	The FMR2 gene, FRAXE and non-specific X-linked mental retardation: clinical and molecular aspects. Annals of Human Genetics, 2000, 64, 95-106.	0.3	49
147	A Recurrent RNA-Splicing Mutation in the SEDL Gene Causes X-Linked Spondyloepiphyseal Dysplasia Tarda. American Journal of Human Genetics, 2001, 68, 1398-1407.	2.6	49
148	HCFC1 loss-of-function mutations disrupt neuronal and neural progenitor cells of the developing brain. Human Molecular Genetics, 2015, 24, 3335-3347.	1.4	47
149	Cloning and expression of the murine homologue of a putative human X-linked nuclear protein gene closely linked to PGK1 in Xq13.3. Human Molecular Genetics, 1994, 3, 39-44.	1.4	46
150	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
151	Interchromosomal Insertional Translocation at Xq26.3 AltersSOX3Expression in an Individual With XX Male Sex Reversal. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E815-E820.	1.8	46
152	Gene Structure and Expression Study of the SEDL Gene for Spondyloepiphyseal Dysplasia Tarda. Genomics, 2000, 69, 242-251.	1.3	45
153	The BÃ¶rjesonâ€“Forssmanâ€“Lehman syndrome (BFLS, MIM #301900). European Journal of Human Genetics, 2006, 14, 1233-1237.	1.4	45
154	Protein and gene expression analysis of Phf6, the gene mutated in the BÃ¶rjesonâ€“Forssmanâ€“Lehmann Syndrome of intellectual disability and obesity. Gene Expression Patterns, 2007, 7, 858-871.	0.3	45
155	Eight further individuals with intellectual disability and epilepsy carrying bi-allelicCNTNAP2aberrations allow delineation of the mutational and phenotypic spectrum. Journal of Medical Genetics, 2016, 53, 820-827.	1.5	45
156	De novo and inherited mutations in the X-linked gene CLCN4 are associated with syndromic intellectual disability and behavior and seizure disorders in males and females. Molecular Psychiatry, 2018, 23, 222-230.	4.1	45
157	Mutation screening in Borjeson-Forssman-Lehmann syndrome: identification of a novel de novo PHF6 mutation in a female patient. Journal of Medical Genetics, 2005, 43, 238-243.	1.5	43
158	Xp11.2 microduplications including IQSEC2, TSPYL2 and KDM5C genes in patients with neurodevelopmental disorders. European Journal of Human Genetics, 2016, 24, 373-380.	1.4	43
159	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	5.8	43
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161	Molecular pathology of expanded polyalanine tract mutations in the Aristaless-related homeobox gene. Genomics, 2007, 90, 59-71.	1.3	42
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