Jozef Gecz

List of Publications by Citations

327 papers	18,736 citations	78 h-index	124 g-index
359 ext. papers	22,369 ext. citations	8.5 avg, IF	6.04 L-index

#	Paper	IF	Citations
327	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, 442-53	11	484
326	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. *Nature Genetics*, **2010**, 42, 203-9	36.3	461
325	A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. *Nature Genetics*, **2009**, 41, 535-43	36.3	454
324	Refining analyses of copy number variation identifies specific genes associated with developmental delay. *Nature Genetics*, **2014**, 46, 1063-71	36.3	429
323	Mutations in the human ortholog of Aristaless cause X-linked mental retardation and epilepsy. *Nature Genetics*, **2002**, 30, 441-5	36.3	356
322	Mutations of CDKL5 cause a severe neurodevelopmental disorder with infantile spasms and mental retardation. *American Journal of Human Genetics*, **2004**, 75, 1079-93	11	352
321	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. *Nature Genetics*, **2008**, 40, 776-81	36.3	328
320	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, 227-36	11	287
319	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. *Nature Genetics*, **2017**, 49, 515-526	36.3	283
318	Identification of the gene FMR2, associated with FRAXE mental retardation. *Nature Genetics*, **1996**, 13, 105-8	36.3	265
317	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. *Nature Genetics*, **2013**, 45, 546-51	36.3	238
316	A novel X-linked trichothiodystrophy associated with a nonsense mutation in RNF113A. *Journal of Medical Genetics*, **2015**, 52, 269-74	5.8	237
315	PHF6 mutations in T-cell acute lymphoblastic leukemia. *Nature Genetics*, **2010**, 42, 338-42	36.3	231
314	Disruption of the serine/threonine kinase 9 gene causes severe X-linked infantile spasms and mental retardation. *American Journal of Human Genetics*, **2003**, 72, 1401-11	11	227
313	Mutations in the X-linked cyclin-dependent kinase-like 5 (CDKL5/STK9) gene are associated with severe neurodevelopmental retardation. *American Journal of Human Genetics*, **2004**, 75, 1149-54	11	226
312	Identification of a microRNA that activates gene expression by repressing nonsense-mediated RNA decay. *Molecular Cell*, **2011**, 42, 500-10	17.6	211
311	ARX, a novel Prd-class-homeobox gene highly expressed in the telencephalon, is mutated in X-linked mental retardation. *Human Molecular Genetics*, **2002**, 11, 981-91	5.6	209

310	Cerebral palsy: causes, pathways, and the role of genetic variants. *American Journal of Obstetrics and Gynecology*, **2015**, 213, 779-88	6.4	206
309	PRRT2 mutations cause benign familial infantile epilepsy and infantile convulsions with choreoathetosis syndrome. *American Journal of Human Genetics*, **2012**, 90, 152-60	11	199
308	Mutations in UPF3B, a member of the nonsense-mediated mRNA decay complex, cause syndromic and nonsyndromic mental retardation. *Nature Genetics*, **2007**, 39, 1127-33	36.3	189
307	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. *American Journal of Human Genetics*, **2016**, 99, 287-98	11	180
306	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, 185-95	11	176
305	SLC9A6 mutations cause X-linked mental retardation, microcephaly, epilepsy, and ataxia, a phenotype mimicking Angelman syndrome. *American Journal of Human Genetics*, **2008**, 82, 1003-10	11	175
304	Mutations in PHF6 are associated with Börjeson-Forssman-Lehmann syndrome. *Nature Genetics*, **2002**, 32, 661-5	36.3	168
303	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. *Molecular Psychiatry*, **2016**, 21, 133-48	15.1	167
302	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. *Genome Biology*, **2016**, 17, 243	18.3	166
301	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, 432-43	11	164
300	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, 345-52	11	163
299	The genetic landscape of intellectual disability arising from chromosome X. *Trends in Genetics*, **2009**, 25, 308-16	8.5	162
298	Identification of the gene (SEDL) causing X-linked spondyloepiphyseal dysplasia tarda. *Nature Genetics*, **1999**, 22, 400-4	36.3	161
297	Mutations in mammalian target of rapamycin regulator DEPDC5 cause focal epilepsy with brain malformations. *Annals of Neurology*, **2014**, 75, 782-7	9.4	153
296	Epilepsy and mental retardation limited to females: an under-recognized disorder. *Brain*, **2008**, 131, 918-27	11.2	152
295	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. *Nature*, **2018**, 562, 268-271	50.4	149
294	Early onset seizures and Rett-like features associated with mutations in CDKL5. *European Journal of Human Genetics*, **2005**, 13, 1113-20	5.3	143
293	Disruption at the PTCHD1 Locus on Xp22.11 in Autism spectrum disorder and intellectual disability. *Science Translational Medicine*, **2010**, 2, 49ra68	17.5	140

292	Infantile spasms, dystonia, and other X-linked phenotypes caused by mutations in Aristaless related homeobox gene, ARX. *Brain and Development*, **2002**, 24, 266-8	2.2	137
291	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, 343-52	11	136
290	XLMR genes: update 2007. *European Journal of Human Genetics*, **2008**, 16, 422-34	5.3	135
289	Mutations in the DLG3 gene cause nonsyndromic X-linked mental retardation. *American Journal of Human Genetics*, **2004**, 75, 318-24	11	134
288	ARX spectrum disorders: making inroads into the molecular pathology. *Human Mutation*, **2010**, 31, 889-900	4.7	131
287	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	3.5	130
286	The original Lujan syndrome family has a novel missense mutation (p.N1007S) in the MED12 gene. *Journal of Medical Genetics*, **2007**, 44, 472-7	5.8	126
285	Mutations in the polyglutamine binding protein 1 gene cause X-linked mental retardation. *Nature Genetics*, **2003**, 35, 313-5	36.3	125
284	A missense mutation in RPS6KA3 (RSK2) responsible for non-specific mental retardation. *Nature Genetics*, **1999**, 22, 13-4	36.3	125
283	Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. *Human Mutation*, **2015**, 36, 1197-204	4.7	122
282	Rett syndrome: clinical review and genetic update. *Journal of Medical Genetics*, **2005**, 42, 1-7	5.8	121
281	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, 982-7	11	120
280	Mutations in the guanine nucleotide exchange factor gene IQSEC2 cause nonsyndromic intellectual disability. *Nature Genetics*, **2010**, 42, 486-8	36.3	118
279	Oligosaccharyltransferase-subunit mutations in nonsyndromic mental retardation. *American Journal of Human Genetics*, **2008**, 82, 1150-7	11	116
278	Whole-exome sequencing points to considerable genetic heterogeneity of cerebral palsy. *Molecular Psychiatry*, **2015**, 20, 176-82	15.1	115
277	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, 444-53	5.3	111
276	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*, **2007**, 16, 265-75	5.6	111
275	ARX: a gene for all seasons. *Current Opinion in Genetics and Development*, **2006**, 16, 308-16	4.9	109

274	La FAM fatale: USP9X in development and disease. *Cellular and Molecular Life Sciences*, **2015**, 72, 2075-89	10.3	107
273	A focal epilepsy and intellectual disability syndrome is due to a mutation in TBC1D24. *American Journal of Human Genetics*, **2010**, 87, 371-5	11	100
272	Mutations in the FTSJ1 gene coding for a novel S-adenosylmethionine-binding protein cause nonsyndromic X-linked mental retardation. *American Journal of Human Genetics*, **2004**, 75, 305-9	11	99
271	Variable expression of mental retardation, autism, seizures, and dystonic hand movements in two families with an identical ARX gene mutation. *American Journal of Medical Genetics Part A*, **2002**, 112, 405-11		98
270	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. *Nature Neuroscience*, **2017**, 20, 1043-1051	25.5	94
269	Fine-scale survey of X chromosome copy number variants and indels underlying intellectual disability. *American Journal of Human Genetics*, **2010**, 87, 173-88	11	93
268	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, 1119-24	11	92
267	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, 1120-30	11	92
266	The genetic basis of cerebral palsy. *Developmental Medicine and Child Neurology*, **2017**, 59, 462-469	3.3	91
265	Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. *Molecular Psychiatry*, **2016**, 21, 126-32	15.1	90
264	Defects in tRNA Anticodon Loop 2'-O-Methylation Are Implicated in Nonsyndromic X-Linked Intellectual Disability due to Mutations in FTSJ1. *Human Mutation*, **2015**, 36, 1176-87	4.7	90
263	XNP mutation in a large family with Juberg-Marsidi syndrome. *Nature Genetics*, **1996**, 12, 359-60	36.3	90
262	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. *American Journal of Human Genetics*, **2016**, 98, 541-552	11	89
261	Mutation frequencies of X-linked mental retardation genes in families from the EuroMRX consortium. *Human Mutation*, **2007**, 28, 207-8	4.7	89
260	A UPF3-mediated regulatory switch that maintains RNA surveillance. *Nature Structural and Molecular Biology*, **2009**, 16, 747-53	17.6	88
259	A mutation in the Golgi Qb-SNARE gene GOSR2 causes progressive myoclonus epilepsy with early ataxia. *American Journal of Human Genetics*, **2011**, 88, 657-63	11	85
258	CASK mutations are frequent in males and cause X-linked nystagmus and variable XLMR phenotypes. *European Journal of Human Genetics*, **2010**, 18, 544-52	5.3	85
257	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, 767-76	15.1	85

256	Nonsense-mediated mRNA decay: inter-individual variability and human disease. *Neuroscience and Biobehavioral Reviews*, **2014**, 46 Pt 2, 175-86	9	83
255	Contribution of copy number variants involving nonsense-mediated mRNA decay pathway genes to neuro-developmental disorders. *Human Molecular Genetics*, **2013**, 22, 1816-25	5.6	82
254	Mutations in USP9X are associated with X-linked intellectual disability and disrupt neuronal cell migration and growth. *American Journal of Human Genetics*, **2014**, 94, 470-8	11	79
253	The molecular basis of X-linked spondyloepiphyseal dysplasia tarda. *American Journal of Human Genetics*, **2001**, 68, 1386-97	11	79
252	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. *Human Molecular Genetics*, **2015**, 24, 5250-9	5.6	78
251	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. *American Journal of Human Genetics*, **2017**, 100, 907-925	11	77
250	TBC1D24 genotype-phenotype correlation: Epilepsies and other neurologic features. *Neurology*, **2016**, 87, 77-85	6.5	75
249	X-linked myoclonic epilepsy with spasticity and intellectual disability: mutation in the homeobox gene ARX. *Neurology*, **2002**, 59, 348-56	6.5	75
248	Characterization of the human glutamate receptor subunit 3 gene (GRIA3), a candidate for bipolar disorder and nonspecific X-linked mental retardation. *Genomics*, **1999**, 62, 356-68	4.3	75
247	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, 4673-87	5.6	74
246	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, 18163-8	11.5	74
245	Transcriptome profiling of UPF3B/NMD-deficient lymphoblastoid cells from patients with various forms of intellectual disability. *Molecular Psychiatry*, **2012**, 17, 1103-15	15.1	71
244	Biallelic SUN5 Mutations Cause Autosomal-Recessive Acephalic Spermatozoa Syndrome. *American Journal of Human Genetics*, **2016**, 99, 942-949	11	70
243	A noncoding, regulatory mutation implicates HCFC1 in nonsyndromic intellectual disability. *American Journal of Human Genetics*, **2012**, 91, 694-702	11	69
242	Fibroblast growth factor homologous factor 2 (FHF2): gene structure, expression and mapping to the BEjeson-Forssman-Lehmann syndrome region in Xq26 delineated by a duplication breakpoint in a BFLS-like patient. *Human Genetics*, **1999**, 104, 56-63	6.3	69
241	ZNF674: a new kruppel-associated box-containing zinc-finger gene involved in nonsyndromic X-linked mental retardation. *American Journal of Human Genetics*, **2006**, 78, 265-78	11	67
240	Mutations in the BRWD3 gene cause X-linked mental retardation associated with macrocephaly. *American Journal of Human Genetics*, **2007**, 81, 367-74	11	66
239	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, 211-6	5.8	64

238	CCDC22 deficiency in humans blunts activation of proinflammatory NF-B signaling. *Journal of Clinical Investigation*, **2013**, 123, 2244-56	15.9	64
237	FOXP1 mutations cause intellectual disability and a recognizable phenotype. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3166-75	2.5	63
236	Two novel JAK2 exon 12 mutations in JAK2V617F-negative polycythaemia vera patients. *Leukemia*, **2008**, 22, 870-3	10.7	63
235	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, 631-9	2.9	61
234	Nonsyndromic X-linked mental retardation: where are the missing mutations?. *Trends in Genetics*, **2003**, 19, 316-20	8.5	61
233	Evolution of the human X--a smart and sexy chromosome that controls speciation and development. *Cytogenetic and Genome Research*, **2002**, 99, 141-5	1.9	60
232	Identification and characterization of a missense mutation in the -linked acetylglucosamine (-GlcNAc) transferase gene that segregates with X-linked intellectual disability. *Journal of Biological Chemistry*, **2017**, 292, 8948-8963	5.4	58
231	Abnormal Cell Sorting Underlies the Unique X-Linked Inheritance of PCDH19 Epilepsy. *Neuron*, **2018**, 97, 59-66.e5	13.9	58
230	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. *Nature Communications*, **2019**, 10, 4919	17.4	58
229	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, 652-9	5.3	57
228	Gene structure and subcellular localization of FMR2, a member of a new family of putative transcription activators. *Genomics*, **1997**, 44, 201-13	4.3	56
227	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, 236-47	3.9	56
226	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, 373-81	11	55
225	X-linked mild non-syndromic mental retardation with neuropsychiatric problems and the missense mutation A365E in PAK3 **2003**, 120A, 509-17		55
224	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, 681-95	11	54
223	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, 1269-79	20.1	53
222	Loss of Usp9x disrupts cortical architecture, hippocampal development and TGF-mediated axonogenesis. *PLoS ONE*, **2013**, 8, e68287	3.7	53
221	A ubiquitin-dependent signalling axis specific for ALKBH-mediated DNA dealkylation repair. *Nature*, **2017**, 551, 389-393	50.4	52

220	Three new families with X-linked mental retardation caused by the 428-451dup(24bp) mutation in ARX. *Clinical Genetics*, **2004**, 66, 39-45	4	52
219	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. *American Journal of Human Genetics*, **2020**, 106, 356-370	11	51
218	Rare copy number variation in cerebral palsy. *European Journal of Human Genetics*, **2014**, 22, 40-5	5.3	51
217	The clinical picture of the B□jeson-Forssman-Lehmann syndrome in males and heterozygous females with PHF6 mutations. *Clinical Genetics*, **2004**, 65, 226-32	4	51
216	Genes for cognitive function: developments on the X. *Genome Research*, **2000**, 10, 157-63	9.7	51
215	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	11	51
214	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, 736-42	5.8	50
213	Fragile XE-associated familial mental retardation protein 2 (FMR2) acts as a potent transcription activator. *Journal of Human Genetics*, **2001**, 46, 251-9	4.3	50
212	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. *Neurology*, **2016**, 87, 1975-1984	6.5	50
211	Seizures are regulated by ubiquitin-specific peptidase 9 X-linked (USP9X), a de-ubiquitinase. *PLoS Genetics*, **2015**, 11, e1005022	6	49
210	M□orth SeaM□progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. *Brain*, **2013**, 136, 1146-54	11.2	49
209	Mutation detection in FGFR2 craniosynostosis syndromes. *Human Genetics*, **1997**, 99, 251-5	6.3	49
208	MCT8 mutation analysis and identification of the first female with Allan-Herndon-Dudley syndrome due to loss of MCT8 expression. *European Journal of Human Genetics*, **2008**, 16, 1029-37	5.3	49
207	Cloning and characterization of a new human Xq13 gene, encoding a putative helicase. *Human Molecular Genetics*, **1994**, 3, 1957-64	5.6	49
206	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. *Nature Communications*, **2019**, 10, 4920	17.4	48
205	Identification and characterization of two novel JARID1C mutations: suggestion of an emerging genotype-phenotype correlation. *European Journal of Human Genetics*, **2010**, 18, 330-5	5.3	48
204	FMR2 expression in families with FRAXE mental retardation. *Human Molecular Genetics*, **1997**, 6, 435-41	5.6	48
203	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	4.3	48

202	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	15.1	47
201	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, 1873-85	5.6	46
200	Hypomorphic temperature-sensitive alleles of NSDHL cause CK syndrome. *American Journal of Human Genetics*, **2010**, 87, 905-14	11	46
199	Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. *Human Genetics*, **2006**, 118, 578-90	6.3	44
198	The FMR2 gene, FRAXE and non-specific X-linked mental retardation: clinical and molecular aspects. *Annals of Human Genetics*, **2000**, 64, 95-106	2.2	44
197	A recurrent RNA-splicing mutation in the SEDL gene causes X-linked spondyloepiphyseal dysplasia tarda. *American Journal of Human Genetics*, **2001**, 68, 1398-407	11	44
196	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	11	44
195	Expanding the molecular basis and phenotypic spectrum of X-linked Joubert syndrome associated with OFD1 mutations. *European Journal of Human Genetics*, **2012**, 20, 806-9	5.3	43
194	Genetic or Other Causation Should Not Change the Clinical Diagnosis of Cerebral Palsy. *Journal of Child Neurology*, **2019**, 34, 472-476	2.5	42
193	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, 539-47	6.3	40
192	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	5.6	39
191	Molecular pathology of expanded polyalanine tract mutations in the Aristaless-related homeobox gene. *Genomics*, **2007**, 90, 59-71	4.3	38
190	The B□jeson-Forssman-Lehman syndrome (BFLS, MIM #301900). *European Journal of Human Genetics*, **2006**, 14, 1233-7	5.3	38
189	Gene structure and expression study of the SEDL gene for spondyloepiphyseal dysplasia tarda. *Genomics*, **2000**, 69, 242-51	4.3	38
188	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. *Nature Genetics*, **2020**, 52, 1046-1056	30.9	38
187	Interchromosomal insertional translocation at Xq26.3 alters SOX3 expression in an individual with XX male sex reversal. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E815-20	5.6	37
186	Protein and gene expression analysis of Phf6, the gene mutated in the B□jeson-Forssman-Lehmann Syndrome of intellectual disability and obesity. *Gene Expression Patterns*, **2007**, 7, 858-71	1.5	37
185	Mutation screening in Borjeson-Forssman-Lehmann syndrome: identification of a novel de novo PHF6 mutation in a female patient. *Journal of Medical Genetics*, **2006**, 43, 238-43	5.8	36

184	Pcdh19 Loss-of-Function Increases Neuronal Migration In Vitro but is Dispensable for Brain Development in Mice. *Scientific Reports*, **2016**, 6, 26765	4.9	36
183	Identification of three novel SEDL mutations, including mutation in the rare, non-canonical splice site of exon 4. *Clinical Genetics*, **2003**, 64, 235-42	4	35
182	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. *American Journal of Human Genetics*, **2015**, 97, 302-10	11	34
181	New insights into Brunner syndrome and potential for targeted therapy. *Clinical Genetics*, **2016**, 89, 120-7	4	34
180	Nance-Horan syndrome protein, NHS, associates with epithelial cell junctions. *Human Molecular Genetics*, **2006**, 15, 1972-83	5.6	34
179	Identification of a mutation in the XNP/ATR-X gene in a family reported as Smith-Fineman-Myers syndrome **2000**, 91, 83-85		34
178	Mutations in the intellectual disability gene KDM5C reduce protein stability and demethylase activity. *Human Molecular Genetics*, **2015**, 24, 2861-72	5.6	33
177	Overlapping submicroscopic deletions in Xq28 in two unrelated boys with developmental disorders: identification of a gene near FRAXE. *American Journal of Human Genetics*, **1995**, 56, 907-14	11	33
176	Xp11.2 microduplications including IQSEC2, TSPYL2 and KDM5C genes in patients with neurodevelopmental disorders. *European Journal of Human Genetics*, **2016**, 24, 373-80	5.3	32
175	A Upf3b-mutant mouse model with behavioral and neurogenesis defects. *Molecular Psychiatry*, **2018**, 23, 1773-1786	15.1	32
174	-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	5.4	32
173	CCDC22: a novel candidate gene for syndromic X-linked intellectual disability. *Molecular Psychiatry*, **2012**, 17, 4-7	15.1	31
172	HCFC1 loss-of-function mutations disrupt neuronal and neural progenitor cells of the developing brain. *Human Molecular Genetics*, **2015**, 24, 3335-47	5.6	30
171	Human wild-type SEDL protein functionally complements yeast Trs20p but some naturally occurring SEDL mutants do not. *Gene*, **2003**, 320, 137-44	3.8	30
170	A regulatory path associated with X-linked intellectual disability and epilepsy links KDM5C to the polyalanine expansions in ARX. *American Journal of Human Genetics*, **2013**, 92, 114-25	11	29
169	Cyclin-dependent kinase-like 5 (CDKL5) mutation screening in Rett syndrome and related disorders. *Twin Research and Human Genetics*, **2010**, 13, 168-78	2.2	29
168	HUWE1 mutations in Juberg-Marsidi and Brooks syndromes: the results of an X-chromosome exome sequencing study. *BMJ Open*, **2016**, 6, e009537	3	29
167	Severe childhood speech disorder: Gene discovery highlights transcriptional dysregulation. *Neurology*, **2020**, 94, e2148-e2167	6.5	28

166	Subtle functional defects in the Arf-specific guanine nucleotide exchange factor IQSEC2 cause non-syndromic X-linked intellectual disability. *Small GTPases*, **2010**, 1, 98-103	2.7	28
165	Eight further individuals with intellectual disability and epilepsy carrying bi-allelic CNTNAP2 aberrations allow delineation of the mutational and phenotypic spectrum. *Journal of Medical Genetics*, **2016**, 53, 820-827	5.8	28
164	Reduced steroidogenesis in patients with PCDH19-female limited epilepsy. *Epilepsia*, **2017**, 58, e91-e95	6.4	27
163	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	11	26
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