Jozef Gecz

List of Publications by Year in Descending Order

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	Oligonucleotide correction of an intronic TIMMDC1 variant in cells of patients with severe neurodegenerative disorder.. *Npj Genomic Medicine*, **2022**, 7, 9	6.2	2
326	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy.. *Neurology: Genetics*, **2022**, 8, e652	3.8	1
325	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders.. *Human Genetics and Genomics Advances*, **2022**, 3, 100075	0.8	1
324	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. *American Journal of Human Genetics*, **2021**, 108, 2006-2016	11	3
323	Whole genome sequencing in transposition of the great arteries and associations with clinically relevant heart, brain and laterality genes. *American Heart Journal*, **2021**, 244, 1-13	4.9	1
322	Transgenic mice with an R342X mutation in Phf6 display clinical features of B□jeson-Forssman-Lehmann Syndrome. *Human Molecular Genetics*, **2021**, 30, 575-594	5.6	0
321	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. *Genome Medicine*, **2021**, 13, 63	14.4	9
320	A 127 kb truncating deletion of PGRMC1 is a novel cause of X-linked isolated paediatric cataract. *European Journal of Human Genetics*, **2021**, 29, 1206-1215	5.3	1
319	Different types of disease-causing noncoding variants revealed by genomic and gene expression analyses in families with X-linked intellectual disability. *Human Mutation*, **2021**, 42, 835-847	4.7	
318	Integrated in silico and experimental assessment of disease relevance of PCDH19□missense variants. *Human Mutation*, **2021**, 42, 1030-1041	4.7	
317	In-depth analysis reveals complex molecular aetiology in a cohort of idiopathic cerebral palsy. *Brain*, **2021**,	11.2	3
316	Cerebral palsy with autism and ADHD: time to pay attention. *Developmental Medicine and Child Neurology*, **2021**, 63, 247-248	3.3	2
315	Disrupted Excitatory Synaptic Contacts and Altered Neuronal Network Activity Underpins the Neurological Phenotype in PCDH19-Clustering Epilepsy (PCDH19-CE). *Molecular Neurobiology*, **2021**, 58, 2005-2018	6.2	6
314	Human disease genes website series: An international, open and dynamic library for up-to-date clinical information. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1039-1046	2.5	4
313	Further delineation of BCAP31-linked intellectual disability: description of 17 new families with LoF and missense variants. *European Journal of Human Genetics*, **2021**, 29, 1405-1417	5.3	0
312	Association of Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. *Neurology*, **2021**, 96, e2251-e2260	6.5	3
311	Simultaneous Screening of the FRAXA and FRAXE Loci for Rapid Detection of FMR1 CGG and/or AFF2 CCG Repeat Expansions by Triplet-Primed PCR. *Journal of Molecular Diagnostics*, **2021**, 23, 941-951	5.1	0

310	People with Cerebral Palsy and Their Family's Preferences about Genomics Research. *Public Health Genomics*, **2021**, 1-10	1.9	
309	Yield of clinically reportable genetic variants in unselected cerebral palsy by whole genome sequencing. *Npj Genomic Medicine*, **2021**, 6, 74	6.2	0
308	Protocadherin 19 Clustering Epilepsy and Neurosteroids: Opportunities for Intervention. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	2
307	Missense variant contribution to USP9X-female syndrome. *Npj Genomic Medicine*, **2020**, 5, 53	6.2	3
306	RLIM Is a Candidate Dosage-Sensitive Gene for Individuals with Varying Duplications of Xq13, Intellectual Disability, and Distinct Facial Features. *American Journal of Human Genetics*, **2020**, 107, 1157-1169	11	1
305	Constraint and conservation of paired-type homeodomains predicts the clinical outcome of missense variants of uncertain significance. *Human Mutation*, **2020**, 41, 1407-1424	4.7	1
304	A standardized patient-centered characterization of the phenotypic spectrum of PCDH19 girls clustering epilepsy. *Translational Psychiatry*, **2020**, 10, 127	8.6	12
303	Definition and diagnosis of cerebral palsy in genetic studies: a systematic review. *Developmental Medicine and Child Neurology*, **2020**, 62, 1024-1030	3.3	9
302	Familial adult myoclonic epilepsy type 1 SAMD12 TTTCA repeat expansion arose 17,000 years ago and is present in Sri Lankan and Indian families. *European Journal of Human Genetics*, **2020**, 28, 973-978	5.3	9
301	Chromatin-Binding Protein PHF6 Regulates Activity-Dependent Transcriptional Networks to Promote Hunger Response. *Cell Reports*, **2020**, 30, 3717-3728.e6	10.6	5
300	Expanding Clinical Presentations Due to Variations in THOC2 mRNA Nuclear Export Factor. *Frontiers in Molecular Neuroscience*, **2020**, 13, 12	6.1	4
299	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
298	An intellectual disability syndrome with single-nucleotide variants in O-GlcNAc transferase. *European Journal of Human Genetics*, **2020**, 28, 706-714	5.3	17
297	Severe childhood speech disorder: Gene discovery highlights transcriptional dysregulation. *Neurology*, **2020**, 94, e2148-e2167	6.5	28
296	Downregulation of the GHRH/GH/IGF1 axis in a mouse model of Börjeson-Forssman-Lehman syndrome. *Development (Cambridge)*, **2020**, 147,	6.6	1
295	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. *Nature Communications*, **2020**, 11, 4932	17.4	25
294	X-linked intellectual disability: Phenotypic expression in carrier females. *Clinical Genetics*, **2020**, 97, 418-425	4	6
293	Levetiracetam efficacy in PCDH19 Girls Clustering Epilepsy. *European Journal of Paediatric Neurology*, **2020**, 24, 142-147	3.8	9

292	A Survey of Rare Epigenetic Variation in 23,116 Human Genomes Identifies Disease-Relevant Epivariations and CGG Expansions. *American Journal of Human Genetics*, **2020**, 107, 654-669	11	12
291	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. *Nature Genetics*, **2020**, 52, 1046-1056	30.9	38
290	A synonymous UPF3B variant causing a speech disorder implicates NMD as a regulator of neurodevelopmental disorder gene networks. *Human Molecular Genetics*, **2020**, 29, 2568-2578	5.6	4
289	Two novel intragenic variants in the FMR1 gene in patients with suspect clinical diagnosis of Fragile X syndrome and no CGG repeat expansion. *European Journal of Medical Genetics*, **2020**, 63, 104010	2.6	2
288	Disentangling the paradox of the PCDH19 clustering epilepsy, a disorder of cellular mosaics. *Current Opinion in Genetics and Development*, **2020**, 65, 169-175	4.9	12
287	PCDH19 Pathogenic Variants in Males: Expanding the Phenotypic Spectrum. *Advances in Experimental Medicine and Biology*, **2020**, 1298, 177-187	3.6	3
286	EXOME REPORT: Novel mutation in ATP6V1B2 segregating with autosomal dominant epilepsy, intellectual disability and mild gingival and nail abnormalities. *European Journal of Medical Genetics*, **2020**, 63, 103799	2.6	8
285	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Signaling. *Biological Psychiatry*, **2020**, 87, 100-112	7.9	19
284	Disruptive variants of associate with autism and interfere with neuronal development and synaptic transmission. *Science Advances*, **2019**, 5, eaax2166	14.3	16
283	Inhibition of Upf2-Dependent Nonsense-Mediated Decay Leads to Behavioral and Neurophysiological Abnormalities by Activating the Immune Response. *Neuron*, **2019**, 104, 665-679.e8	13.9	16
282	Novel mutations in NLGN3 causing autism spectrum disorder and cognitive impairment. *Human Mutation*, **2019**, 40, 2021-2032	4.7	23
281	Genetic or Other Causation Should Not Change the Clinical Diagnosis of Cerebral Palsy. *Journal of Child Neurology*, **2019**, 34, 472-476	2.5	42
280	Dysregulations of sonic hedgehog signaling in MED12-related X-linked intellectual disability disorders. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00569	2.3	9
279	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
278	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. *Nature Communications*, **2019**, 10, 4679	17.4	21
277	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. *Nature Communications*, **2019**, 10, 4920	17.4	48
276	Histone demethylase KDM5C is a SAHA-sensitive central hub at the crossroads of transcriptional axes involved in multiple neurodevelopmental disorders. *Human Molecular Genetics*, **2019**, 28, 4089-4102	5.6	8
275	Heterozygous loss of function of / leads to increased activated Arf6 and severe neurocognitive seizure phenotype in females. *Life Science Alliance*, **2019**, 2,	5.8	13

274	PHF6 regulates hematopoietic stem and progenitor cells and its loss synergizes with expression of TLX3 to cause leukemia. *Blood*, **2019**, 133, 1729-1741	2.2	18
273	Big issues in neurodevelopment for children and adults with congenital heart disease. *Open Heart*, **2019**, 6, e000998	3	21
272	Targeted resequencing identifies genes with recurrent variation in cerebral palsy. *Npj Genomic Medicine*, **2019**, 4, 27	6.2	15
271	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. *Nature Communications*, **2019**, 10, 4919	17.4	58
270	A mouse model for intellectual disability caused by mutations in the X-linked 2'-O-methyltransferase Ftsj1 gene. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2019**, 1865, 2083-2093	6.9	12
269	Clinical and functional characterization of recurrent missense variants implicated in THOC6-related intellectual disability. *Human Molecular Genetics*, **2019**, 28, 952-960	5.6	9
268	A recurrent missense variant in SLC9A7 causes nonsyndromic X-linked intellectual disability with alteration of Golgi acidification and aberrant glycosylation. *Human Molecular Genetics*, **2019**, 28, 598-614	5.6	16
267	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. *Genetics in Medicine*, **2019**, 21, 1058-1064	8.1	12
266	Pathogenic variants in E3 ubiquitin ligase RLIM/RNF12 lead to a syndromic X-linked intellectual disability and behavior disorder. *Molecular Psychiatry*, **2019**, 24, 1748-1768	15.1	10
265	Analysis of 182 cerebral palsy transcriptomes points to dysregulation of trophic signalling pathways and overlap with autism. *Translational Psychiatry*, **2018**, 8, 88	8.6	16
264	Targeted knockout of a chemokine-like gene increases anxiety and fear responses. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, E1041-E1050	11.5	24
263	Cerebral palsy and genomics: an international consortium. *Developmental Medicine and Child Neurology*, **2018**, 60, 209-210	3.3	11
262	Abnormal Cell Sorting Underlies the Unique X-Linked Inheritance of PCDH19 Epilepsy. *Neuron*, **2018**, 97, 59-66.e5	13.9	58
261	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
260	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	15.1	20
259	A Upf3b-mutant mouse model with behavioral and neurogenesis defects. *Molecular Psychiatry*, **2018**, 23, 1773-1786	15.1	32
258	Severe neurocognitive and growth disorders due to variation in THOC2, an essential component of nuclear mRNA export machinery. *Human Mutation*, **2018**, 39, 1126-1138	4.7	8
257	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. *European Journal of Human Genetics*, **2018**, 26, 54-63	5.3	23

256	Non-Syndromic 46,XY Disorders of Sex Development. *Acta Medica Martiniana*, **2018**, 18, 35-41	0.2	
255	Regulating transcriptional activity by phosphorylation: A new mechanism for the ARX homeodomain transcription factor. *PLoS ONE*, **2018**, 13, e0206914	3.7	9
254	Pathogenic copy number variants that affect gene expression contribute to genomic burden in cerebral palsy. *Npj Genomic Medicine*, **2018**, 3, 33	6.2	16
253	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. *Nature*, **2018**, 562, 268-271	50.4	149
252	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
251	-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
250	PCDH19 regulation of neural progenitor cell differentiation suggests asynchrony of neurogenesis as a mechanism contributing to PCDH19 Girls Clustering Epilepsy. *Neurobiology of Disease*, **2018**, 116, 106-119	7.5	25
249	A genomic cause of cerebral palsy should not change the clinical classification. *Annals of Clinical and Translational Neurology*, **2018**, 5, 1011	5.3	6
248	Robust imaging and gene delivery to study human lymphoblastoid cell lines. *Journal of Human Genetics*, **2018**, 63, 945-955	4.3	2
247	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. *Nature Genetics*, **2017**, 49, 515-526	36.3	283
246	Protocadherin 19 (PCDH19) interacts with paraspeckle protein NONO to co-regulate gene expression with estrogen receptor alpha (ER) *Human Molecular Genetics*, **2017**, 26, 2042-2052	5.6	19
245	Reduced steroidogenesis in patients with PCDH19-female limited epilepsy. *Epilepsia*, **2017**, 58, e91-e95	6.4	27
244	Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. *European Journal of Medical Genetics*, **2017**, 60, 437-443	2.6	9
243	Variant in the X-chromosome spliceosomal gene GPKOW causes male-lethal microcephaly with intrauterine growth restriction. *European Journal of Human Genetics*, **2017**, 25, 1078-1082	5.3	6
242	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. *Nature Neuroscience*, **2017**, 20, 1043-1051	25.5	94
241	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. *American Journal of Human Genetics*, **2017**, 100, 907-925	11	77
240	MED12-related XLID disorders are dose-dependent of immediate early genes (IEGs) expression. *Human Molecular Genetics*, **2017**, 26, 2062-2075	5.6	17
239	USP9X deubiquitylating enzyme maintains RAPTOR protein levels, mTORC1 signalling and proliferation in neural progenitors. *Scientific Reports*, **2017**, 7, 391	4.9	17

238	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
237	The genetic basis of cerebral palsy. *Developmental Medicine and Child Neurology*, **2017**, 59, 462-469	3.3	91
236	A Recurrent De Novo Nonsense Variant in ZSWIM6 Results in Severe Intellectual Disability without Frontonasal or Limb Malformations. *American Journal of Human Genetics*, **2017**, 101, 995-1005	11	18
235	A ubiquitin-dependent signalling axis specific for ALKBH-mediated DNA dealkylation repair. *Nature*, **2017**, 551, 389-393	50.4	52
234	X-Linked Lissencephaly With Absent Corpus Callosum and Abnormal Genitalia: An Evolving Multisystem Syndrome With Severe Congenital Intestinal Diarrhea Disease. *Child Neurology Open*, **2017**, 4, 2329048X17738625	1.3	4
233	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
232	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
231	Protocadherin Mutations in Neurodevelopmental Disorders **2016**, 221-231		1
230	Biallelic SUN5 Mutations Cause Autosomal-Recessive Acephalic Spermatozoa Syndrome. *American Journal of Human Genetics*, **2016**, 99, 942-949	11	70
229	Embryonic forebrain transcriptome of mice with polyalanine expansion mutations in the ARX homeobox gene. *Human Molecular Genetics*, **2016**, 25, 5433-5443	5.6	7
228	TBC1D24 genotype-phenotype correlation: Epilepsies and other neurologic features. *Neurology*, **2016**, 87, 77-85	6.5	75
227	Reply. *American Journal of Obstetrics and Gynecology*, **2016**, 214, 671	6.4	
226	A mutation in COL4A2 causes autosomal dominant porencephaly with cataracts. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1059-63	2.5	13
225	Identification of an IGSF1-specific deletion in a five-generation pedigree with X-linked Central Hypothyroidism without macroorchidism. *Clinical Endocrinology*, **2016**, 85, 609-15	3.4	25
224	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. *Molecular Psychiatry*, **2016**, 21, 133-48	15.1	167
223	Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. *Molecular Psychiatry*, **2016**, 21, 126-32	15.1	90
222	Multiplex families with epilepsy: Success of clinical and molecular genetic characterization. *Neurology*, **2016**, 86, 713-22	6.5	22
221	New insights into Brunner syndrome and potential for targeted therapy. *Clinical Genetics*, **2016**, 89, 120-4		34

220	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
219	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. *American Journal of Human Genetics*, **2016**, 98, 541-552	11	89
218	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2-2q11.2. *Human Genetics*, **2016**, 135, 1117-25	6.3	16
217	HUWE1 mutations in Juberg-Marsidi and Brooks syndromes: the results of an X-chromosome exome sequencing study. *BMJ Open*, **2016**, 6, e009537	3	29
216	Is FGF13 a major contributor to genetic epilepsy with febrile seizures plus?. *Epilepsy Research*, **2016**, 128, 48-51	3	6
215	Disorders of sex development: insights from targeted gene sequencing of a large international patient cohort. *Genome Biology*, **2016**, 17, 243	18.3	166
214	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
213	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
212	A non-coding variant in the 5′UTR of DLG3 attenuates protein translation to cause non-syndromic intellectual disability. *European Journal of Human Genetics*, **2016**, 24, 1612-1616	5.3	9
211	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. *Neurology*, **2016**, 87, 1975-1984	6.5	50
210	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. *American Journal of Human Genetics*, **2016**, 99, 287-98	11	180
209	Mutations in the intellectual disability gene KDM5C reduce protein stability and demethylase activity. *Human Molecular Genetics*, **2015**, 24, 2861-72	5.6	33
208	La FAM fatale: USP9X in development and disease. *Cellular and Molecular Life Sciences*, **2015**, 72, 2075-89	10.3	107
207	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. *Human Molecular Genetics*, **2015**, 24, 2000-10	5.6	14
206	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
205	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. *American Journal of Human Genetics*, **2015**, 97, 302-10	11	34
204	Developmental disorders: deciphering exomes on a grand scale. *Lancet, The*, **2015**, 385, 1266-7	40	2
203	Identical by descent L1CAM mutation in two apparently unrelated families with intellectual disability without L1 syndrome. *European Journal of Medical Genetics*, **2015**, 58, 364-8	2.6	11

202	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. *Human Molecular Genetics*, **2015**, 24, 5250-9	5.6	78
201	Seizures are regulated by ubiquitin-specific peptidase 9 X-linked (USP9X), a de-ubiquitinase. *PLoS Genetics*, **2015**, 11, e1005022	6	49
200	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
199	Copy number variants in patients with intellectual disability affect the regulation of ARX transcription factor gene. *Human Genetics*, **2015**, 134, 1163-82	6.3	9
198	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
197	Novel IL1RAPL1 mutations associated with intellectual disability impair synaptogenesis. *Human Molecular Genetics*, **2015**, 24, 1106-18	5.6	23
196	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
195	Avascular necrosis of bone in childhood cancer patients: a possible role of genetic susceptibility. *Bratislava Medical Journal*, **2015**, 116, 289-95	1.7	2
194	Cerebral palsy: causes, pathways, and the role of genetic variants. *American Journal of Obstetrics and Gynecology*, **2015**, 213, 779-88	6.4	206
193	Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. *Human Mutation*, **2015**, 36, 1197-204	4.7	122
192	Increased STAG2 dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. *Human Molecular Genetics*, **2015**, 24, 7171-81	5.6	24
191	Whole-exome sequencing points to considerable genetic heterogeneity of cerebral palsy. *Molecular Psychiatry*, **2015**, 20, 176-82	15.1	115
190	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
189	A novel X-linked trichothiodystrophy associated with a nonsense mutation in RNF113A. *Journal of Medical Genetics*, **2015**, 52, 269-74	5.8	237
188	Nonsense-mediated mRNA decay: inter-individual variability and human disease. *Neuroscience and Biobehavioral Reviews*, **2014**, 46 Pt 2, 175-86	9	83
187	Refining analyses of copy number variation identifies specific genes associated with developmental delay. *Nature Genetics*, **2014**, 46, 1063-71	36.3	429
186	Phenotype-genotype complexities: opening DOORS. *Lancet Neurology, The*, **2014**, 13, 24-5	24.1	
185	FRA2A is a CGG repeat expansion associated with silencing of AFF3. *PLoS Genetics*, **2014**, 10, e1004242	6	26

184	Mutations in mammalian target of rapamycin regulator DEPDC5 cause focal epilepsy with brain malformations. *Annals of Neurology*, **2014**, 75, 782-7	9.4	153
183	Reduced polyalanine-expanded Arx mutant protein in developing mouse subpallium alters Lmo1 transcriptional regulation. *Human Molecular Genetics*, **2014**, 23, 1084-94	5.6	17
182	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
181	Rare copy number variation in cerebral palsy. *European Journal of Human Genetics*, **2014**, 22, 40-5	5.3	51
180	UPF3B Gene and Nonsense-Mediated mRNA Decay in Autism Spectrum Disorders **2014**, 1663-1678		3
179	NKX2-1 mutation in a family diagnosed with ataxic dyskinetic cerebral palsy. *European Journal of Medical Genetics*, **2013**, 56, 506-9	2.6	13
178	Loss of FMR2 further emphasizes the link between deregulation of immediate early response genes FOS and JUN and intellectual disability. *Human Molecular Genetics*, **2013**, 22, 2984-91	5.6	8
177	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
176	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
175	TBC1D24 mutation associated with focal epilepsy, cognitive impairment and a distinctive cerebro-cerebellar malformation. *Epilepsy Research*, **2013**, 105, 240-4	3	26
174	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
173	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. *Nature Genetics*, **2013**, 45, 546-51	36.3	238
172	PCR amplification and sequence analysis of GC-rich sequences: Aristaless-related homeobox example. *Methods in Molecular Biology*, **2013**, 1017, 105-20	1.4	5
171	Challenges of "sticky" co-immunoprecipitation: polyalanine tract protein-protein interactions. *Methods in Molecular Biology*, **2013**, 1017, 121-33	1.4	1
170	M North Sea M progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. *Brain*, **2013**, 136, 1146-54	11.2	49
169	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
168	FOXP1 mutations cause intellectual disability and a recognizable phenotype. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3166-75	2.5	63
167	CCDC22 deficiency in humans blunts activation of proinflammatory NF-B signaling. *Journal of Clinical Investigation*, **2013**, 123, 2244-56	15.9	64

166	Loss of Usp9x disrupts cortical architecture, hippocampal development and TGF□mediated axonogenesis. *PLoS ONE*, **2013**, 8, e68287	3.7	53
165	New mutations and sporadic intellectual disability. *Lancet, The*, **2012**, 380, 1630-1	40	1
164	Choreoathetosis, congenital hypothyroidism and neonatal respiratory distress syndrome with intact NKX2-1. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 3168-73	2.5	19
163	Broadening the phenotype associated with mutations in UPF3B: two further cases with renal dysplasia and variable developmental delay. *European Journal of Medical Genetics*, **2012**, 55, 476-9	2.6	26
162	A novel contiguous gene deletion of AVPR2 and ARHGAP4 genes in male dizygotic twins with nephrogenic diabetes insipidus and intellectual disability. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2511-8	2.5	15
161	Is there a Mendelian transmission ratio distortion of the c.429_452dup(24bp) polyalanine tract ARX mutation?. *European Journal of Human Genetics*, **2012**, 20, 1311-4	5.3	6
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