

Vera M Kalscheuer

List of Publications by Year in descending order

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158
papers

11,425
citations

32410

55
h-index

38517

99
g-index

164
all docs

164
docs citations

164
times ranked

18174
citing authors

#	ARTICLE	IF	CITATIONS
1	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. <i>Nature</i> , 2011, 478, 57-63.	13.7	805
2	Germline KRAS mutations cause Noonan syndrome. <i>Nature Genetics</i> , 2006, 38, 331-336.	9.4	670
3	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. <i>Nature Genetics</i> , 2010, 42, 1021-1026.	9.4	431
4	Mutations in the JARID1C Gene, Which Is Involved in Transcriptional Regulation and Chromatin Remodeling, Cause X-Linked Mental Retardation. <i>American Journal of Human Genetics</i> , 2005, 76, 227-236.	2.6	349
5	Transcription Factor SOX3 Is Involved in X-Linked Mental Retardation with Growth Hormone Deficiency. <i>American Journal of Human Genetics</i> , 2002, 71, 1450-1455.	2.6	265
6	Disruption of the Serine/Threonine Kinase 9 Gene Causes Severe X-Linked Infantile Spasms and Mental Retardation. <i>American Journal of Human Genetics</i> , 2003, 72, 1401-1411.	2.6	265
7	Mutations in the X-Linked Cyclin-Dependent Kinase-Like 5 (CDKL5/STK9) Gene Are Associated with Severe Neurodevelopmental Retardation. <i>American Journal of Human Genetics</i> , 2004, 75, 1149-1154.	2.6	264
8	ARX, a novel Prd-class-homeobox gene highly expressed in the telencephalon, is mutated in X-linked mental retardation. <i>Human Molecular Genetics</i> , 2002, 11, 981-991.	1.4	248
9	The insulin-like growth factor type-2 receptor gene is imprinted in the mouse but not in humans. <i>Nature Genetics</i> , 1993, 5, 74-78.	9.4	246
10	CDKL5 ensures excitatory synapse stability by reinforcing NGL-1-PSD95 interaction in the postsynaptic compartment and is impaired in patient iPSC-derived neurons. <i>Nature Cell Biology</i> , 2012, 14, 911-923.	4.6	231
11	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	2.6	230
12	Mutations in the Small GTPase Gene RAB39B Are Responsible for X-linked Mental Retardation Associated with Autism, Epilepsy, and Macrocephaly. <i>American Journal of Human Genetics</i> , 2010, 86, 185-195.	2.6	220
13	Mutations in RAB39B Cause X-Linked Intellectual Disability and Early-Onset Parkinson Disease with α -Synuclein Pathology. <i>American Journal of Human Genetics</i> , 2014, 95, 729-735.	2.6	207
14	WDR11, a WD Protein that Interacts with Transcription Factor EMX1, Is Mutated in Idiopathic Hypogonadotropic Hypogonadism and Kallmann Syndrome. <i>American Journal of Human Genetics</i> , 2010, 87, 465-479.	2.6	165
15	Brain white matter oedema due to CIC-2 chloride channel deficiency: an observational analytical study. <i>Lancet Neurology</i> , The, 2013, 12, 659-668.	4.9	152
16	Spectrum of mutations in PTPN11 and genotype-phenotype correlation in 96 patients with Noonan syndrome and five patients with cardio-facio-cutaneous syndrome. <i>European Journal of Human Genetics</i> , 2003, 11, 201-206.	1.4	148
17	The epsilon-sarcoglycan gene (SGCE), mutated in myoclonus-dystonia syndrome, is maternally imprinted. <i>European Journal of Human Genetics</i> , 2003, 11, 138-144.	1.4	148
18	Agenesis and dysgenesis of the corpus callosum: Clinical, genetic and neuroimaging findings in a series of 41 patients. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2501-2511.	0.7	148

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19	Truncation of the Down Syndrome Candidate Gene DYRK1A in Two Unrelated Patients with Microcephaly. <i>American Journal of Human Genetics</i> , 2008, 82, 1165-1170.	2.6	145
20	Mutations at the mouse ichthyosis locus are within the lamin B receptor gene: a single gene model for human Pelger-Huet anomaly. <i>Human Molecular Genetics</i> , 2003, 12, 61-69.	1.4	142
21	Mutations in the polyglutamine binding protein 1 gene cause X-linked mental retardation. <i>Nature Genetics</i> , 2003, 35, 313-315.	9.4	139
22	Genomic Organization and Expression of the Doublesex-Related Gene Cluster in Vertebrates and Detection of Putative Regulatory Regions for DMRT1. <i>Genomics</i> , 2001, 77, 8-17.	1.3	137
23	A balanced chromosomal translocation disrupting <i>ARHGEF9</i> is associated with epilepsy, anxiety, aggression, and mental retardation. <i>Human Mutation</i> , 2009, 30, 61-68.	1.1	131
24	Genetics of intellectual disability in consanguineous families. <i>Molecular Psychiatry</i> , 2019, 24, 1027-1039.	4.1	131
25	Defects in tRNA Anticodon Loop 2'-O-Methylation Are Implicated in Nonsyndromic X-Linked Intellectual Disability due to Mutations in <i>FTSJ1</i> . <i>Human Mutation</i> , 2015, 36, 1176-1187.	1.1	122
26	Mapping translocation breakpoints by next-generation sequencing. <i>Genome Research</i> , 2008, 18, 1143-1149.	2.4	118
27	Mutations in the <i>FTSJ1</i> Gene Coding for a Novel S-Adenosylmethionine ^ε -Binding Protein Cause Nonsyndromic X-Linked Mental Retardation. <i>American Journal of Human Genetics</i> , 2004, 75, 305-309.	2.6	117
28	Mutations in autism susceptibility candidate 2 (<i>AUTS2</i>) in patients with mental retardation. <i>Human Genetics</i> , 2007, 121, 501-509.	1.8	116
29	Mutation of plasma membrane Ca ²⁺ ATPase isoform 3 in a family with X-linked congenital cerebellar ataxia impairs Ca ²⁺ homeostasis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 14514-14519.	3.3	113
30	Integrative analysis revealed the molecular mechanism underlying <i>RBM10</i> -mediated splicing regulation. <i>EMBO Molecular Medicine</i> , 2013, 5, 1431-1442.	3.3	106
31	Mutation frequencies of X-linked mental retardation genes in families from the EuroMRX consortium. <i>Human Mutation</i> , 2007, 28, 207-208.	1.1	103
32	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. <i>American Journal of Human Genetics</i> , 2015, 97, 922-932.	2.6	101
33	Haploinsufficiency of novel <i>FOXC1B</i> variants in a patient with severe mental retardation, brain malformations and microcephaly. <i>Human Genetics</i> , 2005, 117, 536-544.	1.8	96
34	Detecting genomic indel variants with exact breakpoints in single- and paired-end sequencing data using SplazerS. <i>Bioinformatics</i> , 2012, 28, 619-627.	1.8	95
35	Monoallelic Expression of Human <i>PEG1/MEST</i> s Paralleled by Parent-Specific Methylation in Fetuses. <i>Genomics</i> , 1997, 42, 236-244.	1.3	91
36	Disruption of Netrin G1 by a balanced chromosome translocation in a girl with Rett syndrome. <i>European Journal of Human Genetics</i> , 2005, 13, 921-927.	1.4	90

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37	Involvement of the kinesin family members <i>KIF4A</i> and <i>KIF5C</i> in intellectual disability and synaptic function. <i>Journal of Medical Genetics</i> , 2014, 51, 487-494.	1.5	90
38	A Noncoding, Regulatory Mutation Implicates <i>HCFC1</i> in Nonsyndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2012, 91, 694-702.	2.6	89
39	Next-Generation Sequencing Identifies Mutations of <i>SMPX</i> , which Encodes the Small Muscle Protein, X-Linked, as a Cause of Progressive Hearing Impairment. <i>American Journal of Human Genetics</i> , 2011, 88, 628-634.	2.6	88
40	Mutations in <i>SLC33A1</i> Cause a Lethal Autosomal-Recessive Disorder with Congenital Cataracts, Hearing Loss, and Low Serum Copper and Ceruloplasmin. <i>American Journal of Human Genetics</i> , 2012, 90, 61-68.	2.6	85
41	Hi-C Identifies Complex Genomic Rearrangements and TAD-Shuffling in Developmental Diseases. <i>American Journal of Human Genetics</i> , 2020, 106, 872-884.	2.6	85
42	Mutations in the <i>ZNF41</i> Gene Are Associated with Cognitive Deficits: Identification of a New Candidate for X-Linked Mental Retardation. <i>American Journal of Human Genetics</i> , 2003, 73, 1341-1354.	2.6	83
43	<i>THOC2</i> Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. <i>American Journal of Human Genetics</i> , 2015, 97, 302-310.	2.6	82
44	Mutations in the Intellectual Disability Gene <i>Ube2a</i> Cause Neuronal Dysfunction and Impair Parkin-Dependent Mitophagy. <i>Molecular Cell</i> , 2013, 50, 831-843.	4.5	80
45	Multiple giant cell lesions in patients with Noonan syndrome and cardio-facio-cutaneous syndrome. <i>European Journal of Human Genetics</i> , 2009, 17, 420-425.	1.4	79
46	Disruption of the <i>TCF4</i> gene in a girl with mental retardation but without the classical Pitt-Hopkins syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2053-2059.	0.7	68
47	<i>ZC4H2</i> Mutations Are Associated with Arthrogryposis Multiplex Congenita and Intellectual Disability through Impairment of Central and Peripheral Synaptic Plasticity. <i>American Journal of Human Genetics</i> , 2013, 92, 681-695.	2.6	68
48	Nonsyndromic X-linked mental retardation: where are the missing mutations?. <i>Trends in Genetics</i> , 2003, 19, 316-320.	2.9	65
49	Impact of low copy repeats on the generation of balanced and unbalanced chromosomal aberrations in mental retardation. <i>Cytogenetic and Genome Research</i> , 2006, 115, 247-253.	0.6	65
50	X-exome sequencing in Finnish families with Intellectual Disability - four novel mutations and two novel syndromic phenotypes. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 49.	1.2	64
51	Maternal-Specific Methylation of the Human <i>IGF2R</i> Gene Is Not Accompanied by Allele-Specific Transcription. <i>Genomics</i> , 1996, 31, 158-166.	1.3	63
52	Chromosome deletions in 13q33-q34: Report of four patients and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 337-342.	0.7	63
53	Loss of function of <i>KIAA2022</i> causes mild to severe intellectual disability with an autism spectrum disorder and impairs neurite outgrowth. <i>Human Molecular Genetics</i> , 2013, 22, 3306-3314.	1.4	62
54	Breakpoint analysis of balanced chromosome rearrangements by next-generation paired-end sequencing. <i>European Journal of Human Genetics</i> , 2010, 18, 539-543.	1.4	61

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55	Evidence against a major role of PEG1/MEST in Silver-Russell syndrome. <i>European Journal of Human Genetics</i> , 1998, 6, 114-120.	1.4	60
56	Mild phenotypes in a series of patients with Opitz GBBB syndrome with MID1 mutations. <i>American Journal of Medical Genetics, Part A</i> , 2005, 132A, 1-7.	0.7	59
57	Translocations Disrupting PHF21A in the Potocki-Shaffer-Syndrome Region Are Associated with Intellectual Disability and Craniofacial Anomalies. <i>American Journal of Human Genetics</i> , 2012, 91, 56-72.	2.6	59
58	<i>EIF2S3</i> Mutations Associated with Severe X-Linked Intellectual Disability Syndrome MEHMO. <i>Human Mutation</i> , 2017, 38, 409-425.	1.1	57
59	In-frame deletion in MECP2 causes mild nonspecific mental retardation. <i>American Journal of Medical Genetics Part A</i> , 2002, 107, 81-83.	2.4	56
60	An excess of chromosome 1 breakpoints in male infertility. <i>European Journal of Human Genetics</i> , 2004, 12, 993-1000.	1.4	56
61	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. <i>American Journal of Human Genetics</i> , 2017, 101, 833-843.	2.6	56
62	O-GlcNAc transferase missense mutations linked to X-linked intellectual disability deregulate genes involved in cell fate determination and signaling. <i>Journal of Biological Chemistry</i> , 2018, 293, 10810-10824.	1.6	56
63	Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. <i>Human Genetics</i> , 2006, 118, 578-590.	1.8	55
64	Redefining the MED13L syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1308-1317.	1.4	53
65	Modeling Read Counts for CNV Detection in Exome Sequencing Data. <i>Statistical Applications in Genetics and Molecular Biology</i> , 2011, 10, .	0.2	52
66	The X-chromosome-linked intellectual disability protein PQBP1 is a component of neuronal RNA granules and regulates the appearance of stress granules. <i>Human Molecular Genetics</i> , 2011, 20, 4916-4931.	1.4	52
67	New insights into Brunner syndrome and potential for targeted therapy. <i>Clinical Genetics</i> , 2016, 89, 120-127.	1.0	52
68	<i>CDKL5</i> variants. <i>Neurology: Genetics</i> , 2017, 3, e200.	0.9	52
69	TRPV1 acts as a synaptic protein and regulates vesicle recycling. <i>Journal of Cell Science</i> , 2010, 123, 2045-2057.	1.2	51
70	Effect of inbreeding on intellectual disability revisited by trio sequencing. <i>Clinical Genetics</i> , 2019, 95, 151-159.	1.0	49
71	Mutation screening in 86 known X-linked mental retardation genes by droplet-based multiplex PCR and massive parallel sequencing. <i>The HUGO Journal</i> , 2009, 3, 41-49.	4.1	48
72	Characterisation of de novo MAPK10/JNK3 truncation mutations associated with cognitive disorders in two unrelated patients. <i>Human Genetics</i> , 2013, 132, 461-471.	1.8	48

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73	HCFC1 loss-of-function mutations disrupt neuronal and neural progenitor cells of the developing brain. <i>Human Molecular Genetics</i> , 2015, 24, 3335-3347.	1.4	47
74	Dysfunction of the Heteromeric KV7.3/KV7.5 Potassium Channel is Associated with Autism Spectrum Disorders. <i>Frontiers in Genetics</i> , 2013, 4, 54.	1.1	45
75	Absent <i>CNKSR</i> 2 causes seizures and intellectual, attention, and language deficits. <i>Annals of Neurology</i> , 2014, 76, 758-764.	2.8	43
76	Genomic Structure and Comparative Analysis of Nine <i>Fugu</i> Genes: Conservation of Synteny with Human Chromosome Xp22.2â€”p22.1. <i>Genome Research</i> , 1999, 9, 437-448.	2.4	41
77	Early Frameshift Mutation in <i>PIGA</i> Identified in a Large XLID Family Without Neonatal Lethality. <i>Human Mutation</i> , 2014, 35, 350-355.	1.1	39
78	Conflicting Reports of Imprinting Status of Human GRB10 in Developing Brain: How Reliable Are Somatic Cell Hybrids for Predicting Allelic Origin of Expression?. <i>American Journal of Human Genetics</i> , 2001, 68, 543-544.	2.6	38
79	Low incidence of UPD in spontaneous abortions beyond the 5th gestational week. <i>European Journal of Human Genetics</i> , 2001, 9, 910-916.	1.4	37
80	Evidence for a new contiguous gene syndrome, the chromosome 16p13.3 deletion syndrome alias severe Rubinsteinâ€”Taybi syndrome. <i>Human Genetics</i> , 2006, 120, 179-186.	1.8	37
81	Clinical and neurocognitive characterization of a family with a novel <i>MED12</i> gene frameshift mutation. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 3063-3071.	0.7	37
82	Variants in <i>CUL4B</i> are Associated with Cerebral Malformations. <i>Human Mutation</i> , 2015, 36, 106-117.	1.1	37
83	Truncation of the CNS-expressed JNK3 in a patient with a severe developmental epileptic encephalopathy. <i>Human Genetics</i> , 2006, 118, 559-567.	1.8	35
84	<i>ARHGEF9</i> disease. <i>Neurology: Genetics</i> , 2017, 3, e148.	0.9	35
85	Identification of a novel CDKL5 exon and pathogenic mutations in patients with severe mental retardation, early-onset seizures and Rett-like features. <i>Neurogenetics</i> , 2011, 12, 165-167.	0.7	34
86	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. <i>Brain</i> , 2017, 140, 2879-2894.	3.7	33
87	Haplotype Sharing Analysis Identifies a Retroviral dUTPase as Candidate Susceptibility Gene for Psoriasis. <i>Journal of Investigative Dermatology</i> , 2005, 124, 99-102.	0.3	32
88	A new standard nomenclature for proteins related to Apx and Shroom. <i>BMC Cell Biology</i> , 2006, 7, 18.	3.0	31
89	A Y328C missense mutation in spermine synthase causes a mild form of Snyderâ€”Robinson syndrome. <i>Human Molecular Genetics</i> , 2013, 22, 3789-3797.	1.4	31
90	Integrated Sequence Analysis Pipeline Provides One-Stop Solution for Identifying Disease-Causing Mutations. <i>Human Mutation</i> , 2014, 35, 1427-1435.	1.1	31

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91	Epilepsy and intellectual disability linked protein Shrm4 interaction with GABABRs shapes inhibitory neurotransmission. <i>Nature Communications</i> , 2017, 8, 14536.	5.8	31
92	No imprinting involved in the expression of DM-kinase m RNAs in mouse and human tissues. <i>Human Molecular Genetics</i> , 1993, 2, 1221-1227.	1.4	30
93	A 3-base pair deletion, c.9711_9713del, in DMD results in intellectual disability without muscular dystrophy. <i>European Journal of Human Genetics</i> , 2014, 22, 480-485.	1.4	30
94	Evaluation of the IRF-2 Gene as a Candidate for PSORS3. <i>Journal of Investigative Dermatology</i> , 2004, 122, 61-64.	0.3	29
95	Subtelomere FISH in 50 children with mental retardation and minor anomalies, identified by a checklist, detects 10 rearrangements including a de novo balanced translocation of chromosomes 17p13.3 and 20q13.33. <i>American Journal of Medical Genetics Part A</i> , 2004, 128A, 364-373.	2.4	29
96	Expanding the clinical phenotype of patients with a <i>ZDHHC9</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 789-795.	0.7	29
97	Deleterious de novo variants of X-linked <i>ZC4H2</i> in females cause a variable phenotype with neurogenic arthrogyrosis multiplex congenita. <i>Human Mutation</i> , 2019, 40, 2270-2285.	1.1	29
98	A Novel Mutation in <i>RPL10</i> (Ribosomal Protein L10) Causes X-Linked Intellectual Disability, Cerebellar Hypoplasia, and Spondylo-Epiphyseal Dysplasia. <i>Human Mutation</i> , 2015, 36, 1155-1158.	1.1	28
99	Increased <i>STAG2</i> dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. <i>Human Molecular Genetics</i> , 2015, 24, 7171-7181.	1.4	28
100	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. <i>Nature Genetics</i> , 2018, 50, 1442-1451.	9.4	28
101	TAF1, associated with intellectual disability in humans, is essential for embryogenesis and regulates neurodevelopmental processes in zebrafish. <i>Scientific Reports</i> , 2019, 9, 10730.	1.6	28
102	Methylation of L1Hs promoters is lower on the inactive X, has a tendency of being higher on autosomes in smaller genomes and shows inter-individual variability at some loci. <i>Human Molecular Genetics</i> , 2012, 21, 219-235.	1.4	27
103	Mutations in two large pedigrees highlight the role of ZNF711 in X-linked intellectual disability. <i>Gene</i> , 2017, 605, 92-98.	1.0	26
104	Pathogenic variants in E3 ubiquitin ligase RLIM/RNF12 lead to a syndromic X-linked intellectual disability and behavior disorder. <i>Molecular Psychiatry</i> , 2019, 24, 1748-1768.	4.1	26
105	A recurrent missense variant in <i>SLC9A7</i> causes nonsyndromic X-linked intellectual disability with alteration of Golgi acidification and aberrant glycosylation. <i>Human Molecular Genetics</i> , 2019, 28, 598-614.	1.4	25
106	Duplication of the MID1 first exon in a patient with Opitz G/BBB syndrome. <i>Human Genetics</i> , 2003, 112, 249-254.	1.8	24
107	The power of the Mediator complex-Expanding the genetic architecture and phenotypic spectrum of MED12-related disorders. <i>Clinical Genetics</i> , 2018, 94, 450-456.	1.0	24
108	Breakpoint Cloning and Haplotype Analysis Indicate a Single Origin of the Common Inv(10)(p11.2q21.2) Mutation among Northern Europeans. <i>American Journal of Human Genetics</i> , 2006, 78, 878-883.	2.6	23

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109	Novel Missense Mutation A789V in IQSEC2 Underlies X-Linked Intellectual Disability in the MRX78 Family. <i>Frontiers in Molecular Neuroscience</i> , 2015, 8, 85.	1.4	23
110	Regulation of the MID1 protein function is fine-tuned by a complex pattern of alternative splicing. <i>Human Genetics</i> , 2004, 114, 541-552.	1.8	22
111	Characterization of FBX25, encoding a novel brain-expressed F-box protein. <i>Biochimica Et Biophysica Acta - General Subjects</i> , 2006, 1760, 110-118.	1.1	22
112	Chromosome aberrations involving 10q22: report of three overlapping interstitial deletions and a balanced translocation disrupting C10orf11. <i>European Journal of Human Genetics</i> , 2010, 18, 291-295.	1.4	22
113	A region on human chromosome 4 (q35.1â†’qter) induces senescence in cell hybrids and is involved in cervical carcinogenesis. <i>Genes Chromosomes and Cancer</i> , 2005, 43, 260-272.	1.5	21
114	Hybridisation-based resequencing of 17 X-linked intellectual disability genes in 135 patients reveals novel mutations in ATRX, SLC6A8 and PQBP1. <i>European Journal of Human Genetics</i> , 2011, 19, 717-720.	1.4	21
115	Exonic microdeletions in the X-linked PQBP1 gene in mentally retarded patients: a pathogenic mutation and in-frame deletions of uncertain effect. <i>European Journal of Human Genetics</i> , 2006, 14, 418-425.	1.4	20
116	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. <i>Human Molecular Genetics</i> , 2018, 27, 589-600.	1.4	20
117	X chromosome dosage and presence of SRY shape sex-specific differences in DNA methylation at an autosomal region in human cells. <i>Biology of Sex Differences</i> , 2018, 9, 10.	1.8	20
118	TheMASProto-Oncogene Is Not Imprinted in Humans. <i>Genomics</i> , 1996, 35, 380-382.	1.3	19
119	Further delineation of the phenotype maps for partial trisomy 16q24 and Jacobsen syndrome by a subtle familial translocation t(11;16)(q24.2;q24.1). <i>American Journal of Medical Genetics, Part A</i> , 2005, 139A, 19-24.	0.7	19
120	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. <i>Human Molecular Genetics</i> , 2014, 23, 6163-6176.	1.4	19
121	Identification and characterization of G90, a novel mouse RNA that lacks an extensive open reading frame. <i>Gene</i> , 1999, 232, 35-42.	1.0	18
122	Common pathological mutations in <i>PQBP1</i> induce nonsense-mediated mRNA decay and enhance exclusion of the mutant exon. <i>Human Mutation</i> , 2010, 31, 90-98.	1.1	18
123	Genomic structure, chromosome mapping and expression analysis of the human AVIL gene, and its exclusion as a candidate for locus for inflammatory bowel disease at 12q13â€“14 (IBD2). <i>Gene</i> , 2002, 288, 179-185.	1.0	17
124	Identical by descent L1CAM mutation in two apparently unrelated families with intellectual disability without L1 syndrome. <i>European Journal of Medical Genetics</i> , 2015, 58, 364-368.	0.7	17
125	Absence of an Obvious Molecular Imprinting Mechanism in a Human Fetus with MonoallelicIGF2RExpression. <i>Biochemical and Biophysical Research Communications</i> , 1998, 245, 272-277.	1.0	16
126	Comprehensive analysis of human subtelomeres with combined binary ratio labelling fluorescence in situ hybridisation. <i>European Journal of Human Genetics</i> , 2003, 11, 643-651.	1.4	16

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127	Heterotaxy and cardiac defect in a girl with chromosome translocation t(X;1)(q26;p13.1) and involvement of ZIC3. <i>European Journal of Human Genetics</i> , 2006, 14, 1317-1320.	1.4	16
128	Novel <i>GDI1</i> mutation in a large family with nonsyndromic X-linked intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 3067-3070.	0.7	16
129	Characterization of a 16 Mb interstitial chromosome 7q21 deletion by tiling path array CGH. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 333-337.	0.7	15
130	Synaptic MAGUK Multimer Formation Is Mediated by PDZ Domains and Promoted by Ligand Binding. <i>Chemistry and Biology</i> , 2013, 20, 1044-1054.	6.2	15
131	Cyclin Y phosphorylation- and 14-3-3-binding-dependent activation of PCTAIRE-1/CDK16. <i>Biochemical Journal</i> , 2015, 469, 409-420.	1.7	15
132	cDNA cloning and characterization of the human THRAP2 gene which maps to chromosome 12q24, and its mouse ortholog Thrap2. <i>Gene</i> , 2004, 332, 119-127.	1.0	14
133	HUWE1 mutation explains phenotypic severity in a case of familial idiopathic intellectual disability. <i>European Journal of Medical Genetics</i> , 2013, 56, 379-382.	0.7	14
134	Molecular cloning and characterization of the Fugu rubripes MEST/COPG2 imprinting cluster and chromosomal localization in Fugu and Tetraodon nigroviridis. <i>Chromosome Research</i> , 2000, 8, 465-476.	1.0	13
135	Regulation and expression of the murine Pmp22 gene. <i>Mammalian Genome</i> , 1999, 10, 419-422.	1.0	12
136	Gene Dosage Analysis in Silver-Russell Syndrome: Use of Quantitative Competitive PCR and Dual-Color FISH to Estimate the Frequency of Duplications in 7p11.2-p13. <i>Genetic Testing and Molecular Biomarkers</i> , 2001, 5, 261-266.	1.7	12
137	Noonan-like/multiple giant cell lesion syndrome in two adult patients with SOS1 gene mutations. <i>Clinical Dysmorphology</i> , 2010, 19, 157-160.	0.1	12
138	<i>GPR126</i> : A novel candidate gene implicated in autosomal recessive intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 13-19.	0.7	12
139	Tentative clinical diagnosis of Lujan-Fryns syndrome? "A conglomeration of different genetic entities?". <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 94-102.	0.7	11
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141	Variant in the X-chromosome spliceosomal gene GPKOW causes male-lethal microcephaly with intrauterine growth restriction. <i>European Journal of Human Genetics</i> , 2017, 25, 1078-1082.	1.4	10
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143	A missense mutation in the CSTF2 gene that impairs the function of the RNA recognition motif and causes defects in 3' end processing is associated with intellectual disability in humans. <i>Nucleic Acids Research</i> , 2020, 48, 9804-9821.	6.5	10
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147	Small inherited terminal duplication of 7q with hydrocephalus, cleft palate, joint contractures, and severe hypotonia. Clinical Dysmorphology, 2003, 12, 123-7.	0.1	9
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