

# Vera M Kalscheuer

## List of Publications by Year in descending order

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158  
papers

11,425  
citations

28274

55  
h-index

33894

99  
g-index

164  
all docs

164  
docs citations

164  
times ranked

16662  
citing authors

#	ARTICLE	IF	CITATIONS
1	Comprehensive genotype-phenotype correlation in AP4 deficiency syndrome; Adding data from a large cohort of Iranian patients. <i>Clinical Genetics</i> , 2021, 99, 187-192.	2.0	2
2	Novel pathogenic EIF2S3 missense variants causing clinically variable MEHMO syndrome with impaired eIF2 $\beta$ translational function, and literature review. <i>Clinical Genetics</i> , 2020, 98, 507-514.	2.0	9
3	Whole genome sequencing identifies a duplicated region encompassing Xq13.2q13.3 in a large Iranian family with intellectual disability. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1418.	1.2	1
4	A missense mutation in the CSTF2 gene that impairs the function of the RNA recognition motif and causes defects in 5' end processing is associated with intellectual disability in humans. <i>Nucleic Acids Research</i> , 2020, 48, 9804-9821.	14.5	10
5	Hi-C Identifies Complex Genomic Rearrangements and TAD-Shuffling in Developmental Diseases. <i>American Journal of Human Genetics</i> , 2020, 106, 872-884.	6.2	85
6	GPR126: A novel candidate gene implicated in autosomal recessive intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 13-19.	1.2	12
7	TAF1, associated with intellectual disability in humans, is essential for embryogenesis and regulates neurodevelopmental processes in zebrafish. <i>Scientific Reports</i> , 2019, 9, 10730.	3.3	28
8	Deleterious de novo variants of X-linked ZC4H2 in females cause a variable phenotype with neurogenic arthrogyrosis multiplex congenita. <i>Human Mutation</i> , 2019, 40, 2270-2285.	2.5	29
9	Multigenic truncation of the semaphorin-plexin pathway by a germline chromothriptic rearrangement associated with Moebius syndrome. <i>Human Mutation</i> , 2019, 40, 1057-1062.	2.5	4
10	Mutation p.R356Q in the Collybistin Phosphoinositide Binding Site Is Associated With Mild Intellectual Disability. <i>Frontiers in Molecular Neuroscience</i> , 2019, 12, 60.	2.9	10
11	Identification of disease-causing variants in the EXOSC gene family underlying autosomal recessive intellectual disability in Iranian families. <i>Clinical Genetics</i> , 2019, 95, 718-725.	2.0	5
12	A recurrent missense variant in SLC9A7 causes nonsyndromic X-linked intellectual disability with alteration of Golgi acidification and aberrant glycosylation. <i>Human Molecular Genetics</i> , 2019, 28, 598-614.	2.9	25
13	Effect of inbreeding on intellectual disability revisited by trio sequencing. <i>Clinical Genetics</i> , 2019, 95, 151-159.	2.0	49
14	Genetics of intellectual disability in consanguineous families. <i>Molecular Psychiatry</i> , 2019, 24, 1027-1039.	7.9	131
15	Pathogenic variants in E3 ubiquitin ligase RLM/RNF12 lead to a syndromic X-linked intellectual disability and behavior disorder. <i>Molecular Psychiatry</i> , 2019, 24, 1748-1768.	7.9	26
16	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. <i>Human Molecular Genetics</i> , 2018, 27, 589-600.	2.9	20
17	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.	21.4	28
18	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.	3.4	56

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19	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.	2.0	24
20	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.	4.1	20
21	<i>EIF2S3</i> Mutations Associated with Severe X-Linked Intellectual Disability Syndrome MEHMO. <i>Human Mutation</i> , 2017, 38, 409-425.	2.5	57
22	Epilepsy and intellectual disability linked protein Shrm4 interaction with GABABRs shapes inhibitory neurotransmission. <i>Nature Communications</i> , 2017, 8, 14536.	12.8	31
23	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.	2.8	10
24	<i>ARHGEF9</i> disease. <i>Neurology: Genetics</i> , 2017, 3, e148.	1.9	35
25	Mutations in two large pedigrees highlight the role of ZNF711 in X-linked intellectual disability. <i>Gene</i> , 2017, 605, 92-98.	2.2	26
26	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.	6.2	56
27	<i>CDKL5</i> variants. <i>Neurology: Genetics</i> , 2017, 3, e200.	1.9	52
28	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. <i>Brain</i> , 2017, 140, 2879-2894.	7.6	33
29	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.	1.2	11
30	A balanced chromosomal translocation involving chromosomes 3 and 16 in a patient with Mayer-Rokitansky-Kuster-Hauser syndrome reveals new candidate genes at 3p22.3 and 16p13.3. <i>Molecular Cytogenetics</i> , 2016, 9, 57.	0.9	8
31	New insights into Brunner syndrome and potential for targeted therapy. <i>Clinical Genetics</i> , 2016, 89, 120-127.	2.0	52
32	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.	2.5	28
33	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.	2.5	122
34	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. <i>American Journal of Human Genetics</i> , 2015, 97, 922-932.	6.2	101
35	Cyclin Y phosphorylation- and 14-3-3-binding-dependent activation of PCTAIRE-1/CDK16. <i>Biochemical Journal</i> , 2015, 469, 409-420.	3.7	15
36	Increased <i>STAG2</i> dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. <i>Human Molecular Genetics</i> , 2015, 24, 7171-7181.	2.9	28

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37	Variants in <i>CUL4B</i> are Associated with Cerebral Malformations. <i>Human Mutation</i> , 2015, 36, 106-117.	2.5	37
38	Redefining the MED13L syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1308-1317.	2.8	53
39	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.	6.2	230
40	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. <i>American Journal of Human Genetics</i> , 2015, 97, 302-310.	6.2	82
41	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.	1.3	17
42	HCFC1 loss-of-function mutations disrupt neuronal and neural progenitor cells of the developing brain. <i>Human Molecular Genetics</i> , 2015, 24, 3335-3347.	2.9	47
43	Novel Missense Mutation A789V in IQSEC2 Underlies X-Linked Intellectual Disability in the MRX78 Family. <i>Frontiers in Molecular Neuroscience</i> , 2015, 8, 85.	2.9	23
44	Early Frameshift Mutation in <i>PIGA</i> Identified in a Large XLID Family Without Neonatal Lethality. <i>Human Mutation</i> , 2014, 35, 350-355.	2.5	39
45	Mutations in RAB39B Cause X-Linked Intellectual Disability and Early-Onset Parkinson Disease with $\alpha$ -Synuclein Pathology. <i>American Journal of Human Genetics</i> , 2014, 95, 729-735.	6.2	207
46	Integrated Sequence Analysis Pipeline Provides One-Stop Solution for Identifying Disease-Causing Mutations. <i>Human Mutation</i> , 2014, 35, 1427-1435.	2.5	31
47	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. <i>Human Molecular Genetics</i> , 2014, 23, 6163-6176.	2.9	19
48	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.	2.8	30
49	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.	3.2	90
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.	2.7	64
51	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.	1.2	29
52	Absent <i>CNKSRL2</i> causes seizures and intellectual, attention, and language deficits. <i>Annals of Neurology</i> , 2014, 76, 758-764.	5.3	43
53	Characterisation of de novo MAPK10/JNK3 truncation mutations associated with cognitive disorders in two unrelated patients. <i>Human Genetics</i> , 2013, 132, 461-471.	3.8	48
54	Integrative analysis revealed the molecular mechanism underlying <i>RBM10</i> -mediated splicing regulation. <i>EMBO Molecular Medicine</i> , 2013, 5, 1431-1442.	6.9	106

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55	A Y328C missense mutation in spermine synthase causes a mild form of Snyder-Robinson syndrome. <i>Human Molecular Genetics</i> , 2013, 22, 3789-3797.	2.9	31
56	ZC4H2 Mutations Are Associated with Arthrogyriposis Multiplex Congenita and Intellectual Disability through Impairment of Central and Peripheral Synaptic Plasticity. <i>American Journal of Human Genetics</i> , 2013, 92, 681-695.	6.2	68
57	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.	1.2	37
58	Synaptic MAGUK Multimer Formation Is Mediated by PDZ Domains and Promoted by Ligand Binding. <i>Chemistry and Biology</i> , 2013, 20, 1044-1054.	6.0	15
59	HUWE1 mutation explains phenotypic severity in a case of familial idiopathic intellectual disability. <i>European Journal of Medical Genetics</i> , 2013, 56, 379-382.	1.3	14
60	Mutations in the Intellectual Disability Gene Ube2a Cause Neuronal Dysfunction and Impair Parkin-Dependent Mitophagy. <i>Molecular Cell</i> , 2013, 50, 831-843.	9.7	80
61	Brain white matter oedema due to CIC-2 chloride channel deficiency: an observational analytical study. <i>Lancet Neurology</i> , 2013, 12, 659-668.	10.2	152
62	Loss of function of KIAA2022 causes mild to severe intellectual disability with an autism spectrum disorder and impairs neurite outgrowth. <i>Human Molecular Genetics</i> , 2013, 22, 3306-3314.	2.9	62
63	Dysfunction of the Heteromeric KV7.3/KV7.5 Potassium Channel is Associated with Autism Spectrum Disorders. <i>Frontiers in Genetics</i> , 2013, 4, 54.	2.3	45
64	Detecting genomic indel variants with exact breakpoints in single- and paired-end sequencing data using SplazerS. <i>Bioinformatics</i> , 2012, 28, 619-627.	4.1	95
65	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.	2.9	27
66	Mutation of plasma membrane Ca <sup>2+</sup> ATPase isoform 3 in a family with X-linked congenital cerebellar ataxia impairs Ca <sup>2+</sup> homeostasis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 14514-14519.	7.1	113
67	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.	6.2	59
68	A Noncoding, Regulatory Mutation Implicates HCFC1 in Nonsyndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2012, 91, 694-702.	6.2	89
69	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.	10.3	231
70	Mutations in SLC33A1 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.	6.2	85
71	Biparental inheritance of chromosomal abnormalities in male twins with non-syndromic mental retardation. <i>European Journal of Medical Genetics</i> , 2011, 54, e383-e388.	1.3	7
72	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. <i>Nature</i> , 2011, 478, 57-63.	27.8	805

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73	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.	2.8	21
74	Next-Generation Sequencing Identifies Mutations of SMPX, 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.	6.2	88
75	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.	1.4	34
76	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.	1.2	16
77	Modeling Read Counts for CNV Detection in Exome Sequencing Data. <i>Statistical Applications in Genetics and Molecular Biology</i> , 2011, 10, .	0.6	52
78	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.	2.9	52
79	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.	6.2	220
80	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.	6.2	165
81	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.	2.5	18
82	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.	2.8	22
83	Breakpoint analysis of balanced chromosome rearrangements by next-generation paired-end sequencing. <i>European Journal of Human Genetics</i> , 2010, 18, 539-543.	2.8	61
84	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. <i>Nature Genetics</i> , 2010, 42, 1021-1026.	21.4	431
85	<i>CDKL5</i> truncation due to a t(X;2)(p22.1;p25.3) in a girl with X-linked infantile spasm syndrome. <i>Clinical Genetics</i> , 2010, 77, 92-96.	2.0	8
86	TRPV1 acts as a synaptic protein and regulates vesicle recycling. <i>Journal of Cell Science</i> , 2010, 123, 2045-2057.	2.0	51
87	Noonan-like/multiple giant cell lesion syndrome in two adult patients with SOS1 gene mutations. <i>Clinical Dysmorphology</i> , 2010, 19, 157-160.	0.3	12
88	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.	2.5	131
89	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
90	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.	2.8	79

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91	Chromosome deletions in 13q33â€“34: Report of four patients and review of the literature. American Journal of Medical Genetics, Part A, 2008, 146A, 337-342.	1.2	63
92	Disruption of the <i>TCF4</i> gene in a girl with mental retardation but without the classical Pittâ€“Hopkins syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 2053-2059.	1.2	68
93	Agenesis and dysgenesis of the corpus callosum: Clinical, genetic and neuroimaging findings in a series of 41 patients. American Journal of Medical Genetics, Part A, 2008, 146A, 2501-2511.	1.2	148
94	Truncation of the Down Syndrome Candidate Gene DYRK1A in Two Unrelated Patients with Microcephaly. American Journal of Human Genetics, 2008, 82, 1165-1170.	6.2	145
95	Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome in a Girl with Chromosome Translocation t(2;3)(q33;q23). Ophthalmic Genetics, 2008, 29, 37-40.	1.2	6
96	Mapping translocation breakpoints by next-generation sequencing. Genome Research, 2008, 18, 1143-1149.	5.5	118
97	Characterization of a 16 Mb interstitial chromosome 7q21 deletion by tiling path array CGH. American Journal of Medical Genetics, Part A, 2007, 143A, 333-337.	1.2	15
98	Mutation frequencies of X-linked mental retardation genes in families from the EuroMRX consortium. Human Mutation, 2007, 28, 207-208.	2.5	103
99	Mutations in autism susceptibility candidate 2 (AUTS2) in patients with mental retardation. Human Genetics, 2007, 121, 501-509.	3.8	116
100	Breakpoint Cloning and Haplotype Analysis Indicate a Single Origin of the Common Inv(10)(p11.2q21.2) Mutation among Northern Europeans. American Journal of Human Genetics, 2006, 78, 878-883.	6.2	23
101	Characterization of FBX25, encoding a novel brain-expressed F-box protein. Biochimica Et Biophysica Acta - General Subjects, 2006, 1760, 110-118.	2.4	22
102	First report of a partial trisomy 3q12-q23 de novoâ€“FISH breakpoint determination and phenotypic characterization. European Journal of Medical Genetics, 2006, 49, 225-234.	1.3	4
103	Unexpected identification of two interstitial deletions in a patient with a pericentric inversion of a chromosome 4 and an abnormal phenotype. European Journal of Medical Genetics, 2006, 49, 215-223.	1.3	9
104	A new standard nomenclature for proteins related to Apx and Shroom. BMC Cell Biology, 2006, 7, 18.	3.0	31
105	Germline KRAS mutations cause Noonan syndrome. Nature Genetics, 2006, 38, 331-336.	21.4	670
106	Exonic microdeletions in the X-linked PQBP1 gene in mentally retarded patients: a pathogenic mutation and in-frame deletions of uncertain effect. European Journal of Human Genetics, 2006, 14, 418-425.	2.8	20
107	Heterotaxy and cardiac defect in a girl with chromosome translocation t(X;1)(q26;p13.1) and involvement of ZIC3. European Journal of Human Genetics, 2006, 14, 1317-1320.	2.8	16
108	Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. Human Genetics, 2006, 118, 578-590.	3.8	55

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109	Truncation of the CNS-expressed JNK3 in a patient with a severe developmental epileptic encephalopathy. <i>Human Genetics</i> , 2006, 118, 559-567.	3.8	35
110	Evidence for a new contiguous gene syndrome, the chromosome 16p13.3 deletion syndrome alias severe Rubinstein-Taybi syndrome. <i>Human Genetics</i> , 2006, 120, 179-186.	3.8	37
111	Molecular cytogenetic analysis of a de novo interstitial chromosome 10q22 deletion. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1108-1110.	1.2	9
112	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.	1.1	65
113	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.	2.8	90
114	Haplotype Sharing Analysis Identifies a Retroviral dUTPase as Candidate Susceptibility Gene for Psoriasis. <i>Journal of Investigative Dermatology</i> , 2005, 124, 99-102.	0.7	32
115	A region on human chromosome 4 (q35.1qter) induces senescence in cell hybrids and is involved in cervical carcinogenesis. <i>Genes Chromosomes and Cancer</i> , 2005, 43, 260-272.	2.8	21
116	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.	1.2	59
117	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.	1.2	19
118	Haploinsufficiency of novel FOXP1 variants in a patient with severe mental retardation, brain malformations and microcephaly. <i>Human Genetics</i> , 2005, 117, 536-544.	3.8	96
119	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.	6.2	349
120	An excess of chromosome 1 breakpoints in male infertility. <i>European Journal of Human Genetics</i> , 2004, 12, 993-1000.	2.8	56
121	Regulation of the MID1 protein function is fine-tuned by a complex pattern of alternative splicing. <i>Human Genetics</i> , 2004, 114, 541-552.	3.8	22
122	Evaluation of the IRF-2 Gene as a Candidate for PSORS3. <i>Journal of Investigative Dermatology</i> , 2004, 122, 61-64.	0.7	29
123	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
124	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.	2.2	14
125	Mutations in the FTS1 Gene Coding for a Novel S-Adenosylmethionine-Binding Protein Cause Nonsyndromic X-Linked Mental Retardation. <i>American Journal of Human Genetics</i> , 2004, 75, 305-309.	6.2	117
126	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.	6.2	264



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127	Duplication of the MID1 first exon in a patient with Opitz G/BBB syndrome. <i>Human Genetics</i> , 2003, 112, 249-254.	3.8	24
128	Nonsyndromic X-linked mental retardation: where are the missing mutations?. <i>Trends in Genetics</i> , 2003, 19, 316-320.	6.7	65
129	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.	2.8	148
130	The epsilon-sarcoglycan gene (SGCE), mutated in myoclonus-dystonia syndrome, is maternally imprinted. <i>European Journal of Human Genetics</i> , 2003, 11, 138-144.	2.8	148
131	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.	2.8	16
132	Mutations in the polyglutamine binding protein 1 gene cause X-linked mental retardation. <i>Nature Genetics</i> , 2003, 35, 313-315.	21.4	139
133	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.	6.2	265
134	Mutations in the ZNF41 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.	6.2	83
135	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.	2.9	142
136	Small inherited terminal duplication of 7q with hydrocephalus, cleft palate, joint contractures, and severe hypotonia. <i>Clinical Dysmorphology</i> , 2003, 12, 123-7.	0.3	9
137	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.	2.9	248
138	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.	6.2	265
139	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 12q13-q14 (IBD2). <i>Gene</i> , 2002, 288, 179-185.	2.2	17
140	In-frame deletion in <i>MECP2</i> causes mild nonspecific mental retardation. <i>American Journal of Medical Genetics Part A</i> , 2002, 107, 81-83.	2.4	56
141	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.	2.9	137
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