Vera M Kalscheuer

List of Publications by Year
in descending order

158	11,425	32410	38517
papers	citations	55 h-index	99 g-index

164	164	164	18174
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Comprehensive <scp>genotypeâ€phenotype</scp> correlation in <scp>AP</scp>â€4 deficiency syndrome; Adding data from a large cohort of Iranian patients. Clinical Genetics, 2021, 99, 187-192.	1.0	2
2	Novel pathogenic <scp>EIF2S3</scp> missense variants causing clinically variable <scp>MEHMO</scp> syndrome with impaired <scp>eIF2Î³</scp> translational function, and literature review. Clinical Genetics, 2020, 98, 507-514.	1.0	9
3	Whole genome sequencing identifies a duplicated region encompassing Xq13.2q13.3 in a large Iranian family with intellectual disability. Molecular Genetics & Genomic Medicine, 2020, 8, e1418.	0.6	1
4	A missense mutation in the CSTF2 gene that impairs the function of the RNA recognition motif and causes defects in 3â€² end processing is associated with intellectual disability in humans. Nucleic Acids Research, 2020, 48, 9804-9821.	6.5	10
5	Hi-C Identifies Complex Genomic Rearrangements and TAD-Shuffling in Developmental Diseases. American Journal of Human Genetics, 2020, 106, 872-884.	2.6	85
6	GPR126: A novel candidate gene implicated in autosomal recessive intellectual disability. American Journal of Medical Genetics, Part A, 2019, 179, 13-19.	0.7	12
7	TAF1, associated with intellectual disability in humans, is essential for embryogenesis and regulates neurodevelopmental processes in zebrafish. Scientific Reports, 2019, 9, 10730.	1.6	28
8	Deleterious de novo variants of Xâ€linked ZC4H2 in females cause a variable phenotype with neurogenic arthrogryposis multiplex congenita. Human Mutation, 2019, 40, 2270-2285.	1.1	29
9	Multigenic truncation of the semaphorinâ€“plexin pathway by a germline chromothriptic rearrangement associated with Moebius syndrome. Human Mutation, 2019, 40, 1057-1062.	1.1	4
10	Mutation p.R356Q in the Collybistin Phosphoinositide Binding Site Is Associated With Mild Intellectual Disability. Frontiers in Molecular Neuroscience, 2019, 12, 60.	1.4	10
11	Identification of diseaseâ€causing variants in the EXOSC gene family underlying autosomal recessive intellectual disability in Iranian families. Clinical Genetics, 2019, 95, 718-725.	1.0	5
12	A recurrent missense variant inSLC9A7causes nonsyndromic X-linked intellectual disability with alteration of Golgi acidification and aberrant glycosylation. Human Molecular Genetics, 2019, 28, 598-614.	1.4	25
13	Effect of inbreeding on intellectual disability revisited by trio sequencing. Clinical Genetics, 2019, 95, 151-159.	1.0	49
14	Genetics of intellectual disability in consanguineous families. Molecular Psychiatry, 2019, 24, 1027-1039.	4.1	131
15	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.	4.1	26
16	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. Human Molecular Genetics, 2018, 27, 589-600.	1.4	20
17	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. Nature Genetics, 2018, 50, 1442-1451.	9.4	28
18	O-GlcNAc transferase missense mutations linked to X-linked intellectual disability deregulate genes involved in cell fate determination and signaling. Journal of Biological Chemistry, 2018, 293, 10810-10824.	1.6	56

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19	The power of the Mediator complex-Expanding the genetic architecture and phenotypic spectrum of MED12 -related disorders. Clinical Genetics, 2018, 94, 450-456.	1.0	24
20	X chromosome dosage and presence of SRY shape sex-specific differences in DNA methylation at an autosomal region in human cells. Biology of Sex Differences, 2018, 9, 10.	1.8	20
21	EIF2S3Mutations Associated with Severe X-Linked Intellectual Disability Syndrome MEHMO. Human Mutation, 2017, 38, 409-425.	1.1	57
22	Epilepsy and intellectual disability linked protein Shrm4 interaction with GABABRs shapes inhibitory neurotransmission. Nature Communications, 2017, 8, 14536.	5.8	31
23	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.	1.4	10
24	ARHGEF9 disease. Neurology: Genetics, 2017, 3, e148.	0.9	35
25	Mutations in two large pedigrees highlight the role of ZNF711 in X-linked intellectual disability. Gene, 2017, 605, 92-98.	1.0	26
26	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. American Journal of Human Genetics, 2017, 101, 833-843.	2.6	56
27	CDKL5 variants. Neurology: Genetics, 2017, 3, e200.	0.9	52
28	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. Brain, 2017, 140, 2879-2894.	3.7	33
29	Tentative clinical diagnosis of Lujanâ€Fryns syndromeâ€"A conglomeration of different genetic entities?. American Journal of Medical Genetics, Part A, 2016, 170, 94-102.	0.7	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. Molecular Cytogenetics, 2016, 9, 57.	0.4	8
31	New insights into Brunner syndrome and potential for targeted therapy. Clinical Genetics, 2016, 89, 120-127.	1.0	52
32	A Novel Mutation inRPL10(Ribosomal Protein L10) Causes X-Linked Intellectual Disability, Cerebellar Hypoplasia, and Spondylo-Epiphyseal Dysplasia. Human Mutation, 2015, 36, 1155-1158.	1.1	28
33	Defects in tRNA Anticodon Loop 2â€²-O-Methylation Are Implicated in Nonsyndromic X-Linked Intellectual Disability due to Mutations inFTSJ1. Human Mutation, 2015, 36, 1176-1187.	1.1	122
34	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	2.6	101
35	Cyclin Y phosphorylation- and 14-3-3-binding-dependent activation of PCTAIRE-1/CDK16. Biochemical Journal, 2015, 469, 409-420.	1.7	15
36	IncreasedSTAG2dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. Human Molecular Genetics, 2015, 24, 7171-7181.	1.4	28

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37	Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117.	1.1	37
38	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	1.4	53
39	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	2.6	230
40	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. American Journal of Human Genetics, 2015, 97, 302-310.	2.6	82
41	Identical by descent L1CAM mutation in two apparently unrelated families with intellectual disability without L1 syndrome. European Journal of Medical Genetics, 2015, 58, 364-368.	0.7	17
42	HCFC1 loss-of-function mutations disrupt neuronal and neural progenitor cells of the developing brain. Human Molecular Genetics, 2015, 24, 3335-3347.	1.4	47
43	Novel Missense Mutation A789V in IQSEC2 Underlies X-Linked Intellectual Disability in the MRX78 Family. Frontiers in Molecular Neuroscience, 2015, 8, 85.	1.4	23
44	Early Frameshift Mutation inPIGAIdentified in a Large XLID Family Without Neonatal Lethality. Human Mutation, 2014, 35, 350-355.	1.1	39
45	Mutations in RAB39B Cause X-Linked Intellectual Disability and Early-Onset Parkinson Disease with Î±-Synuclein Pathology. American Journal of Human Genetics, 2014, 95, 729-735.	2.6	207
46	Integrated Sequence Analysis Pipeline Provides One-Stop Solution for Identifying Disease-Causing Mutations. Human Mutation, 2014, 35, 1427-1435.	1.1	31
47	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. Human Molecular Genetics, 2014, 23, 6163-6176.	1.4	19
48	A 3-base pair deletion, c.9711_9713del, in DMD results in intellectual disability without muscular dystrophy. European Journal of Human Genetics, 2014, 22, 480-485.	1.4	30
49	Involvement of the kinesin family membersKIF4AandKIF5Cin intellectual disability and synaptic function. Journal of Medical Genetics, 2014, 51, 487-494.	1.5	90
50	X-exome sequencing in Finnish families with Intellectual Disability - four novel mutations and two novel syndromic phenotypes. Orphanet Journal of Rare Diseases, 2014, 9, 49.	1.2	64
51	Expanding the clinical phenotype of patients with a ZDHHC9 mutation. American Journal of Medical Genetics, Part A, 2014, 164, 789-795.	0.7	29
52	Absent <scp>CNKSR</scp>2 causes seizures and intellectual, attention, and language deficits. Annals of Neurology, 2014, 76, 758-764.	2.8	43
53	Characterisation of de novo MAPK10/JNK3 truncation mutations associated with cognitive disorders in two unrelated patients. Human Genetics, 2013, 132, 461-471.	1.8	48
54	Integrative analysis revealed the molecular mechanism underlying <scp>RBM</scp>10â€mediated splicing regulation. EMBO Molecular Medicine, 2013, 5, 1431-1442.	3.3	106

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55	A Y328C missense mutation in spermine synthase causes a mild form of Snyderâ€“Robinson syndrome. Human Molecular Genetics, 2013, 22, 3789-3797.	1.4	31
56	ZC4H2 Mutations Are Associated with Arthrogryposis Multiplex Congenita and Intellectual Disability through Impairment of Central and Peripheral Synaptic Plasticity. American Journal of Human Genetics, 2013, 92, 681-695.	2.6	68
57	Clinical and neurocognitive characterization of a family with a novel MED12 gene frameshift mutation. American Journal of Medical Genetics, Part A, 2013, 161, 3063-3071.	0.7	37
58	Synaptic MAGUK Multimer Formation Is Mediated by PDZ Domains and Promoted by Ligand Binding. Chemistry and Biology, 2013, 20, 1044-1054.	6.2	15
59	HUWE1 mutation explains phenotypic severity in a case of familial idiopathic intellectual disability. European Journal of Medical Genetics, 2013, 56, 379-382.	0.7	14
60	Mutations in the Intellectual Disability Gene Ube2a Cause Neuronal Dysfunction and Impair Parkin-Dependent Mitophagy. Molecular Cell, 2013, 50, 831-843.	4.5	80
61	Brain white matter oedema due to ClC-2 chloride channel deficiency: an observational analytical study. Lancet Neurology, The, 2013, 12, 659-668.	4.9	152
62	Loss of function of KIAA2022 causes mild to severe intellectual disability with an autism spectrum disorder and impairs neurite outgrowth. Human Molecular Genetics, 2013, 22, 3306-3314.	1.4	62
63	Dysfunction of the Heteromeric KV7.3/KV7.5 Potassium Channel is Associated with Autism Spectrum Disorders. Frontiers in Genetics, 2013, 4, 54.	1.1	45
64	Detecting genomic indel variants with exact breakpoints in single- and paired-end sequencing data using SplazerS. Bioinformatics, 2012, 28, 619-627.	1.8	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. Human Molecular Genetics, 2012, 21, 219-235.	1.4	27
66	Mutation of plasma membrane Ca ²⁺ ATPase isoform 3 in a family with X-linked congenital cerebellar ataxia impairs Ca ²⁺ homeostasis. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 14514-14519.	3.3	113
67	Translocations Disrupting PHF21A in the Potocki-Shaffer-Syndrome Region Are Associated with Intellectual Disability and Craniofacial Anomalies. American Journal of Human Genetics, 2012, 91, 56-72.	2.6	59
68	A Noncoding, Regulatory Mutation Implicates HCFC1 in Nonsyndromic Intellectual Disability. American Journal of Human Genetics, 2012, 91, 694-702.	2.6	89
69	CDKL5 ensures excitatory synapse stability by reinforcing NGL-1â€“PSD95 interaction in the postsynaptic compartment and is impaired in patient iPSC-derived neurons. Nature Cell Biology, 2012, 14, 911-923.	4.6	231
70	Mutations in SLC33A1 Cause a Lethal Autosomal-Recessive Disorder with Congenital Cataracts, Hearing Loss, and Low Serum Copper and Ceruloplasmin. American Journal of Human Genetics, 2012, 90, 61-68.	2.6	85
71	Biparental inheritance of chromosomal abnormalities in male twins with non-syndromic mental retardation. European Journal of Medical Genetics, 2011, 54, e383-e388.	0.7	7
72	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. Nature, 2011, 478, 57-63.	13.7	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. European Journal of Human Genetics, 2011, 19, 717-720.	1.4	21
74	Next-Generation Sequencing Identifies Mutations of SMPX, which Encodes the Small Muscle Protein, X-Linked, as a Cause of Progressive Hearing Impairment. American Journal of Human Genetics, 2011, 88, 628-634.	2.6	88
75	Identification of a novel CDKL5 exon and pathogenic mutations in patients with severe mental retardation, early-onset seizures and Rett-like features. Neurogenetics, 2011, 12, 165-167.	0.7	34
76	Novel GDI1 mutation in a large family with nonsyndromic Xâ€linked intellectual disability. American Journal of Medical Genetics, Part A, 2011, 155, 3067-3070.	0.7	16
77	Modeling Read Counts for CNV Detection in Exome Sequencing Data. Statistical Applications in Genetics and Molecular Biology, 2011, 10, .	0.2	52
78	The X-chromosome-linked intellectual disability protein PQBP1 is a component of neuronal RNA granules and regulates the appearance of stress granules. Human Molecular Genetics, 2011, 20, 4916-4931.	1.4	52
79	Mutations in the Small GTPase Gene RAB39B Are Responsible for X-linked Mental Retardation Associated with Autism, Epilepsy, and Macrocephaly. American Journal of Human Genetics, 2010, 86, 185-195.	2.6	220
80	WDR11, a WD Protein that Interacts with Transcription Factor EMX1, Is Mutated in Idiopathic Hypogonadotropic Hypogonadism and Kallmann Syndrome. American Journal of Human Genetics, 2010, 87, 465-479.	2.6	165
81	Common pathological mutations inPQBP1induce nonsense-mediated mRNA decay and enhance exclusion of the mutant exon. Human Mutation, 2010, 31, 90-98.	1.1	18
82	Chromosome aberrations involving 10q22: report of three overlapping interstitial deletions and a balanced translocation disrupting C10orf11. European Journal of Human Genetics, 2010, 18, 291-295.	1.4	22
83	Breakpoint analysis of balanced chromosome rearrangements by next-generation paired-end sequencing. European Journal of Human Genetics, 2010, 18, 539-543.	1.4	61
84	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	9.4	431
85	CDKL5 truncation due to a t(X;2)(p22.1;p25.3) in a girl with Xâ€linked infantile spasm syndrome. Clinical Genetics, 2010, 77, 92-96.	1.0	8
86	TRPV1 acts as a synaptic protein and regulates vesicle recycling. Journal of Cell Science, 2010, 123, 2045-2057.	1.2	51
87	Noonan-like/multiple giant cell lesion syndrome in two adult patients with SOS1 gene mutations. Clinical Dysmorphology, 2010, 19, 157-160.	0.1	12
88	A balanced chromosomal translocation disruptingARHGEF9is associated with epilepsy, anxiety, aggression, and mental retardation. Human Mutation, 2009, 30, 61-68.	1.1	131
89	Mutation screening in 86 known X-linked mental retardation genes by droplet-based multiplex PCR and massive parallel sequencing. The HUGO Journal, 2009, 3, 41-49.	4.1	48
90	Multiple giant cell lesions in patients with Noonan syndrome and cardio-facio-cutaneous syndrome. European Journal of Human Genetics, 2009, 17, 420-425.	1.4	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.	0.7	63
92	Disruption of the TCF4 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.	0.7	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.	0.7	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.	2.6	145
95	Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome in a Girl with Chromosome Translocation t(2;3)(q33;q23). Ophthalmic Genetics, 2008, 29, 37-40.	0.5	6
96	Mapping translocation breakpoints by next-generation sequencing. Genome Research, 2008, 18, 1143-1149.	2.4	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.	0.7	15
98	Mutation frequencies of X-linked mental retardation genes in families from the EuroMRX consortium. Human Mutation, 2007, 28, 207-208.	1.1	103
99	Mutations in autism susceptibility candidate 2 (AUTS2) in patients with mental retardation. Human Genetics, 2007, 121, 501-509.	1.8	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.	2.6	23
101	Characterization of FBX25, encoding a novel brain-expressed F-box protein. Biochimica Et Biophysica Acta - General Subjects, 2006, 1760, 110-118.	1.1	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.	0.7	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.	0.7	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.	9.4	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.	1.4	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.	1.4	16
108	Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. Human Genetics, 2006, 118, 578-590.	1.8	55

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109	Truncation of the CNS-expressed JNK3 in a patient with a severe developmental epileptic encephalopathy. Human Genetics, 2006, 118, 559-567.	1.8	35
110	Evidence for a new contiguous gene syndrome, the chromosome 16p13.3 deletion syndrome alias severe Rubinsteinâ€“Taybi syndrome. Human Genetics, 2006, 120, 179-186.	1.8	37
111	Molecular cytogenetic analysis of a de novo interstitial chromosome 10q22 deletion. American Journal of Medical Genetics, Part A, 2006, 140A, 1108-1110.	0.7	9
112	Impact of low copy repeats on the generation of balanced and unbalanced chromosomal aberrations in mental retardation. Cytogenetic and Genome Research, 2006, 115, 247-253.	0.6	65
113	Disruption of Netrin G1 by a balanced chromosome translocation in a girl with Rett syndrome. European Journal of Human Genetics, 2005, 13, 921-927.	1.4	90
114	Haplotype Sharing Analysis Identifies a Retroviral dUTPase as Candidate Susceptibility Gene for Psoriasis. Journal of Investigative Dermatology, 2005, 124, 99-102.	0.3	32
115	A region on human chromosome 4 (q35.1â†'qter) induces senescence in cell hybrids and is involved in cervical carcinogenesis. Genes Chromosomes and Cancer, 2005, 43, 260-272.	1.5	21
116	Mild phenotypes in a series of patients with Opitz GBBB syndrome withMID1mutations. American Journal of Medical Genetics, Part A, 2005, 132A, 1-7.	0.7	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). American Journal of Medical Genetics, Part A, 2005, 139A, 19-24.	0.7	19
118	Haploinsufficiency of novel FOXG1B variants in a patient with severe mental retardation, brain malformations and microcephaly. Human Genetics, 2005, 117, 536-544.	1.8	96
119	Mutations in the JARID1C Gene, Which Is Involved in Transcriptional Regulation and Chromatin Remodeling, Cause X-Linked Mental Retardation. American Journal of Human Genetics, 2005, 76, 227-236.	2.6	349
120	An excess of chromosome 1 breakpoints in male infertility. European Journal of Human Genetics, 2004, 12, 993-1000.	1.4	56
121	Regulation of the MID1 protein function is fine-tuned by a complex pattern of alternative splicing. Human Genetics, 2004, 114, 541-552.	1.8	22
122	Evaluation of the IRF-2 Gene as a Candidate for PSORS3. Journal of Investigative Dermatology, 2004, 122, 61-64.	0.3	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. American Journal of Medical Genetics Part A, 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. Gene, 2004, 332, 119-127.	1.0	14
125	Mutations in the FTSJ1 Gene Coding for a Novel S-Adenosylmethionineâ€“Binding Protein Cause Nonsyndromic X-Linked Mental Retardation. American Journal of Human Genetics, 2004, 75, 305-309.	2.6	117
126	Mutations in the X-Linked Cyclin-Dependent Kinaseâ€“Like 5 (CDKL5/STK9) Gene Are Associated with Severe Neurodevelopmental Retardation. American Journal of Human Genetics, 2004, 75, 1149-1154.	2.6	264

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127	Duplication of the MID1 first exon in a patient with Opitz G/BBB syndrome. Human Genetics, 2003, 112, 249-254.	1.8	24
128	Nonsyndromic X-linked mental retardation: where are the missing mutations?. Trends in Genetics, 2003, 19, 316-320.	2.9	65
129	Spectrum of mutations in PTPN11 and genotypeâ€“phenotype correlation in 96 patients with Noonan syndrome and five patients with cardio-facio-cutaneous syndrome. European Journal of Human Genetics, 2003, 11, 201-206.	1.4	148
130	The epsilon-sarcoglycan gene (SGCE), mutated in myoclonus-dystonia syndrome, is maternally imprinted. European Journal of Human Genetics, 2003, 11, 138-144.	1.4	148
131	Comprehensive analysis of human subtelomeres with combined binary ratio labelling fluorescence in situ hybridisation. European Journal of Human Genetics, 2003, 11, 643-651.	1.4	16
132	Mutations in the polyglutamine binding protein 1 gene cause X-linked mental retardation. Nature Genetics, 2003, 35, 313-315.	9.4	139
133	Disruption of the Serine/Threonine Kinase 9 Gene Causes Severe X-Linked Infantile Spasms and Mental Retardation. American Journal of Human Genetics, 2003, 72, 1401-1411.	2.6	265
134	Mutations in the ZNF41 Gene Are Associated with Cognitive Deficits: Identification of a New Candidate for X-Linked Mental Retardation. American Journal of Human Genetics, 2003, 73, 1341-1354.	2.6	83
135	Mutations at the mouse ichthyosis locus are within the lamin B receptor gene: a single gene model for human Pelger-Huet anomaly. Human Molecular Genetics, 2003, 12, 61-69.	1.4	142
136	Small inherited terminal duplication of 7q with hydrocephalus, cleft palate, joint contractures, and severe hypotonia. Clinical Dysmorphology, 2003, 12, 123-7.	0.1	9
137	ARX, a novel Prd-class-homeobox gene highly expressed in the telencephalon, is mutated in X-linked mental retardation. Human Molecular Genetics, 2002, 11, 981-991.	1.4	248
138	Transcription Factor SOX3 Is Involved in X-Linked Mental Retardation with Growth Hormone Deficiency. American Journal of Human Genetics, 2002, 71, 1450-1455.	2.6	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 12q13â€“14 (IBD2). Gene, 2002, 288, 179-185.	1.0	17
140	In-frame deletion inMECP2 causes mild nonspecific mental retardation. American Journal of Medical Genetics Part A, 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. Genomics, 2001, 77, 8-17.	1.3	137
142	Conflicting Reports of Imprinting Status of Human GRB10 in Developing Brain: How Reliable Are Somatic Cell Hybrids for Predicting Allelic Origin of Expression?. American Journal of Human Genetics, 2001, 68, 543-544.	2.6	38
143	Preferential Inactivation of a dupX(q23 â†' q27â€“28) Chromosome in a Girl with Mental Retardation and Dysmorphy. Human Heredity, 2001, 52, 177-182.	0.4	7
144	A subtelomeric cryptic unbalanced translocation der (1)t(1;18)(q44;q23) in a severely retarded girl: similarities and differences to the deletion 1q42/43-ter syndrome. Gene Function & Disease, 2001, 2, 165-170.	0.3	1

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145	Low incidence of UPD in spontaneous abortions beyond the 5th gestational week. European Journal of Human Genetics, 2001, 9, 910-916.	1.4	37
146	Gene Dosage Analysis in Silver-Russell Syndrome: Use of Quantitative Competitive PCR and Dual-Color FISH to Estimate the Frequency of Duplications in 7p11.2â€“p13. Genetic Testing and Molecular Biomarkers, 2001, 5, 261-266.	1.7	12
147	Molecular cloning and characterization of the Fugu rubripes MEST/COPG2 imprinting cluster and chromosomal localization in Fugu and Tetraodon nigroviridis. Chromosome Research, 2000, 8, 465-476.	1.0	13
148	Regulation and expression of the murine Pmp22 gene. Mammalian Genome, 1999, 10, 419-422.	1.0	12
149	Identification and characterization of G90, a novel mouse RNA that lacks an extensive open reading frame. Gene, 1999, 232, 35-42.	1.0	18
150	Genomic Structure and Comparative Analysis of Nine Fugu Genes: Conservation of Synteny with Human Chromosome Xp22.2â€“p22.1. Genome Research, 1999, 9, 437-448.	2.4	41
151	Evidence against a major role of PEG1/MEST in Silverâ€“Russell syndrome. European Journal of Human Genetics, 1998, 6, 114-120.	1.4	60
152	Absence of an Obvious Molecular Imprinting Mechanism in a Human Fetus with MonoallelicIGF2RExpression. Biochemical and Biophysical Research Communications, 1998, 245, 272-277.	1.0	16
153	Monoallelic Expression of HumanPEG1/MESTIs Paralleled by Parent-Specific Methylation in Fetuses. Genomics, 1997, 42, 236-244.	1.3	91
154	Maternal-Specific Methylation of the HumanIGF2RGene Is Not Accompanied by Allele-Specific Transcription. Genomics, 1996, 31, 158-166.	1.3	63
155	TheMASProto-Oncogene Is Not Imprinted in Humans. Genomics, 1996, 35, 380-382.	1.3	19
156	The insulinâ€“like growth factor typeâ€“2 receptor gene is imprinted in the mouse but not in humans. Nature Genetics, 1993, 5, 74-78.	9.4	246
157	No imprinting involved in the expression of DM-kinase m RNAs in mouse and human tissues. Human Molecular Genetics, 1993, 2, 1221-1227.	1.4	30
158	DNase I sensitivity of Microtus agrestis active, inactive and reactivated X chromosomes in mouse-Microtus cell hybrids. Chromosoma, 1988, 96, 227-230.	1.0	10