Vera M Kalscheuer

List of Publications by Year
in descending order

158	11,425	28274 55	33894 99
papers	citations	h-index	g-index

164	164	164	16662
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. Nature, 2011, 478, 57-63.	27.8	805
2	Germline KRAS mutations cause Noonan syndrome. Nature Genetics, 2006, 38, 331-336.	21.4	670
3	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	21.4	431
4	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.	6.2	349
5	Transcription Factor SOX3 Is Involved in X-Linked Mental Retardation with Growth Hormone Deficiency. American Journal of Human Genetics, 2002, 71, 1450-1455.	6.2	265
6	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.	6.2	265
7	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.	6.2	264
8	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.	2.9	248
9	The insulinâ€“like growth factor typeâ€“2 receptor gene is imprinted in the mouse but not in humans. Nature Genetics, 1993, 5, 74-78.	21.4	246
10	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.	10.3	231
11	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.	6.2	230
12	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.	6.2	220
13	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.	6.2	207
14	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.	6.2	165
15	Brain white matter oedema due to ClC-2 chloride channel deficiency: an observational analytical study. Lancet Neurology, The, 2013, 12, 659-668.	10.2	152
16	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.	2.8	148
17	The epsilon-sarcoglycan gene (SGCE), mutated in myoclonus-dystonia syndrome, is maternally imprinted. European Journal of Human Genetics, 2003, 11, 138-144.	2.8	148
18	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

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19	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
20	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.	2.9	142
21	Mutations in the polyglutamine binding protein 1 gene cause X-linked mental retardation. Nature Genetics, 2003, 35, 313-315.	21.4	139
22	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.	2.9	137
23	A balanced chromosomal translocation disruptingARHGEF9is associated with epilepsy, anxiety, aggression, and mental retardation. Human Mutation, 2009, 30, 61-68.	2.5	131
24	Genetics of intellectual disability in consanguineous families. Molecular Psychiatry, 2019, 24, 1027-1039.	7.9	131
25	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.	2.5	122
26	Mapping translocation breakpoints by next-generation sequencing. Genome Research, 2008, 18, 1143-1149.	5.5	118
27	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.	6.2	117
28	Mutations in autism susceptibility candidate 2 (AUTS2) in patients with mental retardation. Human Genetics, 2007, 121, 501-509.	3.8	116
29	Mutation of plasma membrane Ca ²⁺ ATPase isoform 3 in a family with X-linked congenital cerebellar ataxia impairs Ca ²⁺ homeostasis. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 14514-14519.	7.1	113
30	Integrative analysis revealed the molecular mechanism underlying <scp>RBM</scp>10â€mediated splicing regulation. EMBO Molecular Medicine, 2013, 5, 1431-1442.	6.9	106
31	Mutation frequencies of X-linked mental retardation genes in families from the EuroMRX consortium. Human Mutation, 2007, 28, 207-208.	2.5	103
32	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	6.2	101
33	Haploinsufficiency of novel FOXG1B variants in a patient with severe mental retardation, brain malformations and microcephaly. Human Genetics, 2005, 117, 536-544.	3.8	96
34	Detecting genomic indel variants with exact breakpoints in single- and paired-end sequencing data using SplazerS. Bioinformatics, 2012, 28, 619-627.	4.1	95
35	Monoallelic Expression of HumanPEG1/MESTIs Paralleled by Parent-Specific Methylation in Fetuses. Genomics, 1997, 42, 236-244.	2.9	91
36	Disruption of Netrin G1 by a balanced chromosome translocation in a girl with Rett syndrome. European Journal of Human Genetics, 2005, 13, 921-927.	2.8	90

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37	Involvement of the kinesin family membersKIF4AandKIF5Cin intellectual disability and synaptic function. Journal of Medical Genetics, 2014, 51, 487-494.	3.2	90
38	A Noncoding, Regulatory Mutation Implicates HCFC1 in Nonsyndromic Intellectual Disability. American Journal of Human Genetics, 2012, 91, 694-702.	6.2	89
39	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.	6.2	88
40	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.	6.2	85
41	Hi-C Identifies Complex Genomic Rearrangements and TAD-Shuffling in Developmental Diseases. American Journal of Human Genetics, 2020, 106, 872-884.	6.2	85
42	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.	6.2	83
43	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. American Journal of Human Genetics, 2015, 97, 302-310.	6.2	82
44	Mutations in the Intellectual Disability Gene Ube2a Cause Neuronal Dysfunction and Impair Parkin-Dependent Mitophagy. Molecular Cell, 2013, 50, 831-843.	9.7	80
45	Multiple giant cell lesions in patients with Noonan syndrome and cardio-facio-cutaneous syndrome. European Journal of Human Genetics, 2009, 17, 420-425.	2.8	79
46	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.	1.2	68
47	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.	6.2	68
48	Nonsyndromic X-linked mental retardation: where are the missing mutations?. Trends in Genetics, 2003, 19, 316-320.	6.7	65
49	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.	1.1	65
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.	2.7	64
51	Maternal-Specific Methylation of the HumanIGF2RGene Is Not Accompanied by Allele-Specific Transcription. Genomics, 1996, 31, 158-166.	2.9	63
52	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
53	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.	2.9	62
54	Breakpoint analysis of balanced chromosome rearrangements by next-generation paired-end sequencing. European Journal of Human Genetics, 2010, 18, 539-543.	2.8	61

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55	Evidence against a major role of PEG1/MEST in Silverâ€“Russell syndrome. European Journal of Human Genetics, 1998, 6, 114-120.	2.8	60
56	Mild phenotypes in a series of patients with Opitz GBBB syndrome withMID1mutations. American Journal of Medical Genetics, Part A, 2005, 132A, 1-7.	1.2	59
57	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.	6.2	59
58	EIF2S3Mutations Associated with Severe X-Linked Intellectual Disability Syndrome MEHMO. Human Mutation, 2017, 38, 409-425.	2.5	57
59	Inâ€frame deletion in MECP2 causes mild nonspecific mental retardation. American Journal of Medical Genetics Part A, 2002, 107, 81-83.	2.4	56
60	An excess of chromosome 1 breakpoints in male infertility. European Journal of Human Genetics, 2004, 12, 993-1000.	2.8	56
61	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.	6.2	56
62	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.	3.4	56
63	Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. Human Genetics, 2006, 118, 578-590.	3.8	55
64	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	2.8	53
65	Modeling Read Counts for CNV Detection in Exome Sequencing Data. Statistical Applications in Genetics and Molecular Biology, 2011, 10, .	0.6	52
66	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.	2.9	52
67	New insights into Brunner syndrome and potential for targeted therapy. Clinical Genetics, 2016, 89, 120-127.	2.0	52
68	CDKL5 variants. Neurology: Genetics, 2017, 3, e200.	1.9	52
69	TRPV1 acts as a synaptic protein and regulates vesicle recycling. Journal of Cell Science, 2010, 123, 2045-2057.	2.0	51
70	Effect of inbreeding on intellectual disability revisited by trio sequencing. Clinical Genetics, 2019, 95, 151-159.	2.0	49
71	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
72	Characterisation of de novo MAPK10/JNK3 truncation mutations associated with cognitive disorders in two unrelated patients. Human Genetics, 2013, 132, 461-471.	3.8	48

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73	HCFC1 loss-of-function mutations disrupt neuronal and neural progenitor cells of the developing brain. Human Molecular Genetics, 2015, 24, 3335-3347.	2.9	47
74	Dysfunction of the Heteromeric KV7.3/KV7.5 Potassium Channel is Associated with Autism Spectrum Disorders. Frontiers in Genetics, 2013, 4, 54.	2.3	45
75	Absent <scp>CNKSR</scp>2 causes seizures and intellectual, attention, and language deficits. Annals of Neurology, 2014, 76, 758-764.	5.3	43
76	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.	5.5	41
77	Early Frameshift Mutation inPIGAIdentified in a Large XLID Family Without Neonatal Lethality. Human Mutation, 2014, 35, 350-355.	2.5	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?. American Journal of Human Genetics, 2001, 68, 543-544.	6.2	38
79	Low incidence of UPD in spontaneous abortions beyond the 5th gestational week. European Journal of Human Genetics, 2001, 9, 910-916.	2.8	37
80	Evidence for a new contiguous gene syndrome, the chromosome 16p13.3 deletion syndrome alias severe Rubinsteinâ€“Taybi syndrome. Human Genetics, 2006, 120, 179-186.	3.8	37
81	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.	1.2	37
82	Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117.	2.5	37
83	Truncation of the CNS-expressed JNK3 in a patient with a severe developmental epileptic encephalopathy. Human Genetics, 2006, 118, 559-567.	3.8	35
84	ARHGEF9 disease. Neurology: Genetics, 2017, 3, e148.	1.9	35
85	Identification of a novel CDKL5 exon and pathogenic mutations in patients with severe mental retardation, early-onset seizures and Rett-like features. Neurogenetics, 2011, 12, 165-167.	1.4	34
86	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. Brain, 2017, 140, 2879-2894.	7.6	33
87	Haplotype Sharing Analysis Identifies a Retroviral dUTPase as Candidate Susceptibility Gene for Psoriasis. Journal of Investigative Dermatology, 2005, 124, 99-102.	0.7	32
88	A new standard nomenclature for proteins related to Apx and Shroom. BMC Cell Biology, 2006, 7, 18.	3.0	31
89	A Y328C missense mutation in spermine synthase causes a mild form of Snyderâ€“Robinson syndrome. Human Molecular Genetics, 2013, 22, 3789-3797.	2.9	31
90	Integrated Sequence Analysis Pipeline Provides One-Stop Solution for Identifying Disease-Causing Mutations. Human Mutation, 2014, 35, 1427-1435.	2.5	31

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91	Epilepsy and intellectual disability linked protein Shrm4 interaction with GABABRs shapes inhibitory neurotransmission. Nature Communications, 2017, 8, 14536.	12.8	31
92	No imprinting involved in the expression of DM-kinase m RNAs in mouse and human tissues. Human Molecular Genetics, 1993, 2, 1221-1227.	2.9	30
93	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.	2.8	30
94	Evaluation of the IRF-2 Gene as a Candidate for PSORS3. Journal of Investigative Dermatology, 2004, 122, 61-64.	0.7	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. American Journal of Medical Genetics Part A, 2004, 128A, 364-373.	2.4	29
96	Expanding the clinical phenotype of patients with a ZDHHC9 mutation. American Journal of Medical Genetics, Part A, 2014, 164, 789-795.	1.2	29
97	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.	2.5	29
98	A Novel Mutation inRPL10(Ribosomal Protein L10) Causes X-Linked Intellectual Disability, Cerebellar Hypoplasia, and Spondylo-Epiphyseal Dysplasia. Human Mutation, 2015, 36, 1155-1158.	2.5	28
99	IncreasedSTAG2dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. Human Molecular Genetics, 2015, 24, 7171-7181.	2.9	28
100	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. Nature Genetics, 2018, 50, 1442-1451.	21.4	28
101	TAF1, associated with intellectual disability in humans, is essential for embryogenesis and regulates neurodevelopmental processes in zebrafish. Scientific Reports, 2019, 9, 10730.	3.3	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. Human Molecular Genetics, 2012, 21, 219-235.	2.9	27
103	Mutations in two large pedigrees highlight the role of ZNF711 in X-linked intellectual disability. Gene, 2017, 605, 92-98.	2.2	26
104	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.	7.9	26
105	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.	2.9	25
106	Duplication of the MID1 first exon in a patient with Opitz G/BBB syndrome. Human Genetics, 2003, 112, 249-254.	3.8	24
107	The power of the Mediator complex-Expanding the genetic architecture and phenotypic spectrum of MED12 -related disorders. Clinical Genetics, 2018, 94, 450-456.	2.0	24
108	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

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109	Novel Missense Mutation A789V in IQSEC2 Underlies X-Linked Intellectual Disability in the MRX78 Family. Frontiers in Molecular Neuroscience, 2015, 8, 85.	2.9	23
110	Regulation of the MID1 protein function is fine-tuned by a complex pattern of alternative splicing. Human Genetics, 2004, 114, 541-552.	3.8	22
111	Characterization of FBX25, encoding a novel brain-expressed F-box protein. Biochimica Et Biophysica Acta - General Subjects, 2006, 1760, 110-118.	2.4	22
112	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.	2.8	22
113	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.	2.8	21
114	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.	2.8	21
115	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
116	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. Human Molecular Genetics, 2018, 27, 589-600.	2.9	20
117	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.	4.1	20
118	TheMASProto-Oncogene Is Not Imprinted in Humans. Genomics, 1996, 35, 380-382.	2.9	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). American Journal of Medical Genetics, Part A, 2005, 139A, 19-24.	1.2	19
120	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. Human Molecular Genetics, 2014, 23, 6163-6176.	2.9	19
121	Identification and characterization of G90, a novel mouse RNA that lacks an extensive open reading frame. Gene, 1999, 232, 35-42.	2.2	18
122	Common pathological mutations inPQBP1induce nonsense-mediated mRNA decay and enhance exclusion of the mutant exon. Human Mutation, 2010, 31, 90-98.	2.5	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). Gene, 2002, 288, 179-185.	2.2	17
124	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.	1.3	17
125	Absence of an Obvious Molecular Imprinting Mechanism in a Human Fetus with MonoallelicIGF2RExpression. Biochemical and Biophysical Research Communications, 1998, 245, 272-277.	2.1	16
126	Comprehensive analysis of human subtelomeres with combined binary ratio labelling fluorescence in situ hybridisation. European Journal of Human Genetics, 2003, 11, 643-651.	2.8	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. European Journal of Human Genetics, 2006, 14, 1317-1320.	2.8	16
128	Novel GDI1 mutation in a large family with nonsyndromic Xâ€linked intellectual disability. American Journal of Medical Genetics, Part A, 2011, 155, 3067-3070.	1.2	16
129	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
130	Synaptic MAGUK Multimer Formation Is Mediated by PDZ Domains and Promoted by Ligand Binding. Chemistry and Biology, 2013, 20, 1044-1054.	6.0	15
131	Cyclin Y phosphorylation- and 14-3-3-binding-dependent activation of PCTAIRE-1/CDK16. Biochemical Journal, 2015, 469, 409-420.	3.7	15
132	cDNA cloning and characterization of the human THRAP2 gene which maps to chromosome 12q24, and its mouse ortholog Thrap2. Gene, 2004, 332, 119-127.	2.2	14
133	HUWE1 mutation explains phenotypic severity in a case of familial idiopathic intellectual disability. European Journal of Medical Genetics, 2013, 56, 379-382.	1.3	14
134	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.	2.2	13
135	Regulation and expression of the murine Pmp22 gene. Mammalian Genome, 1999, 10, 419-422.	2.2	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 7p11.2â€“p13. Genetic Testing and Molecular Biomarkers, 2001, 5, 261-266.	1.7	12
137	Noonan-like/multiple giant cell lesion syndrome in two adult patients with SOS1 gene mutations. Clinical Dysmorphology, 2010, 19, 157-160.	0.3	12
138	GPR126: A novel candidate gene implicated in autosomal recessive intellectual disability. American Journal of Medical Genetics, Part A, 2019, 179, 13-19.	1.2	12
139	Tentative clinical diagnosis of Lujanâ€Fryns syndromeâ€”A conglomeration of different genetic entities?. American Journal of Medical Genetics, Part A, 2016, 170, 94-102.	1.2	11
140	DNase I sensitivity of Microtus agrestis active, inactive and reactivated X chromosomes in mouse-Microtus cell hybrids. Chromosoma, 1988, 96, 227-230.	2.2	10
141	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.	2.8	10
142	Mutation p.R356Q in the Collybistin Phosphoinositide Binding Site Is Associated With Mild Intellectual Disability. Frontiers in Molecular Neuroscience, 2019, 12, 60.	2.9	10
143	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.	14.5	10
144	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

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145	Molecular cytogenetic analysis of a de novo interstitial chromosome 10q22 deletion. American Journal of Medical Genetics, Part A, 2006, 140A, 1108-1110.	1.2	9
146	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.	2.0	9
147	Small inherited terminal duplication of 7q with hydrocephalus, cleft palate, joint contractures, and severe hypotonia. Clinical Dysmorphology, 2003, 12, 123-7.	0.3	9
148	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.	2.0	8
149	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.	0.9	8
150	Preferential Inactivation of a dupX(q23 â†' q27â€"28) Chromosome in a Girl with Mental Retardation and Dysmorphy. Human Heredity, 2001, 52, 177-182.	0.8	7
151	Biparental inheritance of chromosomal abnormalities in male twins with non-syndromic mental retardation. European Journal of Medical Genetics, 2011, 54, e383-e388.	1.3	7
152	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
153	Identification of diseaseâ€causing variants in the EXOSC gene family underlying autosomal recessive intellectual disability in Iranian families. Clinical Genetics, 2019, 95, 718-725.	2.0	5
154	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
155	Multigenic truncation of the semaphorinâ€"plexin pathway by a germline chromothriptic rearrangement associated with Moebius syndrome. Human Mutation, 2019, 40, 1057-1062.	2.5	4
156	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.	2.0	2
157	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
158	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.	1.2	1