Virginia E Kimonis

List of Publications by Year
in descending order

159	10,177	50276	37204
papers	citations	46 h-index	96 g-index

164	164	164	13051
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mosaic de novo SNRPN gene variant associated with Prader-Willi syndrome. Journal of Medical Genetics, 2022, 59, 719-722.	3.2	6
2	Development of a standard of care for patients with valosin-containing protein associated multisystem proteinopathy. Orphanet Journal of Rare Diseases, 2022, 17, 23.	2.7	19
3	VCP/p97 inhibitor CB-5083 modulates muscle pathology in a mouse model of VCP inclusion body myopathy. Journal of Translational Medicine, 2022, 20, 21.	4.4	6
4	Safety and effectiveness of resistance training in patients with late onset Pompe disease - a pilot study. Neuromuscular Disorders, 2022, 32, 284-294.	0.6	4
5	Severe cardiomyopathy associated with the VCP p.R155C and c.177_187del MYBPC3 gene variants. European Journal of Medical Genetics, 2022, 65, 104480.	1.3	3
6	Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918.	2.5	8
7	Molecular Classes and Growth Hormone Treatment Effects on Behavior and Emotion in Patients with Praderâ€“Willi Syndrome. Journal of Clinical Medicine, 2022, 11, 2572.	2.4	1
8	Pathogenic variants of Valosinâ€containing protein induce lysosomal damage and transcriptional activation of autophagy regulators in neuronal cells. Neuropathology and Applied Neurobiology, 2022, 48, e12818.	3.2	5
9	Multisystem Proteinopathy Due to VCP Mutations: A Review of Clinical Heterogeneity and Genetic Diagnosis. Genes, 2022, 13, 963.	2.4	28
10	A clinicopathologic study of malignancy in VCP-associated multisystem proteinopathy. Orphanet Journal of Rare Diseases, 2022, 17, .	2.7	2
11	Stroke and Chronic Kidney Disease in Fabry Disease. Journal of Stroke and Cerebrovascular Diseases, 2021, 30, 105423.	1.6	4
12	NUBPL mitochondrial disease: new patients and review of the genetic and clinical spectrum. Journal of Medical Genetics, 2021, 58, 314-325.	3.2	9
13	Multisystem proteinopathy: Where myopathy and motor neuron disease converge. Muscle and Nerve, 2021, 63, 442-454.	2.2	33
14	Variable clinical features of patients with Fabry disease and outcome of enzyme replacement therapy. Molecular Genetics and Metabolism Reports, 2021, 26, 100700.	1.1	8
15	Influence of molecular classes and growth hormone treatment on growth and dysmorphology in <scp>Praderâ€Willi</scp> syndrome: A multicenter study. Clinical Genetics, 2021, 100, 29-39.	2.0	8
16	A p97/Valosin-Containing Protein Inhibitor Drug CB-5083 Has a Potent but Reversible Off-Target Effect on Phosphodiesterase-6. Journal of Pharmacology and Experimental Therapeutics, 2021, 378, 31-41.	2.5	17
17	Ataxia and Parkinsonism in a Woman With a VCP Variant and Long-Normal Repeats in the SCA2 Allele. Neurology: Genetics, 2021, 7, e595.	1.9	4
18	Characteristics of VCP mutation-associated cardiomyopathy. Neuromuscular Disorders, 2021, 31, 701-705.	0.6	8

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19	Severe manifestations and treatment of COVID-19 in a transplanted patient with Fabry disease. Molecular Genetics and Metabolism Reports, 2021, 29, 100802.	1.1	2
20	A unique case of progressive hemifacial microsomia or Parry-Romberg syndrome associated with limb and brain anomalies with normal neurological findings: A review of the literature. European Journal of Medical Genetics, 2021, 64, 104234.	1.3	1
21	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. Pediatric Neurology, 2021, 123, 30-37.	2.1	21
22	Prevalence of cerebral small vessel disease in a Fabry disease cohort. Molecular Genetics and Metabolism Reports, 2021, 29, 100815.	1.1	5
23	Ceramide contributes to pathogenesis and may be targeted for therapy in VCP inclusion body myopathy. Human Molecular Genetics, 2021, 29, 3945-3953.	2.9	10
24	Regional Strain Pattern and Correlation with Cardiac Magnetic Resonance Imaging in Fabry Disease.. Journal of Cardiovascular Echography, 2021, 31, 131-136.	0.4	2
25	Homozygous B4GALNT1 mutation and biochemical glutaric acidemia type II: A case report. Clinical Neurology and Neurosurgery, 2020, 189, 105553.	1.4	3
26	Molecular subtype and growth hormone effects on dysmorphology in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 169-175.	1.2	7
27	Genetic Subtype-Phenotype Analysis of Growth Hormone Treatment on Psychiatric Behavior in Prader-Willi Syndrome. Genes, 2020, 11, 1250.	2.4	9
28	Diffuse large B-cell non-Hodgkin's lymphoma in Gaucher disease. Molecular Genetics and Metabolism Reports, 2020, 25, 100663.	1.1	4
29	Expression level of R155H mRNA in the knock-in mouse model. Biochemical and Biophysical Research Communications, 2020, 523, 985-986.	2.1	0
30	Hereditary Leiomyomatosis and Renal Cell Cancer (HLRCC): Report of a Family Pedigree. American Journal of the Medical Sciences, 2020, 360, 724-727.	1.1	0
31	Newborn screening for Praderâ€“Willi syndrome is feasible: Early diagnosis for better outcomes. American Journal of Medical Genetics, Part A, 2019, 179, 29-36.	1.2	15
32	Meta-analysis of genotype-phenotype analysis of OPA1 mutations in autosomal dominant optic atrophy. Mitochondrion, 2019, 46, 262-269.	3.4	29
33	Molecular genetic classification in Prader-Willi syndrome: a multisite cohort study. Journal of Medical Genetics, 2019, 56, 149-153.	3.2	112
34	Impact of genetic subtypes of Praderâ€“Willi syndrome with growth hormone therapy on intelligence and body mass index. American Journal of Medical Genetics, Part A, 2019, 179, 1826-1835.	1.2	23
35	GAA variants and phenotypes among 1,079 patients with Pompe disease: Data from the Pompe Registry. Human Mutation, 2019, 40, 2146-2164.	2.5	51
36	SMCHD1 mutation spectrum for facioscapulohumeral muscular dystrophy type 2 (FSHD2) and Bosma arhinia microphthalmia syndrome (BAMS) reveals disease-specific localisation of variants in the ATPase domain. Journal of Medical Genetics, 2019, 56, 693-700.	3.2	27

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37	Early Diagnosis in Praderâ€“Willi Syndrome Reduces Obesity and Associated Co-Morbidities. Genes, 2019, 10, 898.	2.4	25
38	A placebo-controlled trial of folic acid and betaine in identical twins with Angelman syndrome. Orphanet Journal of Rare Diseases, 2019, 14, 232.	2.7	2
39	A randomized pilot efficacy and safety trial of diazoxide choline controlled-release in patients with Prader-Willi syndrome. PLoS ONE, 2019, 14, e0221615.	2.5	30
40	ALG11â€CDG syndrome: Expanding the phenotype. American Journal of Medical Genetics, Part A, 2019, 179, 498-502.	1.2	8
41	Birth seasonality studies in a large Praderâ€“Willi syndrome cohort. American Journal of Medical Genetics, Part A, 2019, 179, 1531-1534.	1.2	2
42	Cover Image, Volume 179A, Number 3, March 2019. American Journal of Medical Genetics, Part A, 2019, 179, i.	1.2	0
43	Splice-Break: exploiting an RNA-seq splice junction algorithm to discover mitochondrial DNA deletion breakpoints and analyses of psychiatric disorders. Nucleic Acids Research, 2019, 47, e59-e59.	14.5	22
44	Homozygosity for the A431E mutation in PSEN1 presenting with a relatively aggressive phenotype. Neuroscience Letters, 2019, 699, 195-198.	2.1	8
45	Antisense oligonucleotide treatment targeting glycogen synthase (GYS1) in a mouse model of Pompe disease. Molecular Genetics and Metabolism, 2019, 126, S85.	1.1	5
46	New family with HSPB 8-associated autosomal dominant rimmed vacuolar myopathy. Neurology: Genetics, 2019, 5, e349.	1.9	24
47	Front Cover, Volume 40, Issue 11. Human Mutation, 2019, 40, i.	2.5	0
48	Contributing factors of mortality in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 196-205.	1.2	50
49	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
50	Variable clinical features and genotype-phenotype correlations in 18 patients with late-onset Pompe disease. Annals of Translational Medicine, 2019, 7, 276-276.	1.7	13
51	Comparison of perinatal factors in deletion versus uniparental disomy in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1161-1165.	1.2	17
52	Novel valosin-containing protein mutations associated with multisystem proteinopathy. Neuromuscular Disorders, 2018, 28, 491-501.	0.6	20
53	Praderâ€“Willi syndrome and earlyâ€onset morbid obesity NIH rare disease consortium: A review of natural history study. American Journal of Medical Genetics, Part A, 2018, 176, 368-375.	1.2	34
54	Genotypeâ€phenotype study in patients with valosinâ€containing protein mutations associated with multisystem proteinopathy. Clinical Genetics, 2018, 93, 119-125.	2.0	100

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55	Pathogenic mutations in NUBPL affect complex I activity and cold tolerance in the yeast model Yarrowia lipolytica. Human Molecular Genetics, 2018, 27, 3697-3709.	2.9	8
56	Multicentre study of maternal and neonatal outcomes in individuals with Prader-Willi syndrome. Journal of Medical Genetics, 2018, 55, 594-598.	3.2	25
57	A cross-sectional analysis of clinical evaluation in 35 individuals with mutations of the valosin-containing protein gene. Neuromuscular Disorders, 2018, 28, 778-786.	0.6	7
58	Phenotypic diversity of patients diagnosed with VACTERL association. American Journal of Medical Genetics, Part A, 2018, 176, 1830-1837.	1.2	7
59	Prospective phenotyping of NGLY1-CDDG, the first congenital disorder of deglycosylation. Genetics in Medicine, 2017, 19, 160-168.	2.4	124
60	Two cases of Leggâ€“Perthes and intellectual disability in Trichoâ€“Rhinoâ€“Phalangeal syndrome type 1 associated with novel TRPS1 mutations. American Journal of Medical Genetics, Part A, 2017, 173, 1663-1667.	1.2	4
61	Oxytocin treatment in children with Praderâ€“Willi syndrome: A doubleâ€blind, placeboâ€controlled, crossover study. American Journal of Medical Genetics, Part A, 2017, 173, 1243-1250.	1.2	90
62	Dysfunctional oleoylethanolamide signaling in a mouse model of Prader-Willi syndrome. Pharmacological Research, 2017, 117, 75-81.	7.1	16
63	A novel mutation of orthodenticle homeobox 2 contributing to a case of otocephaly initially diagnosed by prenatal ultrasound in the first trimester. Clinical Dysmorphology, 2017, 26, 98-100.	0.3	3
64	Activation of the NLRP3 Inflammasome Is Associated with Valosin-Containing Protein Myopathy. Inflammation, 2017, 40, 21-41.	3.8	32
65	Myogenic differentiation of VCP disease-induced pluripotent stem cells: A novel platform for drug discovery. PLoS ONE, 2017, 12, e0176919.	2.5	10
66	Response to Growth Hormone Treatment in a Patient with Insulin-Like Growth Factor 1 Receptor Deletion. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2017, 9, 380-386.	0.9	6
67	Gain-of-Function Mutations inRARBCause Intellectual Disability with Progressive Motor Impairment. Human Mutation, 2016, 37, 786-793.	2.5	34
68	Prader-Willi Syndrome due to an Unbalanced de novo Translocation t(15;19)(q12;p13.3). Cytogenetic and Genome Research, 2016, 150, 29-34.	1.1	3
69	The Myoblast C2C12 Transfected with Mutant Valosin-Containing Protein Exhibits Delayed Stress Granule Resolution on Oxidative Stress. American Journal of Pathology, 2016, 186, 1623-1634.	3.8	21
70	215th ENMC International Workshop VCP-related multi-system proteinopathy (IBMPFD) 13â€“15 November 2015, Heemskerk, The Netherlands. Neuromuscular Disorders, 2016, 26, 535-547.	0.6	38
71	Growth Charts for Prader-Willi Syndrome During Growth Hormone Treatment. Clinical Pediatrics, 2016, 55, 957-974.	0.8	37
72	DVL3 Alleles Resulting in a â^'1 Frameshift of the Last Exon Mediate Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2016, 98, 553-561.	6.2	88

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73	Rapamycin and Chloroquine: The In Vitro and In Vivo Effects of Autophagy-Modifying Drugs Show Promising Results in Valosin Containing Protein Multisystem Proteinopathy. PLoS ONE, 2015, 10, e0122888.	2.5	70
74	Administration of CoQ10 analogue ameliorates dysfunction of the mitochondrial respiratory chain in a mouse model of Angelman syndrome. Neurobiology of Disease, 2015, 76, 77-86.	4.4	25
75	A case report comparing clinical, imaging and neuropsychological assessment findings in twins discordant for the VCP p.R155C mutation. Neuromuscular Disorders, 2015, 25, 177-183.	0.6	6
76	In vitro studies in VCP-associated multisystem proteinopathy suggest altered mitochondrial bioenergetics. Mitochondrion, 2015, 22, 1-8.	3.4	22
77	Psychological Impact of Predictive Genetic Testing in VCP Inclusion Body Myopathy, Paget Disease of Bone and Frontotemporal Dementia. Journal of Genetic Counseling, 2015, 24, 842-850.	1.6	14
78	Clinical characterization of int22h1/int22h2-mediated Xq28 duplication/deletion: new cases and literature review. BMC Medical Genetics, 2015, 16, 12.	2.1	37
79	Targeted Excision of VCP R155H Mutation by Cre-LoxPTechnology as a Promising Therapeutic Strategy for Valosin-Containing Protein Disease. Human Gene Therapy Methods, 2015, 26, 13-24.	2.1	11
80	Growth Charts for Non-Growth Hormone Treated Prader-Willi Syndrome. Pediatrics, 2015, 135, e126-e135.	2.1	56
81	A Fine Balance of Dietary Lipids Improves Pathology of a Murine Model of VCP-Associated Multisystem Proteinopathy. PLoS ONE, 2015, 10, e0131995.	2.5	6
82	Effect of genetic subtypes and growth hormone treatment on bone mineral density in Prader-Willi syndrome. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 511-8.	0.9	18
83	Mild Phenotype in a Male with Pyruvate Dehydrogenase Complex Deficiency Associated with Novel Hemizygous In-Frame Duplication of the E1Î± Subunit Gene (PDHA1). Neuropediatrics, 2014, 45, 056-060.	0.6	16
84	Lipid-enriched diet rescues lethality and slows down progression in a murine model of VCP-associated disease. Human Molecular Genetics, 2014, 23, 1333-1344.	2.9	20
85	Familial incidence and associated symptoms in a population of individuals with nonsyndromic craniosynostosis. Genetics in Medicine, 2014, 16, 302-310.	2.4	70
86	Dysmorphology of inborn errors of metabolism. Molecular Cytogenetics, 2014, 7, I39.	0.9	0
87	Clinical utility and dilemmas of SNP microarray testing. Molecular Cytogenetics, 2014, 7, I34.	0.9	0
88	Frequency of Praderâ€“Willi syndrome in births conceived via assisted reproductive technology. Genetics in Medicine, 2014, 16, 164-169.	2.4	20
89	A splice donor mutation inNAA10results in the dysregulation of the retinoic acid signalling pathway and causes Lenz microphthalmia syndrome. Journal of Medical Genetics, 2014, 51, 185-196.	3.2	86
90	The Influence of Diet and Exercise on the Physical Health of Affected Individuals with VCP Disease. International Journal of Biotechnology for Wellness Industries, 2014, 3, 46-52.	0.3	4

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91	Clinical and radiological features in young individuals with nevoid basal cell carcinoma syndrome. Genetics in Medicine, 2013, 15, 79-83.	2.4	43
92	Clinical exome sequencing leads to the diagnosis of mitochondrial complex I deficiency in a family with global developmental delays, ataxia, and cerebellar and pons hypoplasia. Mitochondrion, 2013, 13, 942.	3.4	0
93	A progressive translational mouse model of human valosinâ€containing protein disease: The VCP^{R155H/+} mouse. Muscle and Nerve, 2013, 47, 260-270.	2.2	58
94	Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. Nature, 2013, 495, 467-473.	27.8	1,249
95	The Novel Desmin Mutant p.A120D Impairs Filament Formation, Prevents Intercalated Disk Localization, and Causes Sudden Cardiac Death. Circulation: Cardiovascular Genetics, 2013, 6, 615-623.	5.1	46
96	Exercise Training Reverses Skeletal Muscle Atrophy in an Experimental Model of VCP Disease. PLoS ONE, 2013, 8, e76187.	2.5	24
97	Methylation-Specific Multiplex Ligation-Dependent Probe Amplification and Identification of Deletion Genetic Subtypes in Prader-Willi Syndrome. Genetic Testing and Molecular Biomarkers, 2012, 16, 178-186.	0.7	37
98	A genome-wide association study identifies susceptibility loci for nonsyndromic sagittal craniosynostosis near BMP2 and within BBS9. Nature Genetics, 2012, 44, 1360-1364.	21.4	120
99	Clinical geneticists' views of VACTERL/VATER association. American Journal of Medical Genetics, Part A, 2012, 158A, 3087-3100.	1.2	78
100	Valosin-containing protein mutation and Parkinsonâ€™s disease. Parkinsonism and Related Disorders, 2012, 18, 107-109.	2.2	53
101	ALX4 gain-of-function mutations in nonsyndromic craniosynostosis. Human Mutation, 2012, 33, 1626-1629.	2.5	28
102	Genetics of hearing loss: where are we standing now?. European Archives of Oto-Rhino-Laryngology, 2012, 269, 1733-1745.	1.6	23
103	Primate Genome Gain and Loss: A Bone Dysplasia, Muscular Dystrophy, and Bone Cancer Syndrome Resulting from Mutated Retroviral-Derived MTAP Transcripts. American Journal of Human Genetics, 2012, 90, 614-627.	6.2	39
104	Radiological features of Paget disease of bone associated with VCP myopathy. Skeletal Radiology, 2012, 41, 329-337.	2.0	15
105	The Homozygote VCPR155H/R155H Mouse Model Exhibits Accelerated Human VCP-Associated Disease Pathology. PLoS ONE, 2012, 7, e46308.	2.5	56
106	Mitochondrial dysfunction in CA1 hippocampal neurons of the UBE3A deficient mouse model for Angelman syndrome. Neuroscience Letters, 2011, 487, 129-133.	2.1	65
107	The Multiple Faces of Valosin-Containing Protein-Associated Diseases: Inclusion Body Myopathy with Pagetâ€™s Disease of Bone, Frontotemporal Dementia, and Amyotrophic Lateral Sclerosis. Journal of Molecular Neuroscience, 2011, 45, 522-531.	2.3	126
108	Nutritional phases in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1040-1049.	1.2	325

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109	Growth Standards of Infants With Prader-Willi Syndrome. Pediatrics, 2011, 127, 687-695.	2.1	53
110	Molecular analysis expands the spectrum of phenotypes associated with GLI3 mutations. Human Mutation, 2010, 31, 1142-1154.	2.5	111
111	Doubleâ€blind therapeutic trial in Angelman syndrome using betaine and folic acid. American Journal of Medical Genetics, Part A, 2010, 152A, 1994-2001.	1.2	37
112	VCP/p97 is essential for maturation of ubiquitin-containing autophagosomes and this function is impaired by mutations that cause IBMPFD. Autophagy, 2010, 6, 217-227.	9.1	389
113	VCP Associated Inclusion Body Myopathy and Paget Disease of Bone Knock-In Mouse Model Exhibits Tissue Pathology Typical of Human Disease. PLoS ONE, 2010, 5, e13183.	2.5	109
114	Is gestation in Prader-Willi syndrome affected by the genetic subtype?. Journal of Assisted Reproduction and Genetics, 2009, 26, 461-466.	2.5	43
115	Valosin-containing protein disease: Inclusion body myopathy with Pagetâ€™s disease of the bone and fronto-temporal dementia. Neuromuscular Disorders, 2009, 19, 308-315.	0.6	205
116	Valosin containing protein associated inclusion body myopathy: abnormal vacuolization, autophagy and cell fusion in myoblasts. Neuromuscular Disorders, 2009, 19, 766-772.	0.6	59
117	Humoral Immune Deficiency and Hemifacial Microsomia Seen in One Family. Cleft Palate-Craniofacial Journal, 2009, 46, 477-480.	0.9	2
118	Specific Loss of Histone H3 Lysine 9 Trimethylation and HP1Î³/Cohesin Binding at D4Z4 Repeats Is Associated with Facioscapulohumeral Dystrophy (FSHD). PLoS Genetics, 2009, 5, e1000559.	3.5	234
119	Identification of 34 novel and 56 knownFOXL2mutations in patients with blepharophimosis syndrome. Human Mutation, 2008, 29, E205-E219.	2.5	42
120	Clinical studies in familial VCP myopathy associated with Paget disease of bone and frontotemporal dementia. American Journal of Medical Genetics, Part A, 2008, 146A, 745-757.	1.2	153
121	VCP disease associated with myopathy, Paget disease of bone and frontotemporal dementia: Review of a unique disorder. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2008, 1782, 744-748.	3.8	202
122	TDP-43 accumulation in inclusion body myopathy muscle suggests a common pathogenic mechanism with frontotemporal dementia. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 79, 1186-1189.	1.9	249
123	Subglossopalatal Synechia in Association with Cardiac and Digital Anomalies. Cleft Palate-Craniofacial Journal, 2008, 45, 217-221.	0.9	6
124	APOE is a potential modifier gene in an autosomal dominant form of frontotemporal dementia (IBMPFD). Genetics in Medicine, 2007, 9, 9-13.	2.4	46
125	Pathological consequences of VCP mutations on human striated muscle. Brain, 2007, 130, 381-393.	7.6	148
126	TDP-43 in the Ubiquitin Pathology of Frontotemporal Dementia With VCP Gene Mutations. Journal of Neuropathology and Experimental Neurology, 2007, 66, 152-157.	1.7	295

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127	Identification of novel deletions of 15q11q13 in Angelman syndrome by array-CGH: molecular characterization and genotypeâ€“phenotype correlations. European Journal of Human Genetics, 2007, 15, 943-949.	2.8	75
128	What Syndrome Is This?. Pediatric Dermatology, 2007, 24, 306-308.	0.9	9
129	Genetics of Craniosynostosis. Seminars in Pediatric Neurology, 2007, 14, 150-161.	2.0	155
130	Evaluation of the role of Valosin-containing protein in the pathogenesis of familial and sporadic Paget's disease of bone. Bone, 2006, 38, 280-285.	2.9	38
131	Recurrent miscarriage in a carrier of a balanced cytogenetically undetectable subtelomeric rearrangement: How many are we missing?. Prenatal Diagnosis, 2006, 26, 291-293.	2.3	4
132	Apert syndrome: what prenatal radiographic findings should prompt its consideration?. Prenatal Diagnosis, 2006, 26, 966-972.	2.3	25
133	Duplication of 5q15-q23.2: Case report and literature review. Birth Defects Research Part A: Clinical and Molecular Teratology, 2006, 76, 272-276.	1.6	8
134	Findings from aCGH in patients with congenital diaphragmatic hernia (CDH): A possible locus for Fryns syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 17-23.	1.2	67
135	Novel Ubiquitin Neuropathology in Frontotemporal Dementia WithValosin-Containing ProteinGene Mutations. Journal of Neuropathology and Experimental Neurology, 2006, 65, 571-581.	1.7	206
136	Genomewide scans in North American families reveal genetic linkage of essential tremor to a region on chromosome 6p23. Brain, 2006, 129, 2318-2331.	7.6	132
137	Peters Anomaly in Association with Multiple Midline Anomalies and a Familial Chromosome 4 Inversion. Ophthalmic Genetics, 2006, 27, 63-65.	1.2	14
138	Autosomal Dominant Inclusion Body Myopathy, Paget Disease of Bone, and Frontotemporal Dementia. Alzheimer Disease and Associated Disorders, 2005, 19, S44-S47.	1.3	51
139	Mutant valosinâ€containing protein causes a novel type of frontotemporal dementia. Annals of Neurology, 2005, 57, 457-461.	5.3	160
140	Role of ?-galactosidase and elastin binding protein in lysosomal and nonlysosomal complexes of patients with GM1-gangliosidosis. Human Mutation, 2005, 25, 285-292.	2.5	43
141	Smith-Lemli-Opitz syndrome in trisomy 13: How does the mix work?. Birth Defects Research Part A: Clinical and Molecular Teratology, 2005, 73, 569-571.	1.6	2
142	Mapping autosomal dominant progressive limb-girdle myopathy with bone fragility to chromosome 9p21-p22: a novel locus for a musculoskeletal syndrome. Human Genetics, 2005, 118, 508-514.	3.8	4
143	Krabbe Disease: Severe Neonatal Presentation With a Family History of Multiple Sclerosis. Journal of Child Neurology, 2005, 20, 826-828.	1.4	13
144	Radiological features in 82 patients with nevoid basal cell carcinoma (NBCC or Gorlin) syndrome. Genetics in Medicine, 2004, 6, 495-502.	2.4	161

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145	Inclusion body myopathy associated with Paget disease of bone and frontotemporal dementia is caused by mutant valosin-containing protein. Nature Genetics, 2004, 36, 377-381.	21.4	1,257
146	Reciprocal fusion transcripts of two novel Zn-finger genes in a female with absence of the corpus callosum, ocular colobomas and a balanced translocation between chromosomes 2p24 and 9q32. European Journal of Human Genetics, 2003, 11, 527-534.	2.8	36
147	Clinical and genetic heterogeneity in chromosome 9p associated hereditary inclusion body myopathy: exclusion of GNE and three other candidate genes. Neuromuscular Disorders, 2003, 13, 559-567.	0.6	50
148	Temtamy-like syndrome associated with translocation of 2p24 and 9q32. Clinical Dysmorphology, 2003, 12, 175-177.	0.3	14
149	Clinical Delineation and Localization to Chromosome 9p13.3â€“p12 of a Unique Dominant Disorder in Four Families: Hereditary Inclusion Body Myopathy, Paget Disease of Bone, and Frontotemporal Dementia. Molecular Genetics and Metabolism, 2001, 74, 458-475.	1.1	191
150	Genetic heterogeneity in autosomal dominant essential tremor. Genetics in Medicine, 2001, 3, 197-199.	2.4	62
151	Manifestations in four males with and an obligate carrier of the Lenz microphthalmia syndrome. American Journal of Medical Genetics Part A, 2001, 98, 92-100.	2.4	30
152	Chest wall hamartoma with Wiedemann-Beckwith syndrome: Clinical report and brief review of chromosome 11p15.5-related tumors. American Journal of Medical Genetics Part A, 2001, 101, 221-225.	2.4	14
153	Increased fertility in a woman with classic galactosaemia. Journal of Inherited Metabolic Disease, 2001, 24, 507-608.	3.6	14
154	Clinical and molecular studies in a unique family with autosomal dominant limb-girdle muscular dystrophy and Paget disease of bone. Genetics in Medicine, 2000, 2, 232-241.	2.4	121
155	Molecular mechanism for duplication 17p11.2â€” the homologous recombination reciprocal of the Smith-Magenis microdeletion. Nature Genetics, 2000, 24, 84-87.	21.4	297
156	Clinical heterogeneity in autosomal dominant optic atrophy in two 3q28-qter linked central Illinois families. Genetics in Medicine, 2000, 2, 283-289.	2.4	8
157	A Unique Point Mutation in the PMP22 Gene Is Associated with Charcot-Marie-Tooth Disease and Deafness. American Journal of Human Genetics, 1999, 64, 1580-1593.	6.2	74
158	Possible new autosomal recessive syndrome of partial agenesis of the corpus callosum, pontine hypoplasia, focal white matter changes, hypotonia, mental retardation, and minor anomalies. American Journal of Medical Genetics Part A, 1997, 73, 184-188.	2.4	6
159	A Mutation in the V1 End Domain of Keratin 1 in Non-Epidermolytic Palmar-Plantar Keratoderma. Journal of Investigative Dermatology, 1994, 103, 764-769.	0.7	139