Virginia E Kimonis

List of Publications by Year in Descending Order

151 papers	8,027 citations	44 h-index	88 g-index
164 ext. papers	9,166 ext. citations	4.9 avg, IF	5.37 L-index

#	Paper	IF	Citations
151	Development of a standard of care for patients with valosin-containing protein associated multisystem proteinopathy.. *Orphanet Journal of Rare Diseases*, **2022**, 17, 23	4.2	4
150	VCP/p97 inhibitor CB-5083 modulates muscle pathology in a mouse model of VCP inclusion body myopathy.. *Journal of Translational Medicine*, **2022**, 20, 21	8.5	0
149	Severe cardiomyopathy associated with the VCP p.R155C and c.177_187del MYBPC3 gene variants.. *European Journal of Medical Genetics*, **2022**, 104480	2.6	1
148	Pathogenic variants of Valosin Containing Protein induce lysosomal damage and transcriptional activation of autophagy regulators in neuronal cells.. *Neuropathology and Applied Neurobiology*, **2022**, e12818	5.2	0
147	Multisystem Proteinopathy Due to VCP Mutations: A Review of Clinical Heterogeneity and Genetic Diagnosis. *Genes*, **2022**, 13, 963	4.2	2
146	Prevalence of cerebral small vessel disease in a Fabry disease cohort. *Molecular Genetics and Metabolism Reports*, **2021**, 29, 100815	1.8	1
145	Ceramide contributes to pathogenesis and may be targeted for therapy in VCP inclusion body myopathy. *Human Molecular Genetics*, **2021**, 29, 3945-3953	5.6	2
144	Stroke and Chronic Kidney Disease in Fabry Disease. *Journal of Stroke and Cerebrovascular Diseases*, **2021**, 30, 105423	2.8	2
143	Variable clinical features of patients with Fabry disease and outcome of enzyme replacement therapy. *Molecular Genetics and Metabolism Reports*, **2021**, 26, 100700	1.8	1
142	Influence of molecular classes and growth hormone treatment on growth and dysmorphology in Prader-Willi syndrome: A multicenter study. *Clinical Genetics*, **2021**, 100, 29-39	4	3
141	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	4.7	4
140	Mosaic de novo gene variant associated with Prader-Willi syndrome. *Journal of Medical Genetics*, **2021**,	5.8	1
139	mitochondrial disease: new patients and review of the genetic and clinical spectrum. *Journal of Medical Genetics*, **2021**, 58, 314-325	5.8	2
138	Multisystem proteinopathy: Where myopathy and motor neuron disease converge. *Muscle and Nerve*, **2021**, 63, 442-454	3.4	8
137	Ataxia and Parkinsonism in a Woman With a VCP Variant and Long-Normal Repeats in the SCA2 Allele. *Neurology: Genetics*, **2021**, 7, e595	3.8	2
136	Characteristics of VCP mutation-associated cardiomyopathy. *Neuromuscular Disorders*, **2021**, 31, 701-705	2.9	3
135	Severe manifestations and treatment of COVID-19 in a transplanted patient with Fabry disease. *Molecular Genetics and Metabolism Reports*, **2021**, 29, 100802	1.8	0

134	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	2.6	0
133	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. *Pediatric Neurology*, **2021**, 123, 30-37	2.9	3
132	Regional Strain Pattern and Correlation with Cardiac Magnetic Resonance Imaging in Fabry Disease.. *Journal of Cardiovascular Echography*, **2021**, 31, 131-136	0.6	
131	Diffuse large B-cell non-Hodgkin lymphoma in Gaucher disease. *Molecular Genetics and Metabolism Reports*, **2020**, 25, 100663	1.8	0
130	Expression level of R155H mRNA in the knock-in mouse model. *Biochemical and Biophysical Research Communications*, **2020**, 523, 985-986	3.4	
129	Hereditary Leiomyomatosis and Renal Cell Cancer (HLRCC): Report of a Family Pedigree. *American Journal of the Medical Sciences*, **2020**, 360, 724-727	2.2	
128	Homozygous B4GALNT1 mutation and biochemical glutaric acidemia type II: A case report. *Clinical Neurology and Neurosurgery*, **2020**, 189, 105553	2	2
127	Molecular subtype and growth hormone effects on dysmorphology in Prader-Willi syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 169-175	2.5	3
126	Genetic Subtype-Phenotype Analysis of Growth Hormone Treatment on Psychiatric Behavior in Prader-Willi Syndrome. *Genes*, **2020**, 11,	4.2	4
125	A placebo-controlled trial of folic acid and betaine in identical twins with Angelman syndrome. *Orphanet Journal of Rare Diseases*, **2019**, 14, 232	4.2	1
124	A randomized pilot efficacy and safety trial of diazoxide choline controlled-release in patients with Prader-Willi syndrome. *PLoS ONE*, **2019**, 14, e0221615	3.7	16
123	ALG11-CDG syndrome: Expanding the phenotype. *American Journal of Medical Genetics, Part A*, **2019**, 179, 498-502	2.5	3
122	Birth seasonality studies in a large Prader-Willi syndrome cohort. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1531-1534	2.5	2
121	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	20.1	10
120	Homozygosity for the A431E mutation in PSEN1 presenting with a relatively aggressive phenotype. *Neuroscience Letters*, **2019**, 699, 195-198	3.3	4
119	Antisense oligonucleotide treatment targeting glycogen synthase (GYS1) in a mouse model of Pompe disease. *Molecular Genetics and Metabolism*, **2019**, 126, S85	3.7	5
118	Meta-analysis of genotype-phenotype analysis of OPA1 mutations in autosomal dominant optic atrophy. *Mitochondrion*, **2019**, 46, 262-269	4.9	16
117	Molecular genetic classification in Prader-Willi syndrome: a multisite cohort study. *Journal of Medical Genetics*, **2019**, 56, 149-153	5.8	64

116	Impact of genetic subtypes of Prader-Willi syndrome with growth hormone therapy on intelligence and body mass index. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1826-1835	2.5	12
115	GAA variants and phenotypes among 1,079 patients with Pompe disease: Data from the Pompe Registry. *Human Mutation*, **2019**, 40, 2146-2164	4.7	26
114	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	5.8	14
113	Early Diagnosis in Prader-Willi Syndrome Reduces Obesity and Associated Co-Morbidities. *Genes*, **2019**, 10,	4.2	14
112	Variable clinical features and genotype-phenotype correlations in 18 patients with late-onset Pompe disease. *Annals of Translational Medicine*, **2019**, 7, 276	3.2	7
111	New family with 8-associated autosomal dominant rimmed vacuolar myopathy. *Neurology: Genetics*, **2019**, 5, e349	3.8	17
110	Contributing factors of mortality in Prader-Willi syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 196-205	2.5	27
109	Cornelia de Lange syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2019**, 179, 150-158	2.5	25
108	Newborn screening for Prader-Willi syndrome is feasible: Early diagnosis for better outcomes. *American Journal of Medical Genetics, Part A*, **2019**, 179, 29-36	2.5	5
107	Comparison of perinatal factors in deletion versus uniparental disomy in Prader-Willi syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1161-1165	2.5	8
106	Novel valosin-containing protein mutations associated with multisystem proteinopathy. *Neuromuscular Disorders*, **2018**, 28, 491-501	2.9	15
105	Prader-Willi syndrome and early-onset morbid obesity NIH rare disease consortium: A review of natural history study. *American Journal of Medical Genetics, Part A*, **2018**, 176, 368-375	2.5	24
104	Genotype-phenotype study in patients with valosin-containing protein mutations associated with multisystem proteinopathy. *Clinical Genetics*, **2018**, 93, 119-125	4	55
103	A cross-sectional analysis of clinical evaluation in 35 individuals with mutations of the valosin-containing protein gene. *Neuromuscular Disorders*, **2018**, 28, 778-786	2.9	3
102	Phenotypic diversity of patients diagnosed with VACTERL association. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1830-1837	2.5	3
101	Pathogenic mutations in NUBPL affect complex I activity and cold tolerance in the yeast model Yarrowia lipolytica. *Human Molecular Genetics*, **2018**, 27, 3697-3709	5.6	7
100	Multicentre study of maternal and neonatal outcomes in individuals with Prader-Willi syndrome. *Journal of Medical Genetics*, **2018**, 55, 594-598	5.8	16
99	Prospective phenotyping of NGLY1-CDDG, the first congenital disorder of deglycosylation. *Genetics in Medicine*, **2017**, 19, 160-168	8.1	72

98	Two cases of Legg-Perthes and intellectual disability in Tricho-Rhino-Phalangeal syndrome type 1 associated with novel TRPS1 mutations. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1663-1667	2.5	2
97	Oxytocin treatment in children with Prader-Willi syndrome: A double-blind, placebo-controlled, crossover study. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1243-1250	2.5	64
96	Dysfunctional oleoylethanolamide signaling in a mouse model of Prader-Willi syndrome. *Pharmacological Research*, **2017**, 117, 75-81	10.2	14
95	Myogenic differentiation of VCP disease-induced pluripotent stem cells: A novel platform for drug discovery. *PLoS ONE*, **2017**, 12, e0176919	3.7	9
94	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	0.9	2
93	Activation of the NLRP3 Inflammasome Is Associated with Valosin-Containing Protein Myopathy. *Inflammation*, **2017**, 40, 21-41	5.1	21
92	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	1.9	4
91	215th ENMC International Workshop VCP-related multi-system proteinopathy (IBMPFD) 13-15 November 2015, Heemskerk, The Netherlands. *Neuromuscular Disorders*, **2016**, 26, 535-47	2.9	24
90	Growth Charts for Prader-Willi Syndrome During Growth Hormone Treatment. *Clinical Pediatrics*, **2016**, 55, 957-74	1.2	28
89	DVL3 Alleles Resulting in a -1 Frameshift of the Last Exon Mediate Autosomal-Dominant Robinow Syndrome. *American Journal of Human Genetics*, **2016**, 98, 553-561	11	63
88	Gain-of-Function Mutations in RARB Cause Intellectual Disability with Progressive Motor Impairment. *Human Mutation*, **2016**, 37, 786-93	4.7	24
87	Prader-Willi Syndrome due to an Unbalanced de novo Translocation t(15;19)(q12;p13.3). *Cytogenetic and Genome Research*, **2016**, 150, 29-34	1.9	3
86	The Myoblast C2C12 Transfected with Mutant Valosin-Containing Protein Exhibits Delayed Stress Granule Resolution on Oxidative Stress. *American Journal of Pathology*, **2016**, 186, 1623-34	5.8	8
85	In vitro studies in VCP-associated multisystem proteinopathy suggest altered mitochondrial bioenergetics. *Mitochondrion*, **2015**, 22, 1-8	4.9	15
84	Psychological Impact of Predictive Genetic Testing in VCP Inclusion Body Myopathy, Paget Disease of Bone and Frontotemporal Dementia. *Journal of Genetic Counseling*, **2015**, 24, 842-50	2.5	12
83	Clinical characterization of int22h1/int22h2-mediated Xq28 duplication/deletion: new cases and literature review. *BMC Medical Genetics*, **2015**, 16, 12	2.1	22
82	Targeted excision of VCP R155H mutation by Cre-LoxP technology as a promising therapeutic strategy for valosin-containing protein disease. *Human Gene Therapy Methods*, **2015**, 26, 13-24	4.9	9
81	Growth charts for non-growth hormone treated Prader-Willi syndrome. *Pediatrics*, **2015**, 135, e126-35	7.4	43

80	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	3.7	52
79	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	7.5	22
78	A case report comparing clinical, imaging and neuropsychological assessment findings in twins discordant for the VCP p.R155C mutation. *Neuromuscular Disorders*, **2015**, 25, 177-83	2.9	5
77	A Fine Balance of Dietary Lipids Improves Pathology of a Murine Model of VCP-Associated Multisystem Proteinopathy. *PLoS ONE*, **2015**, 10, e0131995	3.7	3
76	Dysmorphology of inborn errors of metabolism. *Molecular Cytogenetics*, **2014**, 7, I39	2	
75	Clinical utility and dilemmas of SNP microarray testing. *Molecular Cytogenetics*, **2014**, 7, I34	2	
74	Frequency of Prader-Willi syndrome in births conceived via assisted reproductive technology. *Genetics in Medicine*, **2014**, 16, 164-9	8.1	17
73	A splice donor mutation in NAA10 results in the dysregulation of the retinoic acid signalling pathway and causes Lenz microphthalmia syndrome. *Journal of Medical Genetics*, **2014**, 51, 185-96	5.8	65
72	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	1.6	15
71	Mild phenotype in a male with pyruvate dehydrogenase complex deficiency associated with novel hemizygous in-frame duplication of the E1 subunit gene (PDHA1). *Neuropediatrics*, **2014**, 45, 56-60	1.6	15
70	Lipid-enriched diet rescues lethality and slows down progression in a murine model of VCP-associated disease. *Human Molecular Genetics*, **2014**, 23, 1333-44	5.6	15
69	Familial incidence and associated symptoms in a population of individuals with nonsyndromic craniosynostosis. *Genetics in Medicine*, **2014**, 16, 302-10	8.1	47
68	Clinical and radiological features in young individuals with nevoid basal cell carcinoma syndrome. *Genetics in Medicine*, **2013**, 15, 79-83	8.1	30
67	A progressive translational mouse model of human valosin-containing protein disease: the VCP(R155H/+) mouse. *Muscle and Nerve*, **2013**, 47, 260-70	3.4	48
66	Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. *Nature*, **2013**, 495, 467-73	50.4	965
65	The novel desmin mutant p.A120D impairs filament formation, prevents intercalated disk localization, and causes sudden cardiac death. *Circulation: Cardiovascular Genetics*, **2013**, 6, 615-23		33
64	Exercise training reverses skeletal muscle atrophy in an experimental model of VCP disease. *PLoS ONE*, **2013**, 8, e76187	3.7	18
63	Radiological features of Paget disease of bone associated with VCP myopathy. *Skeletal Radiology*, **2012**, 41, 329-37	2.7	11

62	Clinical geneticists' views of VACTERL/VATER association. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 3087-100	2.5	57
61	Valosin-containing protein mutation and Parkinson's disease. *Parkinsonism and Related Disorders*, **2012**, 18, 107-9	3.6	39
60	ALX4 gain-of-function mutations in nonsyndromic craniosynostosis. *Human Mutation*, **2012**, 33, 1626-9	4.7	22
59	Genetics of hearing loss: where are we standing now?. *European Archives of Oto-Rhino-Laryngology*, **2012**, 269, 1733-45	3.5	20
58	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, 614-27	11	30
57	Methylation-specific multiplex ligation-dependent probe amplification and identification of deletion genetic subtypes in Prader-Willi syndrome. *Genetic Testing and Molecular Biomarkers*, **2012**, 16, 178-86	1.6	28
56	A genome-wide association study identifies susceptibility loci for nonsyndromic sagittal craniosynostosis near BMP2 and within BBS9. *Nature Genetics*, **2012**, 44, 1360-4	36.3	93
55	The homozygote VCP(R□□H/R□□H) mouse model exhibits accelerated human VCP-associated disease pathology. *PLoS ONE*, **2012**, 7, e46308	3.7	45
54	Mitochondrial dysfunction in CA1 hippocampal neurons of the UBE3A deficient mouse model for Angelman syndrome. *Neuroscience Letters*, **2011**, 487, 129-33	3.3	53
53	The multiple faces of valosin-containing protein-associated diseases: inclusion body myopathy with Paget's disease of bone, frontotemporal dementia, and amyotrophic lateral sclerosis. *Journal of Molecular Neuroscience*, **2011**, 45, 522-31	3.3	110
52	Nutritional phases in Prader-Willi syndrome. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1040-9	2.5	235
51	Growth standards of infants with Prader-Willi syndrome. *Pediatrics*, **2011**, 127, 687-95	7.4	44
50	VCP/p97 is essential for maturation of ubiquitin-containing autophagosomes and this function is impaired by mutations that cause IBMPFD. *Autophagy*, **2010**, 6, 217-27	10.2	339
49	Molecular analysis expands the spectrum of phenotypes associated with GLI3 mutations. *Human Mutation*, **2010**, 31, 1142-54	4.7	90
48	Double-blind therapeutic trial in Angelman syndrome using betaine and folic acid. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1994-2001	2.5	33
47	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	3.7	100
46	Is gestation in Prader-Willi syndrome affected by the genetic subtype?. *Journal of Assisted Reproduction and Genetics*, **2009**, 26, 461-6	3.4	39
45	Valosin-containing protein disease: inclusion body myopathy with Paget's disease of the bone and fronto-temporal dementia. *Neuromuscular Disorders*, **2009**, 19, 308-15	2.9	164

44	Valosin containing protein associated inclusion body myopathy: abnormal vacuolization, autophagy and cell fusion in myoblasts. *Neuromuscular Disorders*, **2009**, 19, 766-72	2.9	53
43	Humoral immune deficiency and hemifacial microsomia seen in one family. *Cleft Palate-Craniofacial Journal*, **2009**, 46, 477-80	1.9	1
42	Specific loss of histone H3 lysine 9 trimethylation and HP1gamma/cohesin binding at D4Z4 repeats is associated with facioscapulohumeral dystrophy (FSHD). *PLoS Genetics*, **2009**, 5, e1000559	6	207
41	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, 744-8	6.9	163
40	TDP-43 accumulation in inclusion body myopathy muscle suggests a common pathogenic mechanism with frontotemporal dementia. *Journal of Neurology, Neurosurgery and Psychiatry*, **2008** , 79, 1186-9	5.5	206
39	Subglossopalatal synechia in association with cardiac and digital anomalies. *Cleft Palate-Craniofacial Journal*, **2008**, 45, 217-21	1.9	3
38	Identification of 34 novel and 56 known FOXL2 mutations in patients with Blepharophimosis syndrome. *Human Mutation*, **2008**, 29, E205-19	4.7	41
37	Clinical studies in familial VCP myopathy associated with Paget disease of bone and frontotemporal dementia. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 745-57	2.5	127
36	Identification of novel deletions of 15q11q13 in Angelman syndrome by array-CGH: molecular characterization and genotype-phenotype correlations. *European Journal of Human Genetics*, **2007**, 15, 943-9	5.3	71
35	What syndrome is this? Laryngo-onycho-cutaneous syndrome. *Pediatric Dermatology*, **2007**, 24, 306-8	1.9	7
34	Genetics of craniosynostosis. *Seminars in Pediatric Neurology*, **2007**, 14, 150-61	2.9	127
33	APOE is a potential modifier gene in an autosomal dominant form of frontotemporal dementia (IBMPFD). *Genetics in Medicine*, **2007**, 9, 9-13	8.1	39
32	Pathological consequences of VCP mutations on human striated muscle. *Brain*, **2007**, 130, 381-93	11.2	128
31	TDP-43 in the ubiquitin pathology of frontotemporal dementia with VCP gene mutations. *Journal of Neuropathology and Experimental Neurology*, **2007**, 66, 152-7	3.1	256
30	Duplication of 5q15-q23.2: case report and literature review. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2006**, 76, 272-6		7
29	Findings from aCGH in patients with congenital diaphragmatic hernia (CDH): a possible locus for Fryns syndrome. *American Journal of Medical Genetics, Part A*, **2006**, 140, 17-23	2.5	58
28	Novel ubiquitin neuropathology in frontotemporal dementia with valosin-containing protein gene mutations. *Journal of Neuropathology and Experimental Neurology*, **2006**, 65, 571-81	3.1	182
27	Genomewide scans in North American families reveal genetic linkage of essential tremor to a region on chromosome 6p23. *Brain*, **2006**, 129, 2318-31	11.2	112

26	Peters anomaly in association with multiple midline anomalies and a familial chromosome 4 inversion. *Ophthalmic Genetics*, **2006**, 27, 63-5	1.2	10
25	Evaluation of the role of Valosin-containing protein in the pathogenesis of familial and sporadic Paget's disease of bone. *Bone*, **2006**, 38, 280-5	4.7	35
24	Recurrent miscarriage in a carrier of a balanced cytogenetically undetectable subtelomeric rearrangement: how many are we missing?. *Prenatal Diagnosis*, **2006**, 26, 291-3	3.2	3
23	Apert syndrome: what prenatal radiographic findings should prompt its consideration?. *Prenatal Diagnosis*, **2006**, 26, 966-72	3.2	20
22	Autosomal dominant inclusion body myopathy, Paget disease of bone, and frontotemporal dementia. *Alzheimer Disease and Associated Disorders*, **2005**, 19 Suppl 1, S44-7	2.5	45
21	Mutant valosin-containing protein causes a novel type of frontotemporal dementia. *Annals of Neurology*, **2005**, 57, 457-61	9.4	131
20	Role of beta-galactosidase and elastin binding protein in lysosomal and nonlysosomal complexes of patients with GM1-gangliosidosis. *Human Mutation*, **2005**, 25, 285-92	4.7	42
19	Smith-Lemli-Opitz syndrome in trisomy 13: how does the mix work?. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2005**, 73, 569-71		2
18	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, 508-14	6.3	4
17	Krabbe disease: severe neonatal presentation with a family history of multiple sclerosis. *Journal of Child Neurology*, **2005**, 20, 826-8	2.5	12
16	Radiological features in 82 patients with nevoid basal cell carcinoma (NBCC or Gorlin) syndrome. *Genetics in Medicine*, **2004**, 6, 495-502	8.1	129
15	Inclusion body myopathy associated with Paget disease of bone and frontotemporal dementia is caused by mutant valosin-containing protein. *Nature Genetics*, **2004**, 36, 377-81	36.3	1088
14	Temtamy-like syndrome associated with translocation of 2p24 and 9q32. *Clinical Dysmorphology*, **2003**, 12, 175-7	0.9	12
13	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, 527-34	5.3	29
12	Clinical and genetic heterogeneity in chromosome 9p associated hereditary inclusion body myopathy: exclusion of GNE and three other candidate genes. *Neuromuscular Disorders*, **2003**, 13, 559-67	2.9	44
11	Genetic heterogeneity in autosomal dominant essential tremor. *Genetics in Medicine*, **2001**, 3, 197-9	8.1	58
10	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		27
9	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, 221-5		13

8	Increased fertility in a woman with classic galactosaemia. *Journal of Inherited Metabolic Disease*, **2001**, 24, 607-8	5.4	13
7	Clinical delineation and localization to chromosome 9p13.3-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, 458-75	3.7	168
6	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, 232-41	8.1	101
5	Molecular mechanism for duplication 17p11.2- the homologous recombination reciprocal of the Smith-Magenis microdeletion. *Nature Genetics*, **2000**, 24, 84-7	36.3	262
4	Clinical heterogeneity in autosomal dominant optic atrophy in two 3q28-qter linked central Illinois families. *Genetics in Medicine*, **2000**, 2, 283-9	8.1	8
3	A unique point mutation in the PMP22 gene is associated with Charcot-Marie-Tooth disease and deafness. *American Journal of Human Genetics*, **1999**, 64, 1580-93	11	64
2	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		5
1	A mutation in the V1 end domain of keratin 1 in non-epidermolytic palmar-plantar keratoderma. *Journal of Investigative Dermatology*, **1994**, 103, 764-9	4.3	120