

# Virginia E Kimonis

## List of Publications by Citations

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151  
papers

8,027  
citations

44  
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164  
ext. papers

9,166  
ext. citations

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avg, IF

5.37  
L-index

#	Paper	IF	Citations
151	Inclusion body myopathy associated with Paget disease of bone and frontotemporal dementia is caused by mutant valosin-containing protein. <i>Nature Genetics</i> , <b>2004</b> , 36, 377-81	36.3	1088
150	Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. <i>Nature</i> , <b>2013</b> , 495, 467-73	50.4	965
149	VCP/p97 is essential for maturation of ubiquitin-containing autophagosomes and this function is impaired by mutations that cause IBMPFD. <i>Autophagy</i> , <b>2010</b> , 6, 217-27	10.2	339
148	Molecular mechanism for duplication 17p11.2- the homologous recombination reciprocal of the Smith-Magenis microdeletion. <i>Nature Genetics</i> , <b>2000</b> , 24, 84-7	36.3	262
147	TDP-43 in the ubiquitin pathology of frontotemporal dementia with VCP gene mutations. <i>Journal of Neuropathology and Experimental Neurology</i> , <b>2007</b> , 66, 152-7	3.1	256
146	Nutritional phases in Prader-Willi syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2011</b> , 155A, 1040-9	2.5	235
145	Specific loss of histone H3 lysine 9 trimethylation and HP1gamma/cohesin binding at D4Z4 repeats is associated with facioscapulohumeral dystrophy (FSHD). <i>PLoS Genetics</i> , <b>2009</b> , 5, e1000559	6	207
144	TDP-43 accumulation in inclusion body myopathy muscle suggests a common pathogenic mechanism with frontotemporal dementia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2008</b> , 79, 1186-9	5.5	206
143	Novel ubiquitin neuropathology in frontotemporal dementia with valosin-containing protein gene mutations. <i>Journal of Neuropathology and Experimental Neurology</i> , <b>2006</b> , 65, 571-81	3.1	182
142	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. <i>Molecular Genetics and Metabolism</i> , <b>2001</b> , 74, 458-75	3.7	168
141	Valosin-containing protein disease: inclusion body myopathy with Paget disease of the bone and fronto-temporal dementia. <i>Neuromuscular Disorders</i> , <b>2009</b> , 19, 308-15	2.9	164
140	VCP disease associated with myopathy, Paget disease of bone and frontotemporal dementia: review of a unique disorder. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2008</b> , 1782, 744-8	6.9	163
139	Mutant valosin-containing protein causes a novel type of frontotemporal dementia. <i>Annals of Neurology</i> , <b>2005</b> , 57, 457-61	9.4	131
138	Radiological features in 82 patients with nevoid basal cell carcinoma (NBCC or Gorlin) syndrome. <i>Genetics in Medicine</i> , <b>2004</b> , 6, 495-502	8.1	129
137	Pathological consequences of VCP mutations on human striated muscle. <i>Brain</i> , <b>2007</b> , 130, 381-93	11.2	128
136	Genetics of craniosynostosis. <i>Seminars in Pediatric Neurology</i> , <b>2007</b> , 14, 150-61	2.9	127
135	Clinical studies in familial VCP myopathy associated with Paget disease of bone and frontotemporal dementia. <i>American Journal of Medical Genetics, Part A</i> , <b>2008</b> , 146A, 745-57	2.5	127

134	A mutation in the V1 end domain of keratin 1 in non-epidermolytic palmar-plantar keratoderma. <i>Journal of Investigative Dermatology</i> , <b>1994</b> , 103, 764-9	4.3	120
133	Genomewide scans in North American families reveal genetic linkage of essential tremor to a region on chromosome 6p23. <i>Brain</i> , <b>2006</b> , 129, 2318-31	11.2	112
132	The multiple faces of valosin-containing protein-associated diseases: inclusion body myopathy with Paget disease of bone, frontotemporal dementia, and amyotrophic lateral sclerosis. <i>Journal of Molecular Neuroscience</i> , <b>2011</b> , 45, 522-31	3.3	110
131	Clinical and molecular studies in a unique family with autosomal dominant limb-girdle muscular dystrophy and Paget disease of bone. <i>Genetics in Medicine</i> , <b>2000</b> , 2, 232-41	8.1	101
130	VCP associated inclusion body myopathy and paget disease of bone knock-in mouse model exhibits tissue pathology typical of human disease. <i>PLoS ONE</i> , <b>2010</b> , 5, e13183	3.7	100
129	A genome-wide association study identifies susceptibility loci for nonsyndromic sagittal craniosynostosis near BMP2 and within BBS9. <i>Nature Genetics</i> , <b>2012</b> , 44, 1360-4	36.3	93
128	Molecular analysis expands the spectrum of phenotypes associated with GLI3 mutations. <i>Human Mutation</i> , <b>2010</b> , 31, 1142-54	4.7	90
127	Prospective phenotyping of NGLY1-CDDG, the first congenital disorder of deglycosylation. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 160-168	8.1	72
126	Identification of novel deletions of 15q11q13 in Angelman syndrome by array-CGH: molecular characterization and genotype-phenotype correlations. <i>European Journal of Human Genetics</i> , <b>2007</b> , 15, 943-9	5.3	71
125	A splice donor mutation in NAA10 results in the dysregulation of the retinoic acid signalling pathway and causes Lenz microphthalmia syndrome. <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 185-96	5.8	65
124	Oxytocin treatment in children with Prader-Willi syndrome: A double-blind, placebo-controlled, crossover study. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 1243-1250	2.5	64
123	Molecular genetic classification in Prader-Willi syndrome: a multisite cohort study. <i>Journal of Medical Genetics</i> , <b>2019</b> , 56, 149-153	5.8	64
122	A unique point mutation in the PMP22 gene is associated with Charcot-Marie-Tooth disease and deafness. <i>American Journal of Human Genetics</i> , <b>1999</b> , 64, 1580-93	11	64
121	DVL3 Alleles Resulting in a -1 Frameshift of the Last Exon Mediate Autosomal-Dominant Robinow Syndrome. <i>American Journal of Human Genetics</i> , <b>2016</b> , 98, 553-561	11	63
120	Findings from aCGH in patients with congenital diaphragmatic hernia (CDH): a possible locus for Fryns syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2006</b> , 140, 17-23	2.5	58
119	Genetic heterogeneity in autosomal dominant essential tremor. <i>Genetics in Medicine</i> , <b>2001</b> , 3, 197-9	8.1	58
118	Clinical geneticists' views of VACTERL/VATER association. <i>American Journal of Medical Genetics, Part A</i> , <b>2012</b> , 158A, 3087-100	2.5	57
117	Genotype-phenotype study in patients with valosin-containing protein mutations associated with multisystem proteinopathy. <i>Clinical Genetics</i> , <b>2018</b> , 93, 119-125	4	55

116	Mitochondrial dysfunction in CA1 hippocampal neurons of the UBE3A deficient mouse model for Angelman syndrome. <i>Neuroscience Letters</i> , <b>2011</b> , 487, 129-33	3.3	53
115	Valosin containing protein associated inclusion body myopathy: abnormal vacuolization, autophagy and cell fusion in myoblasts. <i>Neuromuscular Disorders</i> , <b>2009</b> , 19, 766-72	2.9	53
114	Rapamycin and chloroquine: the in vitro and in vivo effects of autophagy-modifying drugs show promising results in valosin containing protein multisystem proteinopathy. <i>PLoS ONE</i> , <b>2015</b> , 10, e0122888	3.7	52
113	A progressive translational mouse model of human valosin-containing protein disease: the VCP(R155H/+) mouse. <i>Muscle and Nerve</i> , <b>2013</b> , 47, 260-70	3.4	48
112	Familial incidence and associated symptoms in a population of individuals with nonsyndromic craniosynostosis. <i>Genetics in Medicine</i> , <b>2014</b> , 16, 302-10	8.1	47
111	Autosomal dominant inclusion body myopathy, Paget disease of bone, and frontotemporal dementia. <i>Alzheimer Disease and Associated Disorders</i> , <b>2005</b> , 19 Suppl 1, S44-7	2.5	45
110	The homozygote VCP(R155H/R155H) mouse model exhibits accelerated human VCP-associated disease pathology. <i>PLoS ONE</i> , <b>2012</b> , 7, e46308	3.7	45
109	Growth standards of infants with Prader-Willi syndrome. <i>Pediatrics</i> , <b>2011</b> , 127, 687-95	7.4	44
108	Clinical and genetic heterogeneity in chromosome 9p associated hereditary inclusion body myopathy: exclusion of GNE and three other candidate genes. <i>Neuromuscular Disorders</i> , <b>2003</b> , 13, 559-67	2.9	44
107	Growth charts for non-growth hormone treated Prader-Willi syndrome. <i>Pediatrics</i> , <b>2015</b> , 135, e126-35	7.4	43
106	Role of beta-galactosidase and elastin binding protein in lysosomal and nonlysosomal complexes of patients with GM1-gangliosidosis. <i>Human Mutation</i> , <b>2005</b> , 25, 285-92	4.7	42
105	Identification of 34 novel and 56 known FOXL2 mutations in patients with Blepharophimosis syndrome. <i>Human Mutation</i> , <b>2008</b> , 29, E205-19	4.7	41
104	Valosin-containing protein mutation and Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , <b>2012</b> , 18, 107-9	3.6	39
103	Is gestation in Prader-Willi syndrome affected by the genetic subtype?. <i>Journal of Assisted Reproduction and Genetics</i> , <b>2009</b> , 26, 461-6	3.4	39
102	APOE is a potential modifier gene in an autosomal dominant form of frontotemporal dementia (IBMPFD). <i>Genetics in Medicine</i> , <b>2007</b> , 9, 9-13	8.1	39
101	Evaluation of the role of Valosin-containing protein in the pathogenesis of familial and sporadic Paget's disease of bone. <i>Bone</i> , <b>2006</b> , 38, 280-5	4.7	35
100	The novel desmin mutant p.A120D impairs filament formation, prevents intercalated disk localization, and causes sudden cardiac death. <i>Circulation: Cardiovascular Genetics</i> , <b>2013</b> , 6, 615-23		33
99	Double-blind therapeutic trial in Angelman syndrome using betaine and folic acid. <i>American Journal of Medical Genetics, Part A</i> , <b>2010</b> , 152A, 1994-2001	2.5	33

98	Clinical and radiological features in young individuals with nevoid basal cell carcinoma syndrome. <i>Genetics in Medicine</i> , <b>2013</b> , 15, 79-83	8.1	30
97	Primate genome gain and loss: a bone dysplasia, muscular dystrophy, and bone cancer syndrome resulting from mutated retroviral-derived MTAP transcripts. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 614-27	11	30
96	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. <i>European Journal of Human Genetics</i> , <b>2003</b> , 11, 527-34	5.3	29
95	Growth Charts for Prader-Willi Syndrome During Growth Hormone Treatment. <i>Clinical Pediatrics</i> , <b>2016</b> , 55, 957-74	1.2	28
94	Methylation-specific multiplex ligation-dependent probe amplification and identification of deletion genetic subtypes in Prader-Willi syndrome. <i>Genetic Testing and Molecular Biomarkers</i> , <b>2012</b> , 16, 178-86	1.6	28
93	Manifestations in four males with and an obligate carrier of the Lenz microphthalmia syndrome. <i>American Journal of Medical Genetics Part A</i> , <b>2001</b> , 98, 92-100		27
92	Contributing factors of mortality in Prader-Willi syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 196-205	2.5	27
91	GAA variants and phenotypes among 1,079 patients with Pompe disease: Data from the Pompe Registry. <i>Human Mutation</i> , <b>2019</b> , 40, 2146-2164	4.7	26
90	Cornelia de Lange syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 150-158	2.5	25
89	Prader-Willi syndrome and early-onset morbid obesity NIH rare disease consortium: A review of natural history study. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 368-375	2.5	24
88	215th ENMC International Workshop VCP-related multi-system proteinopathy (IBMPFD) 13-15 November 2015, Heemskerk, The Netherlands. <i>Neuromuscular Disorders</i> , <b>2016</b> , 26, 535-47	2.9	24
87	Gain-of-Function Mutations in RARB Cause Intellectual Disability with Progressive Motor Impairment. <i>Human Mutation</i> , <b>2016</b> , 37, 786-93	4.7	24
86	Clinical characterization of int22h1/int22h2-mediated Xq28 duplication/deletion: new cases and literature review. <i>BMC Medical Genetics</i> , <b>2015</b> , 16, 12	2.1	22
85	Administration of CoQ10 analogue ameliorates dysfunction of the mitochondrial respiratory chain in a mouse model of Angelman syndrome. <i>Neurobiology of Disease</i> , <b>2015</b> , 76, 77-86	7.5	22
84	ALX4 gain-of-function mutations in nonsyndromic craniosynostosis. <i>Human Mutation</i> , <b>2012</b> , 33, 1626-9	4.7	22
83	Activation of the NLRP3 Inflammasome Is Associated with Valosin-Containing Protein Myopathy. <i>Inflammation</i> , <b>2017</b> , 40, 21-41	5.1	21
82	Genetics of hearing loss: where are we standing now?. <i>European Archives of Oto-Rhino-Laryngology</i> , <b>2012</b> , 269, 1733-45	3.5	20
81	Apert syndrome: what prenatal radiographic findings should prompt its consideration?. <i>Prenatal Diagnosis</i> , <b>2006</b> , 26, 966-72	3.2	20

80	Exercise training reverses skeletal muscle atrophy in an experimental model of VCP disease. <i>PLoS ONE</i> , <b>2013</b> , 8, e76187	3.7	18
79	Frequency of Prader-Willi syndrome in births conceived via assisted reproductive technology. <i>Genetics in Medicine</i> , <b>2014</b> , 16, 164-9	8.1	17
78	New family with 8-associated autosomal dominant rimmed vacuolar myopathy. <i>Neurology: Genetics</i> , <b>2019</b> , 5, e349	3.8	17
77	A randomized pilot efficacy and safety trial of diazoxide choline controlled-release in patients with Prader-Willi syndrome. <i>PLoS ONE</i> , <b>2019</b> , 14, e0221615	3.7	16
76	Meta-analysis of genotype-phenotype analysis of OPA1 mutations in autosomal dominant optic atrophy. <i>Mitochondrion</i> , <b>2019</b> , 46, 262-269	4.9	16
75	Multicentre study of maternal and neonatal outcomes in individuals with Prader-Willi syndrome. <i>Journal of Medical Genetics</i> , <b>2018</b> , 55, 594-598	5.8	16
74	In vitro studies in VCP-associated multisystem proteinopathy suggest altered mitochondrial bioenergetics. <i>Mitochondrion</i> , <b>2015</b> , 22, 1-8	4.9	15
73	Novel valosin-containing protein mutations associated with multisystem proteinopathy. <i>Neuromuscular Disorders</i> , <b>2018</b> , 28, 491-501	2.9	15
72	Effect of genetic subtypes and growth hormone treatment on bone mineral density in Prader-Willi syndrome. <i>Journal of Pediatric Endocrinology and Metabolism</i> , <b>2014</b> , 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 $\beta$ subunit gene (PDHA1). <i>Neuropediatrics</i> , <b>2014</b> , 45, 56-60	1.6	15
70	Lipid-enriched diet rescues lethality and slows down progression in a murine model of VCP-associated disease. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 1333-44	5.6	15
69	Dysfunctional oleoylethanolamide signaling in a mouse model of Prader-Willi syndrome. <i>Pharmacological Research</i> , <b>2017</b> , 117, 75-81	10.2	14
68	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. <i>Journal of Medical Genetics</i> , <b>2019</b> , 56, 693-700	5.8	14
67	Early Diagnosis in Prader-Willi Syndrome Reduces Obesity and Associated Co-Morbidities. <i>Genes</i> , <b>2019</b> , 10,	4.2	14
66	Chest wall hamartoma with Wiedemann-Beckwith syndrome: clinical report and brief review of chromosome 11p15.5-related tumors. <i>American Journal of Medical Genetics Part A</i> , <b>2001</b> , 101, 221-5		13
65	Increased fertility in a woman with classic galactosaemia. <i>Journal of Inherited Metabolic Disease</i> , <b>2001</b> , 24, 607-8	5.4	13
64	Psychological Impact of Predictive Genetic Testing in VCP Inclusion Body Myopathy, Paget Disease of Bone and Frontotemporal Dementia. <i>Journal of Genetic Counseling</i> , <b>2015</b> , 24, 842-50	2.5	12
63	Impact of genetic subtypes of Prader-Willi syndrome with growth hormone therapy on intelligence and body mass index. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 1826-1835	2.5	12



62	Temtamy-like syndrome associated with translocation of 2p24 and 9q32. <i>Clinical Dysmorphology</i> , <b>2003</b> , 12, 175-7	0.9	12
61	Krabbe disease: severe neonatal presentation with a family history of multiple sclerosis. <i>Journal of Child Neurology</i> , <b>2005</b> , 20, 826-8	2.5	12
60	Radiological features of Paget disease of bone associated with VCP myopathy. <i>Skeletal Radiology</i> , <b>2012</b> , 41, 329-37	2.7	11
59	Splice-Break: exploiting an RNA-seq splice junction algorithm to discover mitochondrial DNA deletion breakpoints and analyses of psychiatric disorders. <i>Nucleic Acids Research</i> , <b>2019</b> , 47, e59	20.1	10
58	Peters anomaly in association with multiple midline anomalies and a familial chromosome 4 inversion. <i>Ophthalmic Genetics</i> , <b>2006</b> , 27, 63-5	1.2	10
57	Targeted excision of VCP R155H mutation by Cre-LoxP technology as a promising therapeutic strategy for valosin-containing protein disease. <i>Human Gene Therapy Methods</i> , <b>2015</b> , 26, 13-24	4.9	9
56	Myogenic differentiation of VCP disease-induced pluripotent stem cells: A novel platform for drug discovery. <i>PLoS ONE</i> , <b>2017</b> , 12, e0176919	3.7	9
55	Comparison of perinatal factors in deletion versus uniparental disomy in Prader-Willi syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 1161-1165	2.5	8
54	Clinical heterogeneity in autosomal dominant optic atrophy in two 3q28-qter linked central Illinois families. <i>Genetics in Medicine</i> , <b>2000</b> , 2, 283-9	8.1	8
53	The Myoblast C2C12 Transfected with Mutant Valosin-Containing Protein Exhibits Delayed Stress Granule Resolution on Oxidative Stress. <i>American Journal of Pathology</i> , <b>2016</b> , 186, 1623-34	5.8	8
52	Multisystem proteinopathy: Where myopathy and motor neuron disease converge. <i>Muscle and Nerve</i> , <b>2021</b> , 63, 442-454	3.4	8
51	What syndrome is this? Laryngo-onycho-cutaneous syndrome. <i>Pediatric Dermatology</i> , <b>2007</b> , 24, 306-8	1.9	7
50	Duplication of 5q15-q23.2: case report and literature review. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , <b>2006</b> , 76, 272-6		7
49	Variable clinical features and genotype-phenotype correlations in 18 patients with late-onset Pompe disease. <i>Annals of Translational Medicine</i> , <b>2019</b> , 7, 276	3.2	7
48	Pathogenic mutations in NUBPL affect complex I activity and cold tolerance in the yeast model <i>Yarrowia lipolytica</i> . <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 3697-3709	5.6	7
47	Antisense oligonucleotide treatment targeting glycogen synthase (GYS1) in a mouse model of Pompe disease. <i>Molecular Genetics and Metabolism</i> , <b>2019</b> , 126, S85	3.7	5
46	A case report comparing clinical, imaging and neuropsychological assessment findings in twins discordant for the VCP p.R155C mutation. <i>Neuromuscular Disorders</i> , <b>2015</b> , 25, 177-83	2.9	5
45	Possible new autosomal recessive syndrome of partial agenesis of the corpus callosum, pontine hypoplasia, focal white matter changes, hypotonia, mental retardation, and minor anomalies. <i>American Journal of Medical Genetics Part A</i> , <b>1997</b> , 73, 184-188		5

44	Newborn screening for Prader-Willi syndrome is feasible: Early diagnosis for better outcomes. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 29-36	2.5	5
43	Homozygosity for the A431E mutation in PSEN1 presenting with a relatively aggressive phenotype. <i>Neuroscience Letters</i> , <b>2019</b> , 699, 195-198	3.3	4
42	Mapping autosomal dominant progressive limb-girdle myopathy with bone fragility to chromosome 9p21-p22: a novel locus for a musculoskeletal syndrome. <i>Human Genetics</i> , <b>2005</b> , 118, 508-14	6.3	4
41	Development of a standard of care for patients with valosin-containing protein associated multisystem proteinopathy.. <i>Orphanet Journal of Rare Diseases</i> , <b>2022</b> , 17, 23	4.2	4
40	Response to Growth Hormone Treatment in a Patient with Insulin-Like Growth Factor 1 Receptor Deletion. <i>JCRPE Journal of Clinical Research in Pediatric Endocrinology</i> , <b>2017</b> , 9, 380-386	1.9	4
39	Genetic Subtype-Phenotype Analysis of Growth Hormone Treatment on Psychiatric Behavior in Prader-Willi Syndrome. <i>Genes</i> , <b>2020</b> , 11,	4.2	4
38	A p97/Valosin-Containing Protein Inhibitor Drug CB-5083 Has a Potent but Reversible Off-Target Effect on Phosphodiesterase-6. <i>Journal of Pharmacology and Experimental Therapeutics</i> , <b>2021</b> , 378, 31-44	4.7	4
37	ALG11-CDG syndrome: Expanding the phenotype. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 498-502	2.5	3
36	A cross-sectional analysis of clinical evaluation in 35 individuals with mutations of the valosin-containing protein gene. <i>Neuromuscular Disorders</i> , <b>2018</b> , 28, 778-786	2.9	3
35	Phenotypic diversity of patients diagnosed with VACTERL association. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 1830-1837	2.5	3
34	Subglossopalatal synechia in association with cardiac and digital anomalies. <i>Cleft Palate-Craniofacial Journal</i> , <b>2008</b> , 45, 217-21	1.9	3
33	Recurrent miscarriage in a carrier of a balanced cytogenetically undetectable subtelomeric rearrangement: how many are we missing?. <i>Prenatal Diagnosis</i> , <b>2006</b> , 26, 291-3	3.2	3
32	A Fine Balance of Dietary Lipids Improves Pathology of a Murine Model of VCP-Associated Multisystem Proteinopathy. <i>PLoS ONE</i> , <b>2015</b> , 10, e0131995	3.7	3
31	Molecular subtype and growth hormone effects on dysmorphology in Prader-Willi syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 169-175	2.5	3
30	Influence of molecular classes and growth hormone treatment on growth and dysmorphology in Prader-Willi syndrome: A multicenter study. <i>Clinical Genetics</i> , <b>2021</b> , 100, 29-39	4	3
29	Prader-Willi Syndrome due to an Unbalanced de novo Translocation t(15;19)(q12;p13.3). <i>Cytogenetic and Genome Research</i> , <b>2016</b> , 150, 29-34	1.9	3
28	Characteristics of VCP mutation-associated cardiomyopathy. <i>Neuromuscular Disorders</i> , <b>2021</b> , 31, 701-705	2.9	3
27	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. <i>Pediatric Neurology</i> , <b>2021</b> , 123, 30-37	2.9	3



26	Two cases of Legg-Perthes and intellectual disability in Tricho-Rhino-Phalangeal syndrome type 1 associated with novel TRPS1 mutations. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 1663-1667	2.5	2
25	Birth seasonality studies in a large Prader-Willi syndrome cohort. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 1531-1534	2.5	2
24	A novel mutation of orthodenticle homeobox 2 contributing to a case of otocephaly initially diagnosed by prenatal ultrasound in the first trimester. <i>Clinical Dysmorphology</i> , <b>2017</b> , 26, 98-100	0.9	2
23	Smith-Lemli-Opitz syndrome in trisomy 13: how does the mix work?. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , <b>2005</b> , 73, 569-71		2
22	Ceramide contributes to pathogenesis and may be targeted for therapy in VCP inclusion body myopathy. <i>Human Molecular Genetics</i> , <b>2021</b> , 29, 3945-3953	5.6	2
21	Homozygous B4GALNT1 mutation and biochemical glutaric acidemia type II: A case report. <i>Clinical Neurology and Neurosurgery</i> , <b>2020</b> , 189, 105553	2	2
20	Stroke and Chronic Kidney Disease in Fabry Disease. <i>Journal of Stroke and Cerebrovascular Diseases</i> , <b>2021</b> , 30, 105423	2.8	2
19	mitochondrial disease: new patients and review of the genetic and clinical spectrum. <i>Journal of Medical Genetics</i> , <b>2021</b> , 58, 314-325	5.8	2
18	Ataxia and Parkinsonism in a Woman With a VCP Variant and Long-Normal Repeats in the SCA2 Allele. <i>Neurology: Genetics</i> , <b>2021</b> , 7, e595	3.8	2
17	Multisystem Proteinopathy Due to VCP Mutations: A Review of Clinical Heterogeneity and Genetic Diagnosis. <i>Genes</i> , <b>2022</b> , 13, 963	4.2	2
16	A placebo-controlled trial of folic acid and betaine in identical twins with Angelman syndrome. <i>Orphanet Journal of Rare Diseases</i> , <b>2019</b> , 14, 232	4.2	1
15	Humoral immune deficiency and hemifacial microsomia seen in one family. <i>Cleft Palate-Craniofacial Journal</i> , <b>2009</b> , 46, 477-80	1.9	1
14	Prevalence of cerebral small vessel disease in a Fabry disease cohort. <i>Molecular Genetics and Metabolism Reports</i> , <b>2021</b> , 29, 100815	1.8	1
13	Variable clinical features of patients with Fabry disease and outcome of enzyme replacement therapy. <i>Molecular Genetics and Metabolism Reports</i> , <b>2021</b> , 26, 100700	1.8	1
12	Mosaic de novo gene variant associated with Prader-Willi syndrome. <i>Journal of Medical Genetics</i> , <b>2021</b> ,	5.8	1
11	Severe cardiomyopathy associated with the VCP p.R155C and c.177_187del MYBPC3 gene variants.. <i>European Journal of Medical Genetics</i> , <b>2022</b> , 104480	2.6	1
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