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	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
2	Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. Nature, 2013, 495, 467-473.	27.8	1,249
3	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
4	Nutritional phases in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1040-1049.	1.2	325
5	Molecular mechanism for duplication 17p11.2â€" the homologous recombination reciprocal of the Smith-Magenis microdeletion. Nature Genetics, 2000, 24, 84-87.	21.4	297
6	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
7	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
8	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
9	Novel Ubiquitin Neuropathology in Frontotemporal Dementia WithValosin-Containing ProteinGene Mutations. Journal of Neuropathology and Experimental Neurology, 2006, 65, 571-581.	1.7	206
10	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
11	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
12	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
13	Radiological features in 82 patients with nevoid basal cell carcinoma (NBCC or Gorlin) syndrome. Genetics in Medicine, 2004, 6, 495-502.	2.4	161
14	Mutant valosinâ€containing protein causes a novel type of frontotemporal dementia. Annals of Neurology, 2005, 57, 457-461.	5.3	160
15	Genetics of Craniosynostosis. Seminars in Pediatric Neurology, 2007, 14, 150-161.	2.0	155
16	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
17	Pathological consequences of VCP mutations on human striated muscle. Brain, 2007, 130, 381-393.	7.6	148
18	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

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19	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
20	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
21	Prospective phenotyping of NGLY1-CDDG, the first congenital disorder of deglycosylation. Genetics in Medicine, 2017, 19, 160-168.	2.4	124
22	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
23	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
24	Molecular genetic classification in Prader-Willi syndrome: a multisite cohort study. Journal of Medical Genetics, 2019, 56, 149-153.	3.2	112
25	Molecular analysis expands the spectrum of phenotypes associated with GLI3 mutations. Human Mutation, 2010, 31, 1142-1154.	2.5	111
26	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
27	Genotypeâ€phenotype study in patients with valosinâ€containing protein mutations associated with multisystem proteinopathy. Clinical Genetics, 2018, 93, 119-125.	2.0	100
28	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
29	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
30	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
31	Clinical geneticists' views of VACTERL/VATER association. American Journal of Medical Genetics, Part A, 2012, 158A, 3087-3100.	1.2	78
32	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
33	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
34	Familial incidence and associated symptoms in a population of individuals with nonsyndromic craniosynostosis. Genetics in Medicine, 2014, 16, 302-310.	2.4	70
35	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
36	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

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37	Mitochondrial dysfunction in CA1 hippocampal neurons of the UBE3A deficient mouse model for Angelman syndrome. Neuroscience Letters, 2011, 487, 129-133.	2.1	65
38	Genetic heterogeneity in autosomal dominant essential tremor. Genetics in Medicine, 2001, 3, 197-199.	2.4	62
39	Valosin containing protein associated inclusion body myopathy: abnormal vacuolization, autophagy and cell fusion in myoblasts. Neuromuscular Disorders, 2009, 19, 766-772.	0.6	59
40	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
41	Growth Charts for Non-Growth Hormone Treated Prader-Willi Syndrome. Pediatrics, 2015, 135, e126-e135.	2.1	56
42	The Homozygote VCPR155H/R155H Mouse Model Exhibits Accelerated Human VCP-Associated Disease Pathology. PLoS ONE, 2012, 7, e46308.	2.5	56
43	Growth Standards of Infants With Prader-Willi Syndrome. Pediatrics, 2011, 127, 687-695.	2.1	53
44	Valosin-containing protein mutation and Parkinsonâ€™s disease. Parkinsonism and Related Disorders, 2012, 18, 107-109.	2.2	53
45	Autosomal Dominant Inclusion Body Myopathy, Paget Disease of Bone, and Frontotemporal Dementia. Alzheimer Disease and Associated Disorders, 2005, 19, S44-S47.	1.3	51
46	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
47	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
48	Contributing factors of mortality in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 196-205.	1.2	50
49	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
50	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
51	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
52	Is gestation in Prader-Willi syndrome affected by the genetic subtype?. Journal of Assisted Reproduction and Genetics, 2009, 26, 461-466.	2.5	43
53	Clinical and radiological features in young individuals with nevoid basal cell carcinoma syndrome. Genetics in Medicine, 2013, 15, 79-83.	2.4	43
54	Identification of 34 novel and 56 knownFOXL2mutations in patients with blepharophimosis syndrome. Human Mutation, 2008, 29, E205-E219.	2.5	42

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55	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
56	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
57	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
58	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
59	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
60	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
61	Clinical characterization of int22h1/int22h2-mediated Xq28 duplication/deletion: new cases and literature review. BMC Medical Genetics, 2015, 16, 12.	2.1	37
62	Growth Charts for Prader-Willi Syndrome During Growth Hormone Treatment. Clinical Pediatrics, 2016, 55, 957-974.	0.8	37
63	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
64	Gain-of-Function Mutations inRARBCause Intellectual Disability with Progressive Motor Impairment. Human Mutation, 2016, 37, 786-793.	2.5	34
65	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
66	Multisystem proteinopathy: Where myopathy and motor neuron disease converge. Muscle and Nerve, 2021, 63, 442-454.	2.2	33
67	Activation of the NLRP3 Inflammasome Is Associated with Valosin-Containing Protein Myopathy. Inflammation, 2017, 40, 21-41.	3.8	32
68	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
69	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
70	Meta-analysis of genotype-phenotype analysis of OPA1 mutations in autosomal dominant optic atrophy. Mitochondrion, 2019, 46, 262-269.	3.4	29
71	ALX4 gain-of-function mutations in nonsyndromic craniosynostosis. Human Mutation, 2012, 33, 1626-1629.	2.5	28
72	Multisystem Proteinopathy Due to VCP Mutations: A Review of Clinical Heterogeneity and Genetic Diagnosis. Genes, 2022, 13, 963.	2.4	28

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73	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
74	Apert syndrome: what prenatal radiographic findings should prompt its consideration?. Prenatal Diagnosis, 2006, 26, 966-972.	2.3	25
75	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
76	Multicentre study of maternal and neonatal outcomes in individuals with Prader-Willi syndrome. Journal of Medical Genetics, 2018, 55, 594-598.	3.2	25
77	Early Diagnosis in Praderâ€“Willi Syndrome Reduces Obesity and Associated Co-Morbidities. Genes, 2019, 10, 898.	2.4	25
78	New family with HSPB 8-associated autosomal dominant rimmed vacuolar myopathy. Neurology: Genetics, 2019, 5, e349.	1.9	24
79	Exercise Training Reverses Skeletal Muscle Atrophy in an Experimental Model of VCP Disease. PLoS ONE, 2013, 8, e76187.	2.5	24
80	Genetics of hearing loss: where are we standing now?. European Archives of Oto-Rhino-Laryngology, 2012, 269, 1733-1745.	1.6	23
81	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
82	In vitro studies in VCP-associated multisystem proteinopathy suggest altered mitochondrial bioenergetics. Mitochondrion, 2015, 22, 1-8.	3.4	22
83	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
84	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
85	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. Pediatric Neurology, 2021, 123, 30-37.	2.1	21
86	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
87	Frequency of Praderâ€“Willi syndrome in births conceived via assisted reproductive technology. Genetics in Medicine, 2014, 16, 164-169.	2.4	20
88	Novel valosin-containing protein mutations associated with multisystem proteinopathy. Neuromuscular Disorders, 2018, 28, 491-501.	0.6	20
89	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
90	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

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91	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
92	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
93	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
94	Dysfunctional oleoylethanolamide signaling in a mouse model of Prader-Willi syndrome. Pharmacological Research, 2017, 117, 75-81.	7.1	16
95	Radiological features of Paget disease of bone associated with VCP myopathy. Skeletal Radiology, 2012, 41, 329-337.	2.0	15
96	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
97	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
98	Increased fertility in a woman with classic galactosaemia. Journal of Inherited Metabolic Disease, 2001, 24, 507-608.	3.6	14
99	Temtamy-like syndrome associated with translocation of 2p24 and 9q32. Clinical Dysmorphology, 2003, 12, 175-177.	0.3	14
100	Peters Anomaly in Association with Multiple Midline Anomalies and a Familial Chromosome 4 Inversion. Ophthalmic Genetics, 2006, 27, 63-65.	1.2	14
101	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
102	Krabbe Disease: Severe Neonatal Presentation With a Family History of Multiple Sclerosis. Journal of Child Neurology, 2005, 20, 826-828.	1.4	13
103	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
104	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
105	Myogenic differentiation of VCP disease-induced pluripotent stem cells: A novel platform for drug discovery. PLoS ONE, 2017, 12, e0176919.	2.5	10
106	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
107	What Syndrome Is This?. Pediatric Dermatology, 2007, 24, 306-308.	0.9	9
108	Genetic Subtype-Phenotype Analysis of Growth Hormone Treatment on Psychiatric Behavior in Prader-Willi Syndrome. Genes, 2020, 11, 1250.	2.4	9

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109	NUBPL mitochondrial disease: new patients and review of the genetic and clinical spectrum. Journal of Medical Genetics, 2021, 58, 314-325.	3.2	9
110	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
111	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
112	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
113	ALG11â€CDG syndrome: Expanding the phenotype. American Journal of Medical Genetics, Part A, 2019, 179, 498-502.	1.2	8
114	Homozygosity for the A431E mutation in PSEN1 presenting with a relatively aggressive phenotype. Neuroscience Letters, 2019, 699, 195-198.	2.1	8
115	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
116	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
117	Characteristics of VCP mutation-associated cardiomyopathy. Neuromuscular Disorders, 2021, 31, 701-705.	0.6	8
118	Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918.	2.5	8
119	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
120	Phenotypic diversity of patients diagnosed with VACTERL association. American Journal of Medical Genetics, Part A, 2018, 176, 1830-1837.	1.2	7
121	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
122	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
123	Subglossopalatal Synechia in Association with Cardiac and Digital Anomalies. Cleft Palate-Craniofacial Journal, 2008, 45, 217-221.	0.9	6
124	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
125	Mosaic de novo SNRPN gene variant associated with Prader-Willi syndrome. Journal of Medical Genetics, 2022, 59, 719-722.	3.2	6
126	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

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127	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
128	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
129	Antisense oligonucleotide treatment targeting glycogen synthase (GYS1) in a mouse model of Pompe disease. Molecular Genetics and Metabolism, 2019, 126, S85.	1.1	5
130	Prevalence of cerebral small vessel disease in a Fabry disease cohort. Molecular Genetics and Metabolism Reports, 2021, 29, 100815.	1.1	5
131	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
132	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
133	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
134	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
135	Stroke and Chronic Kidney Disease in Fabry Disease. Journal of Stroke and Cerebrovascular Diseases, 2021, 30, 105423.	1.6	4
136	Diffuse large B-cell non-Hodgkin's lymphoma in Gaucher disease. Molecular Genetics and Metabolism Reports, 2020, 25, 100663.	1.1	4
137	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
138	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
139	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
140	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
141	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
142	Homozygous B4GALNT1 mutation and biochemical glutaric acidemia type II: A case report. Clinical Neurology and Neurosurgery, 2020, 189, 105553.	1.4	3
143	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
144	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

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145	Humoral Immune Deficiency and Hemifacial Microsomia Seen in One Family. Cleft Palate-Craniofacial Journal, 2009, 46, 477-480.	0.9	2
146	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
147	Birth seasonality studies in a large Praderâ€“Willi syndrome cohort. American Journal of Medical Genetics, Part A, 2019, 179, 1531-1534.	1.2	2
148	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
149	Regional Strain Pattern and Correlation with Cardiac Magnetic Resonance Imaging in Fabry Disease.. Journal of Cardiovascular Echography, 2021, 31, 131-136.	0.4	2
150	A clinicopathologic study of malignancy in VCP-associated multisystem proteinopathy. Orphanet Journal of Rare Diseases, 2022, 17, .	2.7	2
151	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
152	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
153	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
154	Dysmorphology of inborn errors of metabolism. Molecular Cytogenetics, 2014, 7, I39.	0.9	0
155	Clinical utility and dilemmas of SNP microarray testing. Molecular Cytogenetics, 2014, 7, I34.	0.9	0
156	Cover Image, Volume 179A, Number 3, March 2019. American Journal of Medical Genetics, Part A, 2019, 179, i.	1.2	0
157	Front Cover, Volume 40, Issue 11. Human Mutation, 2019, 40, i.	2.5	0
158	Expression level of R155H mRNA in the knock-in mouse model. Biochemical and Biophysical Research Communications, 2020, 523, 985-986.	2.1	0
159	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