

Recessive mutations in EPG5 cause Vici syndrome, a m autophagy

Nature Genetics

45, 83-87

DOI: [10.1038/ng.2497](https://doi.org/10.1038/ng.2497)

Citation Report

#	ARTICLE	IF	CITATIONS
1	Protein breakdown in muscle wasting: Role of autophagy-lysosome and ubiquitin-proteasome. International Journal of Biochemistry and Cell Biology, 2013, 45, 2121-2129.	1.2	508
2	Misregulation of autophagy and protein degradation systems in myopathies and muscular dystrophies. Journal of Cell Science, 2013, 126, 5325-5333.	1.2	160
3	Autophagy in infection, inflammation and immunity. Nature Reviews Immunology, 2013, 13, 722-737.	10.6	1,571
4	Enrichment of FLI1 and RUNX1 mutations in families with excessive bleeding and platelet dense granule secretion defects. Blood, 2013, 122, 4090-4093.	0.6	108
5	BPAN. International Review of Neurobiology, 2013, 110, 85-90.	0.9	41
6	Deletion of Autophagy-related 5 (Atg5) and Pik3c3 Genes in the Lens Causes Cataract Independent of Programmed Organelle Degradation. Journal of Biological Chemistry, 2013, 288, 11436-11447.	1.6	119
7	Role of <i>Epg5</i> in selective neurodegeneration and Vici syndrome. Autophagy, 2013, 9, 1258-1262.	4.3	47
8	Zebrafish <i>ambra1a</i> and <i>ambra1b</i> Knockdown Impairs Skeletal Muscle Development. PLoS ONE, 2014, 9, e99210.	1.1	36
9	Integrative Data Mining Highlights Candidate Genes for Monogenic Myopathies. PLoS ONE, 2014, 9, e110888.	1.1	16
10	Pathogenic Mechanisms in Centronuclear Myopathies. Frontiers in Aging Neuroscience, 2014, 6, 339.	1.7	89
12	Clinical utility gene card for: Vici Syndrome. European Journal of Human Genetics, 2014, 22, 435-435.	1.4	13
13	Genetic Syndromes with Evidence of Immune Deficiency. , 2014, , 281-324.		3
14	Autophagy and Its Normal and Pathogenic States in the Brain. Annual Review of Neuroscience, 2014, 37, 55-78.	5.0	165
15	Emerging role of autophagy in pediatric neurodegenerative and neurometabolic diseases. Pediatric Research, 2014, 75, 217-226.	1.1	42
16	At the end of the autophagic road: an emerging understanding of lysosomal functions in autophagy. Trends in Biochemical Sciences, 2014, 39, 61-71.	3.7	295
17	Skeletal muscle, autophagy, and physical activity: the <i>m</i> Ã©nage Ã trois of metabolic regulation in health and disease. Journal of Molecular Medicine, 2014, 92, 127-137.	1.7	78
18	Exercise-Induced Skeletal Muscle Remodeling and Metabolic Adaptation: Redox Signaling and Role of Autophagy. Antioxidants and Redox Signaling, 2014, 21, 154-176.	2.5	157
19	Autophagy and human diseases. Cell Research, 2014, 24, 69-79.	5.7	708

#	ARTICLE	IF	CITATIONS
21	Autophagy and human disease: emerging themes. <i>Current Opinion in Genetics and Development</i> , 2014, 26, 16-23.	1.5	280
22	Pathological changes in cardiac muscle and cerebellar cortex in Vici syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 3203-3205.	0.7	13
23	First description of a patient with Vici syndrome due to a mutation affecting the penultimate exon of <i>EPG5</i> and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 3170-3175.	0.7	33
24	Autophagy – a fundamental cellular mechanism on the verge of clinical translation. <i>Neuropathology and Applied Neurobiology</i> , 2015, 41, 598-600.	1.8	1
25	Immunologic manifestations of autophagy. <i>Journal of Clinical Investigation</i> , 2015, 125, 75-84.	3.9	135
26	Congenital myopathies. <i>Neurology</i> , 2015, 84, 28-35.	1.5	106
27	Cardiac Cytoarchitecture. , 2015, , .		4
28	Autophagy and neurodegeneration. <i>Journal of Clinical Investigation</i> , 2015, 125, 65-74.	3.9	288
29	SIL1-related Marinesco-Sjogren syndrome (MSS) with associated motor neuronopathy and bradykinetic movement disorder. <i>Neuromuscular Disorders</i> , 2015, 25, 585-588.	0.3	14
30	Defective autophagy through <i>epg5</i> mutation results in failure to reduce germ plasm and mitochondria. <i>FASEB Journal</i> , 2015, 29, 4145-4161.	0.2	29
31	Eaten alive: novel insights into autophagy from multicellular model systems. <i>Trends in Cell Biology</i> , 2015, 25, 376-387.	3.6	92
32	Biallelic mutations in <i>SNX14</i> cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. <i>Nature Genetics</i> , 2015, 47, 528-534.	9.4	111
33	Severe Central Sleep Apnea in Vici Syndrome. <i>Pediatrics</i> , 2015, 136, e1390-e1394.	1.0	9
34	Protecting the mitochondrial powerhouse. <i>Trends in Cell Biology</i> , 2015, 25, 158-170.	3.6	260
35	Autophagy in Ocular Pathophysiology. , 0, , .		1
37	Autophagy linked FYVE (Alfy/WDFY3) is required for establishing neuronal connectivity in the mammalian brain. <i>ELife</i> , 2016, 5, .	2.8	78
38	mTOR, Autophagy, Aminoacidopathies, and Human Genetic Disorders. , 2016, , 143-166.		1
39	Non-Canonical Cell Death Induced by p53. <i>International Journal of Molecular Sciences</i> , 2016, 17, 2068.	1.8	121

#	ARTICLE	IF	CITATIONS
40	Aging-Related Diseases and Autophagy. , 2016, , .		0
41	KIF1A/UNC-104 Transports ATG-9 to Regulate Neurodevelopment and Autophagy at Synapses. <i>Developmental Cell</i> , 2016, 38, 171-185.	3.1	165
42	Bixin protects mice against ventilation-induced lung injury in an NRF2-dependent manner. <i>Scientific Reports</i> , 2016, 6, 18760.	1.6	58
43	The beneficial role of proteolysis in skeletal muscle growth and stress adaptation. <i>Skeletal Muscle</i> , 2016, 6, 16.	1.9	88
44	Non-parametric Survival Analysis of EPG5 Gene with Age at Onset of Alzheimer's Disease. <i>Journal of Molecular Neuroscience</i> , 2016, 60, 436-444.	1.1	8
45	Crosstalk between autophagy and inflammatory signalling pathways: balancing defence and homeostasis. <i>Nature Reviews Immunology</i> , 2016, 16, 661-675.	10.6	341
46	Mice deficient in the Vici syndrome gene <i>Epg5</i> exhibit features of retinitis pigmentosa. <i>Autophagy</i> , 2016, 12, 2263-2270.	4.3	19
47	TBC1D20 mediates autophagy as a key regulator of autophagosome maturation. <i>Autophagy</i> , 2016, 12, 1759-1775.	4.3	56
48	Disrupted autophagy undermines skeletal muscle adaptation and integrity. <i>Mammalian Genome</i> , 2016, 27, 525-537.	1.0	29
49	Vici syndrome in siblings born to consanguineous parents. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 220-225.	0.7	16
50	Targeted Quantitative Screening of Chromosome 18 Encoded Proteome in Plasma Samples of Astronaut Candidates. <i>Journal of Proteome Research</i> , 2016, 15, 4039-4046.	1.8	33
51	The Vici Syndrome Protein EPG5 Is a Rab7 Effector that Determines the Fusion Specificity of Autophagosomes with Late Endosomes/Lysosomes. <i>Molecular Cell</i> , 2016, 63, 781-795.	4.5	227
54	Autophagy modulators from traditional Chinese medicine: Mechanisms and therapeutic potentials for cancer and neurodegenerative diseases. <i>Journal of Ethnopharmacology</i> , 2016, 194, 861-876.	2.0	68
55	Get the Balance Right Pathological Significance of Autophagy Perturbation in Neuromuscular Disorders. <i>Journal of Neuromuscular Diseases</i> , 2016, 3, 127-155.	1.1	35
56	Aberrant splicing induced by the most common <i>EPG5</i> mutation in an individual with Vici syndrome. <i>Brain</i> , 2016, 139, e52-e52.	3.7	14
57	Reply: Aberrant splicing induced by the most common <i>EPG5</i> mutation in an individual with Vici syndrome. <i>Brain</i> , 2016, 139, e53-e53.	3.7	4
58	Vici syndrome: a review. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 21.	1.2	55
59	Two cases of Vici syndrome associated with Idiopathic Thrombocytopenic Purpura (ITP) with a review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1343-1346.	0.7	9

#	ARTICLE	IF	CITATIONS
60	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). <i>Autophagy</i> , 2016, 12, 1-222.	4.3	4,701
61	The clearance of dying cells: table for two. <i>Cell Death and Differentiation</i> , 2016, 23, 915-926.	5.0	239
62	Homeostatic Control of Innate Lung Inflammation by Vici Syndrome Gene <i>Epg5</i> and Additional Autophagy Genes Promotes Influenza Pathogenesis. <i>Cell Host and Microbe</i> , 2016, 19, 102-113.	5.1	83
63	Autophagy Snuffs a Macrophage's Inner Fire. <i>Cell Host and Microbe</i> , 2016, 19, 9-11.	5.1	2
64	<i>EPG5</i> -related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. <i>Brain</i> , 2016, 139, 765-781.	3.7	99
65	Congenital disorders of autophagy: an emerging novel class of inborn errors of neuro-metabolism. <i>Brain</i> , 2016, 139, 317-337.	3.7	126
66	Autophagy in the lens. <i>Experimental Eye Research</i> , 2016, 144, 22-28.	1.2	50
67	Autophagosome Maturation and Fusion. <i>Journal of Molecular Biology</i> , 2017, 429, 486-496.	2.0	185
68	Prenatal and postnatal presentations of corpus callosum agenesis with polymicrogyria caused by <i>EPG5</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 706-711.	0.7	12
69	The exponential growth of autophagy-related research: from the humble yeast to the Nobel Prize. <i>FEBS Letters</i> , 2017, 591, 681-689.	1.3	33
70	Mutations in <i>INPP5K</i> , Encoding a Phosphoinositide 5-Phosphatase, Cause Congenital Muscular Dystrophy with Cataracts and Mild Cognitive Impairment. <i>American Journal of Human Genetics</i> , 2017, 100, 523-536.	2.6	67
71	Primary Immunodeficiency Diseases. , 2017, , .		22
72	Muscle pathology in Vici syndrome—A case study with a novel mutation in <i>EPG5</i> and a summary of the literature. <i>Neuromuscular Disorders</i> , 2017, 27, 771-776.	0.3	21
73	Defects in autophagosome-lysosome fusion underlie Vici syndrome, a neurodevelopmental disorder with multisystem involvement. <i>Scientific Reports</i> , 2017, 7, 3552.	1.6	46
74	Autophagy: an adaptive physiological countermeasure to cellular senescence and ischaemia/reperfusion-associated cardiac arrhythmias. <i>Journal of Cellular and Molecular Medicine</i> , 2017, 21, 1058-1072.	1.6	49
75	Activation of Autophagy Ameliorates Cardiomyopathy in <i>Mybpc3</i> -Targeted Knockin Mice. <i>Circulation: Heart Failure</i> , 2017, 10, .	1.6	53
76	Rapid Targeted Genomics in Critically Ill Newborns. <i>Pediatrics</i> , 2017, 140, .	1.0	99
77	Autopsy findings in <i>EPG5</i> -related Vici syndrome with antenatal onset. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2522-2527.	0.7	6

#	ARTICLE	IF	CITATIONS
78	Stall in Canonical Autophagy-Lysosome Pathways Prompts Nucleophagy-Based Nuclear Breakdown in Neurodegeneration. <i>Current Biology</i> , 2017, 27, 3626-3642.e6.	1.8	47
79	EPG5-Related Vici Syndrome: A Primary Defect of Autophagic Regulation with an Emerging Phenotype Overlapping with Mitochondrial Disorders. <i>JIMD Reports</i> , 2017, 42, 19-29.	0.7	7
80	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 232-245.	1.5	261
81	Monitoring and Measuring Autophagy. <i>International Journal of Molecular Sciences</i> , 2017, 18, 1865.	1.8	805
83	Genetic aberrations in macroautophagy genes leading to diseases. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2018, 1865, 803-816.	1.9	49
84	A brief history of autophagy from cell biology to physiology and disease. <i>Nature Cell Biology</i> , 2018, 20, 521-527.	4.6	518
85	Mechanism and medical implications of mammalian autophagy. <i>Nature Reviews Molecular Cell Biology</i> , 2018, 19, 349-364.	16.1	1,933
86	Dysregulated autophagy in restrictive cardiomyopathy due to Pro209Leu mutation in BAG3. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 388-399.	0.5	56
87	Low-level expression of EPG5 leads to an attenuated Vici syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1207-1211.	0.7	9
88	Genome-wide scan of depressive symptomatology in two representative cohorts in the United States and the United Kingdom. <i>Journal of Psychiatric Research</i> , 2018, 100, 63-70.	1.5	3
89	Autophagy balances inflammation in innate immunity. <i>Autophagy</i> , 2018, 14, 243-251.	4.3	393
90	Hereditary myopathy with early respiratory failure (HMERF): Still rare, but common enough. <i>Neuromuscular Disorders</i> , 2018, 28, 268-276.	0.3	25
91	Congenital Disorders of Autophagy: What a Pediatric Neurologist Should Know. <i>Neuropediatrics</i> , 2018, 49, 018-025.	0.3	15
92	Neurological Manifestations of Primary Immunodeficiency Diseases. <i>Clinical Pediatrics</i> , 2018, 57, 761-774.	0.4	14
93	The Vici syndrome protein EPG5 regulates intracellular nucleic acid trafficking linking autophagy to innate and adaptive immunity. <i>Autophagy</i> , 2018, 14, 22-37.	4.3	23
94	A rare mutation in the EPG5 gene causes Vici syndrome. <i>Clinical Dysmorphology</i> , 2018, 27, 145-147.	0.1	3
95	Validating the RedMIT/GFP-LC3 Mouse Model by Studying Mitophagy in Autosomal Dominant Optic Atrophy Due to the OPA1Q285STOP Mutation. <i>Frontiers in Cell and Developmental Biology</i> , 2018, 6, 103.	1.8	10
96	Mechanisms of the autophagosome-lysosome fusion step and its relation to non-alcoholic fatty liver disease. <i>Liver Research</i> , 2018, 2, 120-124.	0.5	7

#	ARTICLE	IF	CITATIONS
97	Novel compound heterozygous <i>EPC5</i> mutations consisted with a missense mutation and a microduplication in the exon 1 region identified in a Japanese patient with Vici syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2803-2807.	0.7	3
98	Cardiac Arrhythmias and Antiarrhythmic Drugs: An Autophagic Perspective. <i>Frontiers in Physiology</i> , 2018, 9, 127.	1.3	10
99	Effectiveness of whole exome sequencing in unsolved patients with a clinical suspicion of a mitochondrial disorder in Estonia. <i>Molecular Genetics and Metabolism Reports</i> , 2018, 15, 80-89.	0.4	37
100	Autophagy as a promoter of longevity: insights from model organisms. <i>Nature Reviews Molecular Cell Biology</i> , 2018, 19, 579-593.	16.1	513
101	Altered distribution of ATG9A and accumulation of axonal aggregates in neurons from a mouse model of AP-4 deficiency syndrome. <i>PLoS Genetics</i> , 2018, 14, e1007363.	1.5	85
102	Molecular Classification of Primary Immunodeficiencies of T Lymphocytes. <i>Advances in Immunology</i> , 2018, 138, 99-193.	1.1	9
103	A systematic view on E3 ligase Ring TRIMmers with a focus on cardiac function and disease. <i>Trends in Cardiovascular Medicine</i> , 2019, 29, 1-8.	2.3	20
104	C-myc/miR-150/EPC5 axis mediated dysfunction of autophagy promotes development of non-small cell lung cancer. <i>Theranostics</i> , 2019, 9, 5134-5148.	4.6	42
105	Autophagic Control of Skin Aging. <i>Frontiers in Cell and Developmental Biology</i> , 2019, 7, 143.	1.8	52
106	EPC5 Variants with Modest Functional Impact Result in an Ameliorated and Primarily Neurological Phenotype in a 3.5-Year-Old Patient with Vici Syndrome. <i>Neuropediatrics</i> , 2019, 50, 257-261.	0.3	5
107	A Comprehensive Review of Autophagy and Its Various Roles in Infectious, Non-Infectious, and Lifestyle Diseases: Current Knowledge and Prospects for Disease Prevention, Novel Drug Design, and Therapy. <i>Cells</i> , 2019, 8, 674.	1.8	154
108	Hijacking intracellular membranes to feed autophagosomal growth. <i>FEBS Letters</i> , 2019, 593, 3120-3134.	1.3	12
109	Biallelic DMXL2 mutations impair autophagy and cause Ohtahara syndrome with progressive course. <i>Brain</i> , 2019, 142, 3876-3891.	3.7	23
110	Vici Syndrome with a Novel Mutation in EPC5. <i>Indian Pediatrics</i> , 2019, 56, 603-605.	0.2	1
111	Core autophagy genes and human diseases. <i>Current Opinion in Cell Biology</i> , 2019, 61, 117-125.	2.6	44
112	The RBG-1/RBG-2 complex modulates autophagy activity by regulating lysosomal biogenesis and function. <i>Journal of Cell Science</i> , 2019, 132, .	1.2	8
113	High-resolution quantitative proteomics applied to the study of the specific protein signature in the sputum and saliva of active tuberculosis patients and their infected and uninfected contacts. <i>Journal of Proteomics</i> , 2019, 195, 41-52.	1.2	20
114	A genetic model of CEDNIK syndrome in zebrafish highlights the role of the SNARE protein Snap29 in neuromotor and epidermal development. <i>Scientific Reports</i> , 2019, 9, 1211.	1.6	19

#	ARTICLE	IF	CITATIONS
115	The <i>epg5</i> knockout zebrafish line: a model to study Vici syndrome. <i>Autophagy</i> , 2019, 15, 1438-1454.	4.3	16
116	Beta-propeller protein-associated neurodegeneration (BPAN) as a genetically simple model of multifaceted neuropathology resulting from defects in autophagy. <i>Reviews in the Neurosciences</i> , 2019, 30, 261-277.	1.4	18
117	Autophagosome maturation: An epic journey from the ER to lysosomes. <i>Journal of Cell Biology</i> , 2019, 218, 757-770.	2.3	236
118	Methods to Determine the Role of Autophagy Proteins in <i>C. elegans</i> Aging. <i>Methods in Molecular Biology</i> , 2019, 1880, 561-586.	0.4	1
119	Biological Functions of Autophagy Genes: A Disease Perspective. <i>Cell</i> , 2019, 176, 11-42.	13.5	1,721
120	Autophagic dysfunction in Alzheimer's disease: Cellular and molecular mechanistic approaches to halt Alzheimer's pathogenesis. <i>Journal of Cellular Physiology</i> , 2019, 234, 8094-8112.	2.0	111
121	Autophagy in mammalian neurodevelopment and implications for childhood neurological disorders. <i>Neuroscience Letters</i> , 2019, 697, 29-33.	1.0	13
122	<i>EPG5</i> c.1007A>G mutation in a sibling pair with rapidly progressing Vici syndrome. <i>Annals of Human Genetics</i> , 2020, 84, 80-86.	0.3	6
123	Role of <i>Wdr45b</i> in maintaining neural autophagy and cognitive function. <i>Autophagy</i> , 2020, 16, 615-625.	4.3	41
124	Novel insights into the clinical and molecular spectrum of congenital disorders of autophagy. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 51-62.	1.7	31
125	Comprehensive autophagy evaluation in cardiac disease models. <i>Cardiovascular Research</i> , 2020, 116, 483-504.	1.8	41
126	Autophagosome-Lysosome Fusion. <i>Journal of Molecular Biology</i> , 2020, 432, 2462-2482.	2.0	184
127	The Consequences of Abnormal Gene Dosage: Lessons from Chromosome 18. <i>Trends in Genetics</i> , 2020, 36, 764-776.	2.9	8
128	A <i>tecpr2</i> knockout mouse exhibits age-dependent neuroaxonal dystrophy associated with autophagosome accumulation. <i>Autophagy</i> , 2021, 17, 3082-3095.	4.3	18
129	Lysosomal targeting of autophagosomes by the TECPR domain of TECPR2. <i>Autophagy</i> , 2021, 17, 3096-3108.	4.3	20
130	A high-throughput screening identifies ZNF418 as a novel regulator of the ubiquitin-proteasome system and autophagy-lysosomal pathway. <i>Autophagy</i> , 2021, 17, 3124-3139.	4.3	12
131	Vici syndrome with pathogenic homozygous <i>EPG5</i> gene mutation. <i>Medicine (United States)</i> , 2020, 99, e22302.	0.4	8
132	Autophagic cell death in viral infection: Do TAM receptors play a role?. <i>International Review of Cell and Molecular Biology</i> , 2020, 357, 123-168.	1.6	3

#	ARTICLE	IF	CITATIONS
133	Autophagy in Neuronal Development and Plasticity. Trends in Neurosciences, 2020, 43, 767-779.	4.2	50
134	A new homozygous HERC1 gain-of-function variant in MDFPMR syndrome leads to mTORC1 hyperactivation and reduced autophagy during cell catabolism. Molecular Genetics and Metabolism, 2020, 131, 126-134.	0.5	6
135	Autophagy in the Regulation of Tissue Differentiation and Homeostasis. Frontiers in Cell and Developmental Biology, 2020, 8, 602901.	1.8	29
136	Cellular Protein Quality Control in Diabetic Cardiomyopathy: From Bench to Bedside. Frontiers in Cardiovascular Medicine, 2020, 7, 585309.	1.1	11
137	Mendelian neurodegenerative disease genes involved in autophagy. Cell Discovery, 2020, 6, 24.	3.1	33
138	Autophagosome biogenesis and human health. Cell Discovery, 2020, 6, 33.	3.1	66
139	Tonic NMDA receptor signalling shapes endosomal organisation in mammalian cells. Scientific Reports, 2020, 10, 9315.	1.6	3
140	Common presentations and diagnostic approaches. , 2020, , 3-59.		1
141	Syndromes with less prominent immunologic dysfunction. , 2020, , 269-280.		0
142	Autophagy in Rare (NonLysosomal) Neurodegenerative Diseases. Journal of Molecular Biology, 2020, 432, 2735-2753.	2.0	23
143	Alterations of Brain Quantitative Proteomics Profiling Revealed the Molecular Mechanisms of Diosgenin against Cerebral Ischemia Reperfusion Effects. Journal of Proteome Research, 2020, 19, 1154-1168.	1.8	14
144	MitophAging: Mitophagy in Aging and Disease. Frontiers in Cell and Developmental Biology, 2020, 8, 239.	1.8	87
146	Rapamycin relieves the cataract caused by ablation of Gja8b through stimulating autophagy in zebrafish. Autophagy, 2021, 17, 3323-3337.	4.3	38
147	A new UHPLC-MS/MS method for the screening of urinary oligosaccharides expands the detection of storage disorders. Orphanet Journal of Rare Diseases, 2021, 16, 24.	1.2	7
148	Molecular Basis of Neuronal Autophagy in Ageing: Insights from Caenorhabditis elegans. Cells, 2021, 10, 694.	1.8	10
149	TDRD7 participates in lens development and spermiogenesis by mediating autophagosome maturation. Autophagy, 2021, 17, 3848-3864.	4.3	19
150	Primary immunodeficiency associated with hypopigmentation: A differential diagnosis approach. Allergologia Et Immunopathologia, 2021, 49, 178-190.	1.0	4
151	Co-occurrence of orofacial clefts and clubfoot phenotypes in a sub-Saharan African cohort: Whole-exome sequencing implicates multiple syndromes and genes. Molecular Genetics & Genomic Medicine, 2021, 9, e1655.	0.6	3

#	ARTICLE	IF	CITATIONS
152	Insights on autophagosome-lysosome tethering from structural and biochemical characterization of human autophagy factor EPG5. <i>Communications Biology</i> , 2021, 4, 291.	2.0	12
153	Melatonin induces the rejuvenation of long-term ex vivo expanded periodontal ligament stem cells by modulating the autophagic process. <i>Stem Cell Research and Therapy</i> , 2021, 12, 254.	2.4	26
154	Moments in autophagy and disease: Past and present. <i>Molecular Aspects of Medicine</i> , 2021, 82, 100966.	2.7	22
155	The Rab7 effector WDR91 promotes autophagy-lysosome degradation in neurons by regulating lysosome fusion. <i>Journal of Cell Biology</i> , 2021, 220, .	2.3	29
157	Machinery, regulation and pathophysiological implications of autophagosome maturation. <i>Nature Reviews Molecular Cell Biology</i> , 2021, 22, 733-750.	16.1	223
158	Molecular and cellular basis of genetically inherited skeletal muscle disorders. <i>Nature Reviews Molecular Cell Biology</i> , 2021, 22, 713-732.	16.1	55
159	Towards a better understanding of the neuro-developmental role of autophagy in sickness and in health. <i>Cell Stress</i> , 2021, 5, 99-118.	1.4	13
160	Ophthalmic findings as clues for early diagnosis of Vici syndrome in a neonate. <i>Ophthalmic Genetics</i> , 2021, 42, 780-783.	0.5	0
161	Autophagy in vascular dementia and natural products with autophagy regulating activity. <i>Pharmacological Research</i> , 2021, 170, 105756.	3.1	13
162	Compromised mitochondrial quality control triggers lipin1-related rhabdomyolysis. <i>Cell Reports Medicine</i> , 2021, 2, 100370.	3.3	11
163	Autophagy in axonal and presynaptic development. <i>Current Opinion in Neurobiology</i> , 2021, 69, 139-148.	2.0	10
164	The spectrum of neurodevelopmental, neuromuscular and neurodegenerative disorders due to defective autophagy. <i>Autophagy</i> , 2022, 18, 496-517.	4.3	18
165	Molecular targets and approaches to restore autophagy and lysosomal capacity in neurodegenerative disorders. <i>Molecular Aspects of Medicine</i> , 2021, 82, 101018.	2.7	8
166	Intestinal antiviral signaling is controlled by autophagy gene <i>Epg5</i> independent of the microbiota. <i>Autophagy</i> , 2022, 18, 1062-1077.	4.3	6
167	The first Chinese case of Vici syndrome with novel compound heterozygous sequence variants in <i>EPG5</i> . <i>International Journal of Developmental Neuroscience</i> , 2021, 81, 706-716.	0.7	0
168	Guidelines for the use and interpretation of assays for monitoring autophagy (4th) Tj ETQq1 1 0.784314 rgBT /Overlock 10 Tf 50,142 1,430	4.3	1,430
169	Syndromic Immunodeficiencies. , 2017, , 519-551.		2
170	Genetic Disorders of Immune Regulation. , 2017, , 295-338.		4

#	ARTICLE	IF	CITATIONS
171	Common Presentations and Diagnostic Approaches. , 2014, , 3-59.		2
172	Genetic defects of autophagy linked to disease. Progress in Molecular Biology and Translational Science, 2020, 172, 293-323.	0.9	10
174	Vici syndrome in an Egyptian infant: case report and differential diagnosis of inherited hypopigmented disorders. Egyptian Journal of Medical Human Genetics, 2020, 21, .	0.5	1
175	Identification of Atg2 and ArfGAP1 as Candidate Genetic Modifiers of the Eye Pigmentation Phenotype of Adaptor Protein-3 (AP-3) Mutants in Drosophila melanogaster. PLoS ONE, 2015, 10, e0143026.	1.1	8
176	Lack of collagen VI promotes neurodegeneration by impairing autophagy and inducing apoptosis during aging. Aging, 2016, 8, 1083-1101.	1.4	69
177	Modulating Mitophagy in Mitochondrial Disease. Current Medicinal Chemistry, 2019, 25, 5597-5612.	1.2	65
178	A Saudi Infant with Vici Syndrome: Case Report and Literature Review. Open Access Macedonian Journal of Medical Sciences, 2018, 6, 1081-1084.	0.1	10
179	Ophthalmologic Features of Vici Syndrome. Journal of Pediatric Ophthalmology and Strabismus, 2014, 51, 214-220.	0.3	15
180	Human platelets display dysregulated sepsis-associated autophagy, induced by altered LC3 protein-protein interaction of the Vici-protein EPG5. Autophagy, 2022, 18, 1534-1550.	4.3	7
181	Role of autophagy in muscle disease. Molecular Aspects of Medicine, 2021, 82, 101041.	2.7	26
182	Inflammatory Bowel Disease at the Intersection of Autophagy and Immunity: Insights from Human Genetics. , 2013, , 241-264.		1
183	Developmental disorder linked to autophagy. Nature Middle East, 0, , .	0.0	0
184	Sarcomeric Signaling. , 2015, , 141-160.		0
186	Neurodegenerative Diseases and Autophagy. , 2018, , 299-343.		1
187	Inflammatory Bowel Disease at the Intersection of Autophagy and Immunity: Insights from Human Genetics. , 2019, , 305-328.		2
188	Congenital and Hereditary Cataracts: Epidemiology and Genetics. , 2020, , 3-23.		3
189	Myofibrillar Myopathies and Other Myopathies with Rimmed Vacuoles. , 2020, , 361-388.		0
190	Neurological Manifestations of Primary Immunodeficiencies. Iranian Journal of Child Neurology, 2018, 12, 7-23.	0.2	8

#	ARTICLE	IF	CITATIONS
191	MOLECULAR GENETICS OF CLEFT LIP AND PALATE: A REVIEW. <i>Annals of Ibadan Postgraduate Medicine</i> , 2020, 18, S16-S21.	0.1	0
192	Cardiomyopathies in Children and Systemic Disorders When Is It Useful to Look beyond the Heart?. <i>Journal of Cardiovascular Development and Disease</i> , 2022, 9, 47.	0.8	5
194	Autophagy-Inflammation Interplay During Infection: Balancing Pathogen Clearance and Host Inflammation. <i>Frontiers in Pharmacology</i> , 2022, 13, 832750.	1.6	18
195	Material properties of phase-separated TFEB condensates regulate the autophagy-lysosome pathway. <i>Journal of Cell Biology</i> , 2022, 221, .	2.3	17
202	Macroautophagy in CNS health and disease. <i>Nature Reviews Neuroscience</i> , 2022, 23, 411-427.	4.9	44
203	Multiple roles of endocytosis and autophagy in intracellular remodeling during oocyte-to-embryo transition. <i>Proceedings of the Japan Academy Series B: Physical and Biological Sciences</i> , 2022, 98, 207-221.	1.6	3
204	Myotubularin-related phosphatase 5 is a critical determinant of autophagy in neurons. <i>Current Biology</i> , 2022, 32, 2581-2595.e6.	1.8	7
206	An induced pluripotent stem cell line (CIMRi001-A) from a Vici syndrome donor with a homozygous recessive c.1007A>G (p.Q336R) mutation in the EPG5 gene. <i>Stem Cell Research</i> , 2022, , 102833.	0.3	2
208	Novel EPG5 Mutation Associated with Vici Syndrome Gene. <i>Case Reports in Genetics</i> , 2022, 2022, 1-3.	0.1	0
209	Autophagy-associated immune dysregulation and hyperplasia in a patient with compound heterozygous mutations in <i>ATG9A</i> . <i>Autophagy</i> , 2023, 19, 678-691.	4.3	4
210	<i>PI4K2A</i> deficiency causes innate error in intracellular trafficking with developmental and epileptic–dyskinetic encephalopathy. <i>Annals of Clinical and Translational Neurology</i> , 2022, 9, 1345-1358.	1.7	10
211	Molecular Mechanism and Regulation of Autophagy and Its Potential Role in Epilepsy. <i>Cells</i> , 2022, 11, 2621.	1.8	13
212	Vici syndrome in Israel: Clinical and molecular insights. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	1
213	Interactions of Autophagy and the Immune System in Health and Diseases. , 2022, 1, 438-515.		4
214	Loss-of-function variants in <i>MYCBP2</i> cause neurobehavioural phenotypes and corpus callosum defects. <i>Brain</i> , 2023, 146, 1373-1387.	3.7	9
215	Infections in primary immunodeficiency. , 2022, , 747-790.		0
216	Response of the metabolic and autophagy pathways in <i>Mytilus</i> under starvation. <i>Frontiers in Marine Science</i> , 0, 9, .	1.2	1
217	Phenotypic expansion of EPG5-related Vici syndrome: 15 Dutch patients carrying a founder variant. <i>European Journal of Paediatric Neurology</i> , 2022, 41, 91-98.	0.7	1

#	ARTICLE	IF	CITATIONS
219	Traffic jam within lymphocytes: A clinician's perspective. <i>Frontiers in Immunology</i> , 0, 13, .	2.2	1
220	Autophagy Requirements for Eye Lens Differentiation and Transparency. <i>Cells</i> , 2023, 12, 475.	1.8	5
221	Autophagy in the eye: from physiology to pathophysiology. , 2023, 2, .		2
225	Implications of autophagy in health, disease, and aging. , 2024, , 181-205.		0