

# Vasiliki Kalatzis

## List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

63

papers

2,653

citations

25

h-index

51

g-index

70

ext. papers

2,950

ext. citations

5.9

avg, IF

4.44

L-index

#	Paper	IF	Citations
63	A human homologue of the <i>Drosophila</i> eyes absent gene underlies branchio-oto-renal (BOR) syndrome and identifies a novel gene family. <i>Nature Genetics</i> , <b>1997</b> , 15, 157-64	36.3	566
62	Cystinosis, the protein defective in cystinosis, is a H(+)-driven lysosomal cystine transporter. <i>EMBO Journal</i> , <b>2001</b> , 20, 5940-9	13	213
61	Clustering of mutations responsible for branchio-oto-renal (BOR) syndrome in the eyes absent homologous region ( <i>eyaHR</i> ) of <i>EYA1</i> . <i>Human Molecular Genetics</i> , <b>1997</b> , 6, 2247-55	5.6	160
60	Intralysosomal cystine accumulation in mice lacking cystinosis, the protein defective in cystinosis. <i>Molecular and Cellular Biology</i> , <b>2002</b> , 22, 7622-32	4.8	130
59	A proposed new contiguous gene syndrome on 8q consists of Branchio-Oto-Renal (BOR) syndrome, Duane syndrome, a dominant form of hydrocephalus and trapeze aplasia; implications for the mapping of the BOR gene. <i>Human Molecular Genetics</i> , <b>1994</b> , 3, 1859-66	5.6	105
58	The cell adhesion molecule "CAR" and sialic acid on human erythrocytes influence adenovirus in vivo biodistribution. <i>PLoS Pathogens</i> , <b>2009</b> , 5, e1000277	7.6	100
57	The targeting of cystinosis to the lysosomal membrane requires a tyrosine-based signal and a novel sorting motif. <i>Journal of Biological Chemistry</i> , <b>2001</b> , 276, 13314-21	5.4	98
56	Molecular pathogenesis of cystinosis: effect of CTNS mutations on the transport activity and subcellular localization of cystinosis. <i>Human Molecular Genetics</i> , <b>2004</b> , 13, 1361-71	5.6	93
55	BOR and BO Syndromes Are Allelic Defects of <i>EYA1</i> . <i>European Journal of Human Genetics</i> , <b>1997</b> , 5, 242-246	9.6	92
54	<i>Eya1</i> expression in the developing ear and kidney: towards the understanding of the pathogenesis of Branchio-Oto-Renal (BOR) syndrome. <i>Developmental Dynamics</i> , <b>1998</b> , 213, 486-99	2.9	90
53	Renal phenotype of the cystinosis mouse model is dependent upon genetic background. <i>Nephrology Dialysis Transplantation</i> , <b>2010</b> , 25, 1059-66	4.3	68
52	The fundamental and medical impacts of recent progress in research on hereditary hearing loss. <i>Human Molecular Genetics</i> , <b>1998</b> , 7, 1589-97	5.6	61
51	Functional rescue of REP1 following treatment with PTC124 and novel derivative PTC-414 in human choroideremia fibroblasts and the nonsense-mediated zebrafish model. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 3416-3431	5.6	54
50	Proof of concept for AAV2/5-mediated gene therapy in iPSC-derived retinal pigment epithelium of a choroideremia patient. <i>Molecular Therapy - Methods and Clinical Development</i> , <b>2014</b> , 1, 14011	6.4	52
49	New aspects of the pathogenesis of cystinosis. <i>Pediatric Nephrology</i> , <b>2003</b> , 18, 207-15	3.2	52
48	The gene for the human IgA Fc receptor maps to 19q13.4. <i>Human Genetics</i> , <b>1992</b> , 89, 107-8	6.3	51
47	Identification of 14 novel CTNS mutations and characterization of seven splice site mutations associated with cystinosis. <i>Human Mutation</i> , <b>2002</b> , 20, 439-46	4.7	46

46	A Comprehensive Analysis of Choroideremia: From Genetic Characterization to Clinical Practice. <i>PLoS ONE</i> , <b>2016</b> , 11, e0151943	3.7	37
45	Genome Editing in Patient iPSCs Corrects the Most Prevalent Mutations and Reveals Intriguing Mutant mRNA Expression Profiles. <i>Molecular Therapy - Methods and Clinical Development</i> , <b>2020</b> , 17, 156-173	6.4	35
44	Corneal transduction by intra-stromal injection of AAV vectors in vivo in the mouse and ex vivo in human explants. <i>PLoS ONE</i> , <b>2012</b> , 7, e35318	3.7	31
43	Immunolocalization of cystinosin, the protein defective in cystinosis. <i>Journal of the American Society of Nephrology: JASN</i> , <b>2002</b> , 13, 2046-51	12.7	28
42	Cystinosis: from gene to disease. <i>Nephrology Dialysis Transplantation</i> , <b>2002</b> , 17, 1883-6	4.3	28
41	Genome Editing as a Treatment for the Most Prevalent Causative Genes of Autosomal Dominant Retinitis Pigmentosa. <i>International Journal of Molecular Sciences</i> , <b>2019</b> , 20,	6.3	25
40	Guiding Lights in Genome Editing for Inherited Retinal Disorders: Implications for Gene and Cell Therapy. <i>Neural Plasticity</i> , <b>2018</b> , 2018, 5056279	3.3	25
39	The ocular anomalies in a cystinosis animal model mimic disease pathogenesis. <i>Pediatric Research</i> , <b>2007</b> , 62, 156-62	3.2	25
38	Characterization of a putative founder mutation that accounts for the high incidence of cystinosis in Brittany. <i>Journal of the American Society of Nephrology: JASN</i> , <b>2001</b> , 12, 2170-2174	12.7	24
37	Pathogenicity of a novel missense variant associated with choroideremia and its impact on gene replacement therapy. <i>Human Molecular Genetics</i> , <b>2017</b> , 26, 3573-3584	5.6	22
36	Clinical Evaluation and Cone Alterations in Choroideremia. <i>Ophthalmology</i> , <b>2016</b> , 123, 1830-1832	7.3	22
35	Characterization of a translocation-associated deletion defines the candidate region for the gene responsible for branchio-oto-renal syndrome. <i>Genomics</i> , <b>1996</b> , 34, 422-5	4.3	21
34	The effect of PTC124 on choroideremia fibroblasts and iPSC-derived RPE raises considerations for therapy. <i>Scientific Reports</i> , <b>2018</b> , 8, 8234	4.9	21
33	Zika Virus Efficiently Replicates in Human Retinal Epithelium and Disturbs Its Permeability. <i>Journal of Virology</i> , <b>2017</b> , 91,	6.6	20
32	Zika virus induces strong inflammatory responses and impairs homeostasis and function of the human retinal pigment epithelium. <i>EBioMedicine</i> , <b>2019</b> , 39, 315-331	8.8	20
31	Corrective GUSB transfer to the canine mucopolysaccharidosis VII brain. <i>Molecular Therapy</i> , <b>2014</b> , 22, 762-73	11.7	19
30	Exogenous LRRK2G2019S induces parkinsonian-like pathology in a nonhuman primate. <i>JCI Insight</i> , <b>2018</b> , 3,	9.9	19
29	Side scatter intensity is highly heterogeneous in undifferentiated pluripotent stem cells and predicts clonogenic self-renewal. <i>Stem Cells and Development</i> , <b>2013</b> , 22, 1851-60	4.4	18

28	Cystine accumulation in the CNS results in severe age-related memory deficits. <i>Neurobiology of Aging</i> , <b>2009</b> , 30, 987-1000	5.6	16
27	Corrective GUSB transfer to the canine mucopolysaccharidosis VII cornea using a helper-dependent canine adenovirus vector. <i>Journal of Controlled Release</i> , <b>2014</b> , 181, 22-31	11.7	15
26	Nonsense-mediated mRNA decay efficiency varies in choroideremia providing a target to boost small molecule therapeutics. <i>Human Molecular Genetics</i> , <b>2019</b> , 28, 1865-1871	5.6	14
25	Gene transfer may be preventive but not curative for a lysosomal transport disorder. <i>Molecular Therapy</i> , <b>2008</b> , 16, 1372-81	11.7	13
24	Study of Usutu virus neuropathogenicity in mice and human cellular models. <i>PLoS Neglected Tropical Diseases</i> , <b>2020</b> , 14, e0008223	4.8	12
23	Identification and characterisation of the murine homologue of the gene responsible for cystinosis, Ctns. <i>BMC Genomics</i> , <b>2000</b> , 1, 2	4.5	10
22	Lysosomal and network alterations in human mucopolysaccharidosis type VII iPSC-derived neurons. <i>Scientific Reports</i> , <b>2018</b> , 8, 16644	4.9	10
21	Pathogenicity of novel atypical variants leading to choroideremia as determined by functional analyses. <i>Human Mutation</i> , <b>2019</b> , 40, 31-35	4.7	7
20	Preclinical pharmacology of a lipophenol in a mouse model of light-induced retinopathy. <i>Experimental and Molecular Medicine</i> , <b>2020</b> , 52, 1090-1101	12.8	6
19	Cultured Cells from the Human Oocyte Cumulus Niche Are Efficient Feeders to Propagate Pluripotent Stem Cells. <i>Stem Cells and Development</i> , <b>2015</b> , 24, 2317-27	4.4	6
18	Allele-Specific Knockout by CRISPR/Cas to Treat Autosomal Dominant Retinitis Pigmentosa Caused by the G56R Mutation in NR2E3. <i>International Journal of Molecular Sciences</i> , <b>2021</b> , 22,	6.3	5
17	A Novel Chromosomal Translocation Identified due to Complex Genetic Instability in iPSC Generated for Choroideremia. <i>Cells</i> , <b>2019</b> , 8,	7.9	4
16	Dietary, environmental, and genetic risk factors of Extensive Macular Atrophy with Pseudodrusen, a severe bilateral macular atrophy of middle-aged patients. <i>Scientific Reports</i> , <b>2018</b> , 8, 6840	4.9	4
15	Screening for a canine model of choroideremia exclusively identifies nonpathogenic CHM variants. <i>Ophthalmic Research</i> , <b>2011</b> , 45, 155-63	2.9	4
14	Branchio-Oto-Renal syndrome. <i>Advances in Oto-Rhino-Laryngology</i> , <b>2000</b> , 56, 39-44	1.7	4
13	Generation of an iPSC line, INMi001-A, carrying the two most common USH2A mutations from a compound heterozygote with non-syndromic retinitis pigmentosa. <i>Stem Cell Research</i> , <b>2018</b> , 33, 228-232 <sup>1.6</sup>		4
12	Generation of a human iPSC line, INMi003-A, with a missense mutation in CRX associated with autosomal dominant cone-rod dystrophy. <i>Stem Cell Research</i> , <b>2019</b> , 38, 101478	1.6	3
11	Branchio-otic syndromes imbroglia <b>1999</b> , 82, 440-441		3

10	Generation of a human iPSC line, INMi002-A, carrying the most prevalent USH2A variant associated with Usher syndrome type 2. <i>Stem Cell Research</i> , <b>2018</b> , 33, 247-250	1.6	3
9	Generation of a human iPSC line, INMi004-A, with a point mutation in CRX associated with autosomal dominant Leber congenital amaurosis. <i>Stem Cell Research</i> , <b>2019</b> , 38, 101476	1.6	2
8	Mapping of the chromosome 11 breakpoint of the t(4;11)(q21;p14-15) translocation. <i>Cancer Genetics and Cytogenetics</i> , <b>1993</b> , 69, 122-5		2
7	Hereditary Cystinosis <b>2009</b> , 661-679		1
6	Novel roles for voltage-gated T-type Ca and CLC-2 channels in phagocytosis and angiogenic factor balance identified in human iPSC-derived RPE. <i>FASEB Journal</i> , <b>2021</b> , 35, e21406	0.9	1
5	CRISPR/Cas9-Mediated Genome Editing to Generate Clonal iPSC Lines. <i>Methods in Molecular Biology</i> , <b>2021</b> , 1	1.4	1
4	Identification of the First Single Exon 8 Structural Variants Associated with Autosomal Dominant Hearing Loss.. <i>Diagnostics</i> , <b>2022</b> , 12,	3.8	0
3	Generation of a human iPSC line, INMi005-A, from a patient with non-syndromic USH2A-associated retinitis pigmentosa.. <i>Stem Cell Research</i> , <b>2022</b> , 60, 102738	1.6	0
2	iPSCs for modeling choroideremia <b>2022</b> , 123-151		
1	Molecular Therapy for Choroideremia: Pre-clinical and Clinical Progress to Date. <i>Molecular Diagnosis and Therapy</i> , <b>2021</b> , 25, 661-675	4.5	