

# Ji Yeon Kim

## List of Publications by Year in descending order

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35  
papers

577  
citations

687363

13  
h-index

642732

23  
g-index

35  
all docs

35  
docs citations

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times ranked

1037  
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical Utility of Methylation-Specific Multiplex Ligation-Dependent Probe Amplification for the Diagnosis of Praderâ€“Willi Syndrome and Angelman Syndrome. <i>Annals of Laboratory Medicine</i> , 2022, 42, 79-88.	2.5	2
2	Strabismus in chronic progressive external ophthalmoplegia. <i>Acta Ophthalmologica</i> , 2021, 99, e274-e280.	1.1	0
3	Cigarette smoke extract-induced downregulation of p300 is responsible for the impaired inflammatory cytokine response of macrophages. <i>Cellular Signalling</i> , 2021, 85, 110050.	3.6	5
4	No association between POU4F1, POU4F2, ISL1 polymorphisms and normal-tension glaucoma. <i>Ophthalmic Genetics</i> , 2020, 41, 427-431.	1.2	1
5	Replicationâ€“Based Rearrangements Are a Common Mechanism for SNCA Duplication in Parkinson's Disease. <i>Movement Disorders</i> , 2020, 35, 868-876.	3.9	9
6	Genomic Characterization of TBK1 Duplication in Korean Normal-tension Glaucoma Patients. <i>Journal of Glaucoma</i> , 2020, 29, 331-336.	1.6	8
7	Evaluation of the new Abbott Real-Time EBV assay: fully automated quantification of EBV in whole blood by targeting BLLF1. <i>Diagnostic Microbiology and Infectious Disease</i> , 2019, 94, 135-139.	1.8	3
8	Novel MT-ND5 gene mutation identified in Leber's hereditary optic neuropathy patient using mitochondrial genome sequencing. <i>Journal of the Neurological Sciences</i> , 2017, 375, 301-303.	0.6	4
9	Author reply: â€œMtDNA m.3472T > C could be classified as a primary mutation of Leber's hereditary optic neuropathyâ€“. <i>Journal of the Neurological Sciences</i> , 2017, 382, 166-167.	0.6	0
10	MtDNA m.3472T > C could be classified as a primary mutation of Leber's hereditary optic neuropathy. <i>Journal of the Neurological Sciences</i> , 2017, 380, 174-176.	0.6	3
11	The Relation Between Endothelial Nitric Oxide Synthase Polymorphisms and Normal Tension Glaucoma. <i>Journal of Glaucoma</i> , 2017, 26, 1030-1035.	1.6	19
12	Large Deletions of <i>TSPAN12</i> Cause Familial Exudative Vitreoretinopathy (FEVR)., 2016, 57, 6902.		11
13	Pitfalls of Multiple Ligation-Dependent Probe Amplifications in Detecting DMD Exon Deletions or Duplications. <i>Journal of Molecular Diagnostics</i> , 2016, 18, 253-259.	2.8	23
14	Performance of two commercially available BCR-ABL1 quantification assays that use an international reporting scale. <i>Clinical Chemistry and Laboratory Medicine</i> , 2016, 54, 1157-60.	2.3	0
15	Molecular Characterization of <i>FZD4</i> , <i>LRP5</i> , and <i>TSPAN12</i> in Familial Exudative Vitreoretinopathy. , 2015, 56, 5143.		46
16	Case of mild Schmid-type metaphyseal chondrodysplasia with novel sequence variation involving an unusual mutational site of the COL10A1 gene. <i>European Journal of Medical Genetics</i> , 2015, 58, 175-179.	1.3	12
17	Mutational spectrum of the SPAST and ATL1 genes in Korean patients with hereditary spastic paraplegia. <i>Journal of the Neurological Sciences</i> , 2015, 357, 167-172.	0.6	17
18	Diagnostic Application of an Extensive Gene Panel for Leber Congenital Amaurosis with Severe Genetic Heterogeneity. <i>Journal of Molecular Diagnostics</i> , 2015, 17, 100-105.	2.8	17

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19	Chimerism Monitoring after Allogeneic Hematopoietic Stem Cell Transplantation Using Quantitative Real-Time PCR of Biallelic Insertion/Deletion Polymorphisms. <i>Journal of Molecular Diagnostics</i> , 2014, 16, 679-688.	2.8	31
20	Phase II trial of RAD001 in renal cell carcinoma patients with non-clear cell histology.. <i>Journal of Clinical Oncology</i> , 2012, 30, 4544-4544.	1.6	0
21	Relative contribution of SCA2, SCA3 and SCA17 in Korean patients with parkinsonism and ataxia. <i>Parkinsonism and Related Disorders</i> , 2011, 17, 338-342.	2.2	20
22	Two Parkinson's disease patients with $\hat{L}$ -synuclein gene duplication and rapid cognitive decline. <i>Movement Disorders</i> , 2010, 25, 957-959.	3.9	23
23	Molecular Characterization of the NF2 Gene in Korean Patients with Neurofibromatosis Type 2: A Report of Four Novel Mutations. <i>Annals of Laboratory Medicine</i> , 2010, 30, 190-194.	2.5	7
24	Molecular identification of the novel $G\hat{I}^3\text{-}\hat{I}^2$ hybrid hemoglobin: Hb $G\hat{I}^3\text{-}\hat{I}^2$ Ulsan ( $G\hat{I}^3$ through 13; $\hat{I}^2$ from 19). <i>Blood Cells, Molecules, and Diseases</i> , 2010, 45, 276-279.	1.4	5
25	Low contribution of BRCA1/2 genomic rearrangement to high-risk breast cancer in the Korean population. <i>Familial Cancer</i> , 2009, 8, 505-508.	1.9	20
26	Growth kinetics and transplantation of human retinal progenitor cells. <i>Experimental Eye Research</i> , 2009, 89, 301-310.	2.6	66
27	Ophthalmoplegia Diagnosis. <i>Ophthalmology</i> , 2009, 116, 813-814.e2.	5.2	5
28	<i>LCA5</i> , a Rare Genetic Cause of Leber Congenital Amaurosis in Koreans. <i>Ophthalmic Genetics</i> , 2009, 30, 54-55.	1.2	7
29	False Homozygous Deletions of <i>SMN1</i> Exon 7 Using <i>Dra</i> I PCR-RFLP Caused by a Novel Mutation in Spinal Muscular Atrophy. <i>Genetic Testing and Molecular Biomarkers</i> , 2009, 13, 511-513.	0.7	14
30	Correction of Contracted Nail Deformity by Distraction Lengthening. <i>Annals of Plastic Surgery</i> , 2008, 61, 153-156.	0.9	10
31	Molecular and Clinical Characteristics of Myotonic Dystrophy Type 1 in Koreans. <i>Annals of Laboratory Medicine</i> , 2008, 28, 483-492.	2.5	13
32	Molecular characterization of D- Korean persons: development of a diagnostic strategy. <i>Transfusion</i> , 2005, 45, 345-352.	1.6	85
33	Spectrum of the mitochondrial DNA mutations of Leber's hereditary optic neuropathy in Koreans. <i>Journal of Neurology</i> , 2003, 250, 278-281.	3.6	26
34	Mitochondrial DNA C4171A/ND1 is a novel primary causative mutation of Leber's hereditary optic neuropathy with a good prognosis. <i>Annals of Neurology</i> , 2002, 51, 630-634.	5.3	56
35	Spinocerebellar ataxia type 2 in seven Korean families: CAG trinucleotide expansion and clinical characteristics. <i>Journal of Korean Medical Science</i> , 1999, 14, 659.	2.5	9