Nikolas Pontikos

List of Publications by Year
in descending order

104	6,054	159358	82410
papers	citations	30 h-index	72 g-index

121	121	121	12563
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Reorganizing the protein space at the Universal Protein Resource (UniProt). Nucleic Acids Research, 2012, 40, D71-D75.	6.5	1,196
2	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
3	Ongoing and future developments at the Universal Protein Resource. Nucleic Acids Research, 2011, 39, D214-D219.	6.5	649
4	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	13.9	352
5	Functional variants in the LRRK2 gene confer shared effects on risk for Crohnâ€™s disease and Parkinsonâ€™s disease. Science Translational Medicine, 2018, 10, .	5.8	273
6	Standardizing Flow Cytometry Immunophenotyping Analysis from the Human ImmunoPhenotyping Consortium. Scientific Reports, 2016, 6, 20686.	1.6	240
7	Automated deep learning design for medical image classification by health-care professionals with no coding experience: a feasibility study. The Lancet Digital Health, 2019, 1, e232-e242.	5.9	183
8	Genetic Basis of Inherited Retinal Disease in a Molecularly Characterized Cohort of More Than 3000 Families from the United Kingdom. Ophthalmology, 2020, 127, 1384-1394.	2.5	131
9	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315.	2.6	121
10	Code-free deep learning for multi-modality medical image classification. Nature Machine Intelligence, 2021, 3, 288-298.	8.3	90
11	The Human Salivary Microbiome Is Shaped by Shared Environment Rather than Genetics: Evidence from a Large Family of Closely Related Individuals. MBio, 2017, 8, .	1.8	82
12	The complex genetic landscape of familial MDS and AML reveals pathogenic germline variants. Nature Communications, 2020, 11, 1044.	5.8	81
13	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2020, 107, 802-814.	2.6	75
14	Autosomal-Dominant Corneal Endothelial Dystrophies CHED1 and PPCD1 Are Allelic Disorders Caused by Non-coding Mutations in the Promoter of OVOL2. American Journal of Human Genetics, 2016, 98, 75-89.	2.6	70
15	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. PLoS Genetics, 2018, 14, e1007329.	1.5	66
16	The X-linked retinopathies: Physiological insights, pathogenic mechanisms, phenotypic features and novel therapies. Progress in Retinal and Eye Research, 2021, 82, 100898.	7.3	65
17	Predicting sex from retinal fundus photographs using automated deep learning. Scientific Reports, 2021, 11, 10286.	1.6	65
18	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	2.6	61

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19	Dissection of a Complex Disease Susceptibility Region Using a Bayesian Stochastic Search Approach to Fine Mapping. PLoS Genetics, 2015, 11, e1005272.	1.5	55
20	Genetic Complexity of Crohnâ€™s Disease in Two Large Ashkenazi Jewish Families. Gastroenterology, 2016, 151, 698-709.	0.6	54
21	A Frameshift in CSF2RB Predominant Among Ashkenazi Jews Increases Risk for Crohn's Disease and Reduces Monocyte Signaling via GM-CSF. Gastroenterology, 2016, 151, 710-723.e2.	0.6	51
22	Mutations in CPAMD8 Cause a Unique Form of Autosomal-Recessive Anterior Segment Dysgenesis. American Journal of Human Genetics, 2016, 99, 1338-1352.	2.6	47
23	Ectopic GRHL2 Expression Due to Non-coding Mutations Promotes Cell State Transition and Causes Posterior Polymorphous Corneal Dystrophy 4. American Journal of Human Genetics, 2018, 102, 447-459.	2.6	45
24	Genetic Variants Associated With Corneal Biomechanical Properties and Potentially Conferring Susceptibility to Keratoconus in a Genome-Wide Association Study. JAMA Ophthalmology, 2019, 137, 1005.	1.4	45
25	Deep Phenotyping ofPDE6C-Associated Achromatopsia. , 2019, 60, 5112.		44
26	Clinically relevant deep learning for detection and quantification of geographic atrophy from optical coherence tomography: a model development and external validation study. The Lancet Digital Health, 2021, 3, e665-e675.	5.9	44
27	Association of Steroid 5Î±-Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. JAMA Ophthalmology, 2017, 135, 339.	1.4	43
28	An Improved Phenotype-Driven Tool for Rare Mendelian Variant Prioritization: Benchmarking Exomiser on Real Patient Whole-Exome Data. Genes, 2020, 11, 460.	1.0	42
29	Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. Bioinformatics, 2017, 33, 2421-2423.	1.8	40
30	GUCY2D-Associated Leber Congenital Amaurosis: A Retrospective Natural History Study in Preparation for Trials of Novel Therapies. American Journal of Ophthalmology, 2020, 210, 59-70.	1.7	39
31	The GA4GH Phenopacket schema defines a computable representation of clinical data. Nature Biotechnology, 2022, 40, 817-820.	9.4	38
32	Genome instability is a consequence of transcription deficiency in patients with bone marrow failure harboring biallelic ERCC6L2 variants. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 7777-7782.	3.3	37
33	A multi-ethnic genome-wide association study implicates collagen matrix integrity and cell differentiation pathways in keratoconus. Communications Biology, 2021, 4, 266.	2.0	36
34	Personal Genome Project UK (PGP-UK): a research and citizen science hybrid project in support of personalized medicine. BMC Medical Genomics, 2018, 11, 108.	0.7	34
35	Artificial intelligence extension of the OSCARâ€IB criteria. Annals of Clinical and Translational Neurology, 2021, 8, 1528-1542.	1.7	33
36	Prediction of Causative Genes in Inherited Retinal Disorders from Spectral-Domain Optical Coherence Tomography Utilizing Deep Learning Techniques. Journal of Ophthalmology, 2019, 2019, 1-7.	0.6	32

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37	Juvenile Batten Disease (CLN3): Detailed Ocular Phenotype, Novel Observations, Delayed Diagnosis, Masquerades, and Prospects for Therapy. Ophthalmology Retina, 2020, 4, 433-445.	1.2	31
38	Autosomal Recessive Bestrophinopathy. Ophthalmology, 2021, 128, 706-718.	2.5	31
39	Moorfields AMD database report 2: fellow eye involvement with neovascular age-related macular degeneration. British Journal of Ophthalmology, 2020, 104, 684-690.	2.1	26
40	Identification of genetic factors influencing metabolic dysregulation and retinal support for MacTel, a retinal disorder. Communications Biology, 2021, 4, 274.	2.0	26
41	Recessive Retinopathy Consequent on Mutant G-Protein Î² Subunit 3 (GNB3). JAMA Ophthalmology, 2016, 134, 924.	1.4	25
42	One- and two-year visual outcomes from the Moorfields age-related macular degeneration database: a retrospective cohort study and an open science resource. BMJ Open, 2019, 9, e027441.	0.8	25
43	Loss-of-Function Mutations in the CFH Gene Affecting Alternatively Encoded Factor H-like 1 Protein Cause Dominant Early-Onset Macular Drusen. Ophthalmology, 2019, 126, 1410-1421.	2.5	25
44	The genetic landscape of crystallins in congenital cataract. Orphanet Journal of Rare Diseases, 2020, 15, 333.	1.2	25
45	Clinical and Genetic Characteristics of 18 Patients from 13 Japanese Families with CRX-associated retinal disorder: Identification of Genotype-phenotype Association. Scientific Reports, 2020, 10, 9531.	1.6	24
46	Duplication events downstream of IRX1 cause North Carolina macular dystrophy at the MCDR3 locus. Scientific Reports, 2017, 7, 7512.	1.6	23
47	Missense variants in the X-linked genePRPS1cause retinal degeneration in females. Human Mutation, 2018, 39, 80-91.	1.1	23
48	AlzEye: longitudinal record-level linkage of ophthalmic imaging and hospital admissions of 353â€‰157 patients in London, UK. BMJ Open, 2022, 12, e058552.	0.8	22
49	Clinical and genetic characteristics of 10 Japanese patients with PROM1 â€associated retinal disorder: A report of the phenotype spectrum and a literature review in the Japanese population. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 656-674.	0.7	21
50	Genetic Spectrum of EYS-associated Retinal Disease in a Large Japanese Cohort: Identification of Disease-associated Variants with Relatively High Allele Frequency. Scientific Reports, 2020, 10, 5497.	1.6	21
51	Clinical and Molecular Characterization of Familial Exudative Vitreoretinopathy Associated With Microcephaly. American Journal of Ophthalmology, 2019, 207, 87-98.	1.7	20
52	Unique noncoding variants upstream of PRDM13 are associated with a spectrum of developmental retinal dystrophies including progressive bifocal chorioretinal atrophy. Human Mutation, 2019, 40, 578-587.	1.1	19
53	A novel missense mutation in HSF4 causes autosomal-dominant congenital lamellar cataract in a British family. Eye, 2018, 32, 806-812.	1.1	18
54	Phenotypical Characteristics ofPOC1B-Associated Retinopathy in Japanese Cohort: Cone Dystrophy With Normal Funduscopic Appearance. , 2019, 60, 3432.		18

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55	KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Courseâ€"KCNV2 Study Group Report 1. American Journal of Ophthalmology, 2021, 225, 95-107.	1.7	17
56	Enablers and Barriers to Deployment of Smartphone-Based Home Vision Monitoring in Clinical Practice Settings. JAMA Ophthalmology, 2022, 140, 153.	1.4	17
57	Homozygous OB-fold variants in telomere protein TPP1 are associated with dyskeratosis congenitaâ€"like phenotypes. Blood, 2018, 132, 1349-1353.	0.6	16
58	Myelodysplasia and liver disease extend the spectrum of RTEL1 related telomeropathies. Haematologica, 2017, 102, e293-e296.	1.7	15
59	Clinical and Genetic Characteristics of 15 Affected Patients From 12 Japanese Families with GUCY2D-Associated Retinal Disorder. Translational Vision Science and Technology, 2020, 9, 2.	1.1	15
60	A recurrent splice-site mutation in EPHA2 causing congenital posterior nuclear cataract. Ophthalmic Genetics, 2018, 39, 236-241.	0.5	13
61	Pathogenic NR2F1 variants cause a developmental ocular phenotype recapitulated in a mutant mouse model. Brain Communications, 2021, 3, fcab162.	1.5	13
62	A hybrid qPCR/SNP array approach allows cost efficient assessment of KIR gene copy numbers in large samples. BMC Genomics, 2014, 15, 274.	1.2	12
63	Panelâ€based genetic testing for inherited retinal disease screening 176 genes. Molecular Genetics & Genomic Medicine, 2021, 9, e1663.	0.6	12
64	Prediction of causative genes in inherited retinal disorder from fundus photography and autofluorescence imaging using deep learning techniques. British Journal of Ophthalmology, 2021, 105, 1272-1279.	2.1	12
65	Machine Learning Algorithms to Detect Subclinical Keratoconus: Systematic Review. JMIR Medical Informatics, 2021, 9, e27363.	1.3	12
66	ReLayer: a Free, Online Tool for Extracting Retinal Thickness From Cross-Platform OCT Images. Translational Vision Science and Technology, 2019, 8, 25.	1.1	11
67	KCNV2-Associated Retinopathy: Detailed Retinal Phenotype and Structural Endpointsâ€"KCNV2 Study Group Report 2. American Journal of Ophthalmology, 2021, 230, 1-11.	1.7	11
68	Spatial Functional Characteristics of East Asian Patients With Occult Macular Dystrophy (Miyake) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	1.7	10
69	Pheno4J: a gene to phenotype graph database. Bioinformatics, 2017, 33, 3317-3319.	1.8	9
70	Frequency and distribution of corneal astigmatism and keratometry features in adult life: Methodology and findings of the UK Biobank study. PLoS ONE, 2019, 14, e0218144.	1.1	9
71	Factors in Color Fundus Photographs That Can Be Used by Humans to Determine Sex of Individuals. Translational Vision Science and Technology, 2020, 9, 8.	1.1	9
72	Rare coding variant analysis in a large cohort of Ashkenazi Jewish families with inflammatory bowel disease. Human Genetics, 2018, 137, 723-734.	1.8	8

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73	The utility of massively parallel sequencing for posterior polymorphous corneal dystrophy type 3 molecular diagnosis. Experimental Eye Research, 2019, 182, 160-166.	1.2	8
74	Comment on: Trends in Retina Specialist Imaging Utilization From 2012 to 2016 in the United States Medicare Fee-for-Service Population. American Journal of Ophthalmology, 2020, 211, 229.	1.7	8
75	A novel missense mutation inLIM2causing isolated autosomal dominant congenital cataract. Ophthalmic Genetics, 2020, 41, 131-134.	0.5	8
76	Associations of Alcohol Consumption and Smoking With Disease Risk and Neurodegeneration in Individuals With Multiple Sclerosis in the United Kingdom. JAMA Network Open, 2022, 5, e220902.	2.8	8
77	Electrical responses from human retinal cone pathways associate with a common genetic polymorphism implicated in myopia. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	3.3	8
78	Extending the phenotypic spectrum of PRPF8, PRPH2, RP1 and RPGR, and the genotypic spectrum of early-onset severe retinal dystrophy. Orphanet Journal of Rare Diseases, 2021, 16, 128.	1.2	7
79	Personalized Model to Predict Keratoconus Progression From Demographic, Topographic, and Genetic Data. American Journal of Ophthalmology, 2022, 240, 321-329.	1.7	7
80	Whole-genome sequencing reveals a recurrent missense mutation in the Connexin 46 (GJA3) gene causing autosomal-dominant lamellar cataract. Eye, 2018, 32, 1661-1668.	1.1	6
81	Phenogenon: Gene to phenotype associations for rare genetic diseases. PLoS ONE, 2020, 15, e0230587.	1.1	6
82	Novel homozygous splicing mutations in cause autosomal recessive retinitis pigmentosa. Molecular Vision, 2018, 24, 603-612.	1.1	6
83	Elevation in Cell Cycle and Protein Metabolism Gene Transcription in Inactive Colonic Tissue From Icelandic Patients With Ulcerative Colitis. Inflammatory Bowel Diseases, 2019, 25, 317-327.	0.9	5
84	RP2 â€associated retinal disorder in a Japanese cohort: Report of novel variants and a literature review, identifying a genotypeâ€“phenotype association. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 675-693.	0.7	5
85	Familial Limbal Stem Cell Deficiency: Clinical, Cytological and Genetic Characterization. Stem Cell Reviews and Reports, 2018, 14, 148-151.	5.6	4
86	CUGC for posterior polymorphous corneal dystrophy (PPCD). European Journal of Human Genetics, 2020, 28, 126-131.	1.4	4
87	Whole Exome Sequencing Reveals Novel and Recurrent Disease-Causing Variants in Lens Specific Gap Junctional Protein Encoding Genes Causing Congenital Cataract. Genes, 2020, 11, 512.	1.0	4
88	Pathogenic variants in the CYP21A2 gene cause isolated autosomal dominant congenital posterior polar cataracts. Ophthalmic Genetics, 2022, 43, 218-223.	0.5	4
89	A recurrent variant in LIM2 causes an isolated congenital sutural/lamellar cataract in a Japanese family. Ophthalmic Genetics, 2022, 43, 622-626.	0.5	4
90	Non-Penetrance for Ocular Phenotype in Two Individuals Carrying Heterozygous Loss-of-Function ZEB1 Alleles. Genes, 2021, 12, 677.	1.0	3

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91	Posterior corneal vesicles are not associated with the genetic variants that cause posterior polymorphous corneal dystrophy. Acta Ophthalmologica, 2022, 100, .	0.6	3
92	Genome-wide linkage and haplotype sharing analysis implicates the MCDR3 locus as a candidate region for a developmental macular disorder in association with digit abnormalities. Ophthalmic Genetics, 2017, 38, 511-519.	0.5	2
93	ADDO: a comprehensive toolkit to detect, classify and visualize additive and non-additive quantitative trait loci. Bioinformatics, 2020, 36, 1517-1521.	1.8	2
94	Exploring the potential for acute anterior uveitis (AAU) patients to self-manage recurrences via a mobile application: qualitative analysis of a Moorfields Patient Experience focus group. Eye, 2020, 35, 2895-2896.	1.1	2
95	A frameshift variant in specificity protein 1 triggers superactivation of Sp1-mediated transcription in familial bone marrow failure. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 17151-17155.	3.3	2
96	Variants in PAX6, PITX3 and HSF4 causing autosomal dominant congenital cataracts. Eye, 2022, 36, 1694-1701.	1.1	2
97	Fractionated Stereotactic Radiation Therapy for Pituitary Adenomas: An alternative escalating protocol of hypofractionated stereotactic radiotherapy delivering 35 Gy in 5 fractions. Cancer Radiotherapie: Journal De La Societe Francaise De Radiotherapie Oncologique, 2021, , .	0.6	2
98	Collaborative Research and Development of a Novel, Patient-Centered Digital Platform (MyEyeSite) for Rare Inherited Retinal Disease Data: Acceptability and Feasibility Study. JMIR Formative Research, 2022, 6, e21341.	0.7	2
99	Genome Analysis for Inherited Retinal Disease: The State of the Art. Essentials in Ophthalmology, 2021, , 153-168.	0.0	1
100	Cloud-based genomics pipelines for ophthalmology: reviewed from research to clinical practice. Modeling and Artificial Intelligence in Ophthalmology, 2021, 3, 101-140.	0.1	1
101	Stargardt Macular Dystrophy. , 2022, , 151-168.		1
102	â€˜It's a bit of a grey areaâ€™: challenges faced by stop smoking practitioners when advising on e-cigarettes. Journal of Smoking Cessation, 2020, 15, 44-49.	0.3	0
103	Seqfam: A python package for analysis of Next Generation Sequencing DNA data in families. F1000Research, 0, 7, 281.	0.8	0
104	Integrating exome and whole genome analysis with the Human Phenotype Ontology for discovery of new genes in rare eye diseases. Acta Ophthalmologica, 2019, 97, .	0.6	0