Petra Liskova

List of Publications by Year
in descending order

96 papers	2,382 citations	218677 26 h-index	243625 44 g-index
110 all docs	110 docs citations	110 times ranked	3087 citing authors

#	Article	IF	Citations
1	Posterior corneal vesicles are not associated with the genetic variants that cause posterior polymorphous corneal dystrophy. Acta Ophthalmologica, 2022, 100, .	1.1	3
2	Comprehensive variant spectrum of the CNGA3 gene in patients affected by achromatopsia. Human Mutation, 2022, 43, 832-858.	2.5	8
3	Pigmentary retinopathy can indicate the presence of pathogenic LAMP2 variants even in somatic mosaic carriers with no additional signs of Danon disease. Acta Ophthalmologica, 2021, 99, 61-68.	1.1	5
4	The need for widely available genomic testing in rare eye diseases: an ERN-EYE position statement. Orphanet Journal of Rare Diseases, 2021, 16, 142.	2.7	25
5	A multi-ethnic genome-wide association study implicates collagen matrix integrity and cell differentiation pathways in keratoconus. Communications Biology, 2021, 4, 266.	4.4	36
6	Non-Penetrance for Ocular Phenotype in Two Individuals Carrying Heterozygous Loss-of-Function ZEB1 Alleles. Genes, 2021, 12, 677.	2.4	3
7	Artificial intelligence extension of the OSCARâ€IB criteria. Annals of Clinical and Translational Neurology, 2021, 8, 1528-1542.	3.7	33
8	Comprehensive phenotypic and functional analysis of dominant and recessiveFOXE3alleles in ocular developmental disorders. Human Molecular Genetics, 2021, 30, 1591-1606.	2.9	6
9	ALG3-CDG: a patient with novel variants and review of the genetic and ophthalmic findings. BMC Ophthalmology, 2021, 21, 249.	1.4	4
10	Machine Learning Algorithms to Detect Subclinical Keratoconus: Systematic Review. JMIR Medical Informatics, 2021, 9, e27363.	2.6	12
11	Novel diseaseâ€causing variants and phenotypic features of Xâ€linked megalocornea. Acta Ophthalmologica, 2021, , .	1.1	1
12	Hereditary gelsolin amyloidosis â€“ clinical symptoms and molecular genetic cause. Ceska A Slovenska Neurologie A Neurochirurgie, 2021, 84/117, .	0.1	0
13	Clinical and Genetic Study of X-Linked Juvenile Retinoschisis in the Czech Population. Genes, 2021, 12, 1816.	2.4	4
14	Should Patients with Kearns-Sayre Syndrome and Corneal Endothelial Failure Be Genotyped for a TCF4 Trinucleotide Repeat, Commonly Associated with Fuchs Endothelial Corneal Dystrophy?. Genes, 2021, 12, 1918.	2.4	0
15	CUGC for posterior polymorphous corneal dystrophy (PPCD). European Journal of Human Genetics, 2020, 28, 126-131.	2.8	4
16	Alu â€mediated Xq24 deletion encompassing CUL4B , LAMP2 , ATP1B4 , TMEM255A , and ZBTB33 genes causes Danon disease in a female patient. American Journal of Medical Genetics, Part A, 2020, 182, 219-223.	1.2	9
17	Keratoconus in Children: A Literature Review. Cornea, 2020, 39, 1592-1598.	1.7	25
18	Hereditary hyperferritinemia-cataract syndrome in three Czech families: molecular genetic testing and clinical implications. Journal of AAPOS, 2020, 24, 352.e1-352.e5.	0.3	2

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19	Multirater Validation of Peripapillary Hyperreflective Ovoid Mass-like Structures (PHOMS). Neuro-Ophthalmology, 2020, 44, 413-414.	1.0	15
20	The Phenotypic Spectrum of 47 Czech Patients with Single, Large-Scale Mitochondrial DNA Deletions. Brain Sciences, 2020, 10, 766.	2.3	8
21	Association of Sex With Frequent and Mild ABCA4 Alleles in Stargardt Disease. JAMA Ophthalmology, 2020, 138, 1035.	2.5	31
22	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. Genetics in Medicine, 2020, 22, 1235-1246.	2.4	92
23	Expanding the phenotype spectrum associated with pathogenic variants in the COL2A1 and COL11A1 genes. Annals of Human Genetics, 2020, 84, 380-392.	0.8	11
24	Pseudodominant Nanophthalmos in a Roma Family Caused by a Novel PRSS56 Variant. Journal of Ophthalmology, 2020, 2020, 1-9.	1.3	3
25	A novel missense mutation inLIM2causing isolated autosomal dominant congenital cataract. Ophthalmic Genetics, 2020, 41, 131-134.	1.2	8
26	Brittle cornea syndrome: Disease-causing mutations in ZNF469 and two novel variants identified in a patient followed for 26 years. Biomedical Papers of the Medical Faculty of the University Palacký, Olomouc, Czechoslovakia, 2020, 164, 183-188.	0.6	6
27	Dominant (Kjerâ€™s) optic atrophy as sociated with mutations in OPA1 gene. Ceska A Slovenska Neurologie A Neurochirurgie, 2020, 83/116, 33-42.	0.1	0
28	Phenotype Variability in Czech Patients Carrying PAX6 Disease-Causing Variants. Folia Biologica, 2020, 66, 123-132.	0.6	0
29	IPSC-Derived Corneal Endothelial-like Cells Act as an Appropriate Model System to Assess the Impact of SLC4A11 Variants on Pre-mRNA Splicing. , 2019, 60, 3084.		18
30	Genetic Variants Associated With Corneal Biomechanical Properties and Potentially Conferring Susceptibility to Keratoconus in a Genome-Wide Association Study. JAMA Ophthalmology, 2019, 137, 1005.	2.5	45
31	Paraproteinemic keratopathy associated with monoclonal gammopathy of undetermined significance (<scp>MGUS</scp>): clinical findings in twelve patients including recurrence after keratoplasty. Acta Ophthalmologica, 2019, 97, e987-e992.	1.1	13
32	The utility of massively parallel sequencing for posterior polymorphous corneal dystrophy type 3 molecular diagnosis. Experimental Eye Research, 2019, 182, 160-166.	2.6	8
33	CRISPR/Cas9-targeted enrichment and long-read sequencing of the Fuchs endothelial corneal dystrophyâ€“associated TCF4 triplet repeat. Genetics in Medicine, 2019, 21, 2092-2102.	2.4	56
34	Coincidental Occurrence of Schnyder Corneal Dystrophy and Posterior Polymorphous Corneal Dystrophy Type 3. Cornea, 2019, 38, 758-760.	1.7	3
35	Peripapillary microcirculation in Leber hereditary optic neuropathy. Acta Ophthalmologica, 2019, 97, e71-e76.	1.1	23
36	Congenital fibrosis of the extraocular muscles in a Czech family and its molecular genetic cause. Ceska A Slovenska Neurologie A Neurochirurgie, 2019, 82/115, 561-566.	0.1	0

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37	Molecular Genetic Cause of Achromatopsia in Two Patients of Czech Origin. Ceska A Slovenska Oftalmologie, 2019, 75, 272-276.	0.2	0
38	Review of SRD5A3 Disease-Causing Sequence Variants and Ocular Findings in Steroid 5Î±-Reductase Type 3 Congenital Disorder of Glycosylation, and a Detailed New Case. Folia Biologica, 2019, 65, 134-141.	0.6	5
39	Antisense Therapy for a Common Corneal Dystrophy Ameliorates TCF4 Repeat Expansion-Mediated Toxicity. American Journal of Human Genetics, 2018, 102, 528-539.	6.2	59
40	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.	6.2	45
41	SD-OCT imaging as a valuable tool to support molecular genetic diagnostics of Usher syndrome type 1. Journal of AAPOS, 2018, 22, 312-314.e3.	0.3	2
42	Familial Limbal Stem Cell Deficiency: Clinical, Cytological and Genetic Characterization. Stem Cell Reviews and Reports, 2018, 14, 148-151.	5.6	4
43	Analysis of <scp>KERA</scp> in four families with cornea plana identifies two novel mutations. Acta Ophthalmologica, 2018, 96, e87-e91.	1.1	4
44	Schnyder corneal dystrophy and associated phenotypes caused by novel and recurrent mutations in the UBIAD1 gene. BMC Ophthalmology, 2018, 18, 250.	1.4	9
45	Ex vivo 3D human corneal stroma model for Schnyder corneal dystrophy - role of autophagy in its pathogenesis and resolution. Histology and Histopathology, 2018, 33, 455-462.	0.7	4
46	<scp>OPA</scp>1 analysis in an international series of probands with bilateral optic atrophy. Acta Ophthalmologica, 2017, 95, 363-369.	1.1	7
47	Segregation of aÂnovel p.(Ser270Tyr) MAF mutation and p.(Tyr56âˆ—) CRYGD variant in a family with dominantly inherited congenital cataracts. Molecular Biology Reports, 2017, 44, 435-440.	2.3	9
48	Retinal layer segmentation in multiple sclerosis: a systematic review and meta-analysis. Lancet Neurology, The, 2017, 16, 797-812.	10.2	397
49	Copper in Keratoconic Corneas. Cornea, 2017, 36, e14-e14.	1.7	5
50	Replication of SNP associations with keratoconus in a Czech cohort. PLoS ONE, 2017, 12, e0172365.	2.5	22
51	Myxovirus Resistance Protein A mRNA Expression Kinetics in Multiple Sclerosis Patients Treated with IFNÎ². PLoS ONE, 2017, 12, e0169957.	2.5	1
52	Leber Hereditary Optic Neuropathy. Ceska A Slovenska Neurologie A Neurochirurgie, 2017, 80/113, 534-544.	0.1	2
53	Early detection of bilateral cataracts in utero may represent a manifestation of severe congenital disease. American Journal of Medical Genetics, Part A, 2016, 170, 1843-1848.	1.2	6
54	Analysis of FOXL2 detects three novel mutations and an atypical phenotype of blepharophimosisâ€ptosisâ€epicanthus inversus syndrome. Clinical and Experimental Ophthalmology, 2016, 44, 757-762.	2.6	10

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55	Novel TGFBI mutation p.(Leu558Arg) in a lattice corneal dystrophy patient. Ophthalmic Genetics, 2016, 37, 473-474.	1.2	10
56	Phenotypic features of CRB1-associated early-onset severe retinal dystrophy and the different molecular approaches to identifying the disease-causing variants. Graefe's Archive for Clinical and Experimental Ophthalmology, 2016, 254, 1833-1839.	1.9	19
57	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.	6.2	70
58	Unique presentation of LHON/MELAS overlap syndrome caused by m.13046T>C in MTND5. Ophthalmic Genetics, 2016, 37, 419-423.	1.2	10
59	Heterozygous deletions at the ZEB1 locus verify haploinsufficiency as the mechanism of disease for posterior polymorphous corneal dystrophy type 3. European Journal of Human Genetics, 2016, 24, 985-991.	2.8	33
60	The presence of lysyl oxidase-like enzymes in human control and keratoconic corneas. Histology and Histopathology, 2016, 31, 63-71.	0.7	11
61	Detailed Assessment of Renal Function in a Proband with Harboyan Syndrome Caused by a Novel Homozygous <i>SLC4A11 </i>Nonsense Mutation. Ophthalmic Research, 2015, 53, 30-35.	1.9	9
62	Brittle Cornea Syndrome ZNF469 Mutation Carrier Phenotype and Segregation Analysis of Rare ZNF469 Variants in Familial Keratoconus. Investigative Ophthalmology and Visual Science, 2015, 56, 578-586.	3.3	33
63	Is copper imbalance an environmental factor influencing keratoconus development?. Medical Hypotheses, 2015, 84, 518-524.	1.5	10
64	Validation of rs2956540:G>C and rs3735520:G>A association with keratoconus in a population of European descent. European Journal of Human Genetics, 2015, 23, 1581-1583.	2.8	34
65	Identification of Six Novel Mutations inZEB1and Description of the Associated Phenotypes in Patients with Posterior Polymorphous Corneal Dystrophy 3. Annals of Human Genetics, 2015, 79, 1-9.	0.8	29
66	Corneal Endothelial Findings in a Czech Patient with Compound Heterozygous Mutations inKERA. Ophthalmic Genetics, 2014, 35, 252-254.	1.2	8
67	Crohn's disease: Is there a place for neurological screening?. Scandinavian Journal of Gastroenterology, 2014, 49, 173-176.	1.5	12
68	Macular corneal dystrophy and associated corneal thinning. Eye, 2014, 28, 1201-1205.	2.1	11
69	Severe retinal degeneration in women with a c.2543del mutation in ORF15 of the RPGR gene. Molecular Vision, 2014, 20, 1307-17.	1.1	9
70	Further Genetic and Clinical Insights of Posterior Polymorphous Corneal Dystrophy 3. JAMA Ophthalmology, 2013, 131, 1296.	2.5	35
71	Novel OPA1 missense mutation in a family with optic atrophy and severe widespread neurological disorder. Acta Ophthalmologica, 2013, 91, e225-31.	1.1	39
72	Descemet membrane endothelial keratoplasty with a stromal rim in the treatment of posterior polymorphous corneal dystrophy. Indian Journal of Ophthalmology, 2012, 60, 59.	1.1	8

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73	Changes in lysyl oxidase (LOX) distribution and its decreased activity in keratoconus corneas. Experimental Eye Research, 2012, 104, 74-81.	2.6	83
74	High Prevalence of Posterior Polymorphous Corneal Dystrophy in the Czech Republic; Linkage Disequilibrium Mapping and Dating an Ancestral Mutation. PLoS ONE, 2012, 7, e45495.	2.5	24
75	Keratoconus in 18 pairs of twins. Acta Ophthalmologica, 2012, 90, e482-6.	1.1	102
76	Molecular genetic cause of X-linked retinitis pigmentosa in a Czech family. Acta Ophthalmologica, 2011, 89, e213-e215.	1.1	2
77	Recurrence of posterior polymorphous corneal dystrophy is caused by the overgrowth of the original diseased host endothelium. Histochemistry and Cell Biology, 2011, 136, 93-101.	1.7	10
78	Large Proteoglycan Complexes and Disturbed Collagen Architecture in the Corneal Extracellular Matrix of Mucopolysaccharidosis Type VII (Sly Syndrome). , 2011, 52, 6720.		19
79	Variable ocular phenotypes of posterior polymorphous corneal dystrophy caused by mutations in theZEB1gene. Ophthalmic Genetics, 2010, 31, 230-234.	1.2	32
80	Descemet membrane endothelial keratoplasty with a stromal rim (DMEK-S). British Journal of Ophthalmology, 2010, 94, 909-914.	3.9	81
81	Evidence for Keratoconus Susceptibility Locus on Chromosome 14. JAMA Ophthalmology, 2010, 128, 1191.	2.4	41
82	Role of matrix metalloproteinases in recurrent corneal melting. Experimental Eye Research, 2010, 90, 583-590.	2.6	33
83	Recurrence of posterior polymorphous corneal dystrophy is caused by the overgrowth of the original diseased endothelium. Acta Ophthalmologica, 2010, 88, 0-0.	1.1	0
84	Changes in the localization of collagens IV and VIII in corneas obtained from patients with posterior polymorphous corneal dystrophy. Experimental Eye Research, 2009, 88, 945-952.	2.6	19
85	Sequencing of the CHST6 gene in Czech macular corneal dystrophy patients supports the evidence of a founder mutation. British Journal of Ophthalmology, 2008, 92, 265-267.	3.9	15
86	Phenotype Associated with the H626P Mutation and Other Changes in the <i>TGFBI</i> Gene in Czech Families. Ophthalmic Research, 2008, 40, 105-108.	1.9	10
87	British family with early-onset Fuchs' endothelial corneal dystrophy associated with p.L450W mutation in the COL8A2 gene. British Journal of Ophthalmology, 2007, 91, 1717-1718.	3.9	33
88	Mucolipidosis IV: Report of a Case with Ocular Restricted Phenotype Caused by Leaky Splice Mutation. American Journal of Ophthalmology, 2007, 143, 663-671.e2.	3.3	24
89	Immunohistochemical characterization of cytokeratins in the abnormal corneal endothelium of posterior polymorphous corneal dystrophy patients. Experimental Eye Research, 2007, 84, 680-686.	2.6	53
90	NovelSLC4A11mutations in patients with recessive congenital hereditary endothelial dystrophy (CHED2). Human Mutation, 2007, 28, 522-523.	2.5	80

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91	Novel mutations in theZEB1 gene identified in Czech and British patients with posterior polymorphous corneal dystrophy. Human Mutation, 2007, 28, 638-638.	2.5	67
92	Molecular analysis of the VSX1 gene in familial keratoconus. Molecular Vision, 2007, 13, 1887-91.	1.1	38
93	Study of p.N247S KERA mutation in a British family with cornea plana. Molecular Vision, 2007, 13, 1339-47.	1.1	17
94	Differential immunogold localisation of sulphated and unsulphated keratan sulphate proteoglycans in normal and macular dystrophy cornea using sulphation motif-specific antibodies. Histochemistry and Cell Biology, 2006, 127, 115-120.	1.7	25
95	Posterior Polymorphous Corneal Dystrophy in Czech Families Maps to Chromosome 20 and Excludes theVSX1Gene. , 2005, 46, 4480.		67
96	Changes in the Î±1 - Î±6 collagen IV chains in the corneas of posterior polymorphous corneal dystrophy patients. Acta Ophthalmologica, 0, 85, 0-0.	0.3	0