

Lubica Dudakova

List of Publications by Year in descending order

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Version: 2024-02-01

46
papers

781
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687363

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580821

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#	ARTICLE	IF	CITATIONS
1	Posterior corneal vesicles are not associated with the genetic variants that cause posterior polymorphous corneal dystrophy. <i>Acta Ophthalmologica</i> , 2022, 100, .	1.1	3
2	A multi-ethnic genome-wide association study implicates collagen matrix integrity and cell differentiation pathways in keratoconus. <i>Communications Biology</i> , 2021, 4, 266.	4.4	36
3	Non-Penetrance for Ocular Phenotype in Two Individuals Carrying Heterozygous Loss-of-Function ZEB1 Alleles. <i>Genes</i> , 2021, 12, 677.	2.4	3
4	Comprehensive phenotypic and functional analysis of dominant and recessive <i>FOXE3</i> alleles in ocular developmental disorders. <i>Human Molecular Genetics</i> , 2021, 30, 1591-1606.	2.9	6
5	Novel disease-causing variants and phenotypic features of X-linked megalocornea. <i>Acta Ophthalmologica</i> , 2021, , .	1.1	1
6	Clinical and Genetic Study of X-Linked Juvenile Retinoschisis in the Czech Population. <i>Genes</i> , 2021, 12, 1816.	2.4	4
7	Should Patients with Kearns-Sayre Syndrome and Corneal Endothelial Failure Be Genotyped for a TCF4 Trinucleotide Repeat, Commonly Associated with Fuchs Endothelial Corneal Dystrophy?. <i>Genes</i> , 2021, 12, 1918.	2.4	0
8	CUGC for posterior polymorphous corneal dystrophy (PPCD). <i>European Journal of Human Genetics</i> , 2020, 28, 126-131.	2.8	4
9	Hereditary hyperferritinemia-cataract syndrome in three Czech families: molecular genetic testing and clinical implications. <i>Journal of AAPOS</i> , 2020, 24, 352.e1-352.e5.	0.3	2
10	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. <i>Genetics in Medicine</i> , 2020, 22, 1235-1246.	2.4	92
11	Pseudodominant Nanophthalmos in a Roma Family Caused by a Novel PRSS56 Variant. <i>Journal of Ophthalmology</i> , 2020, 2020, 1-9.	1.3	3
12	A novel missense mutation in <i>LIM2</i> causing isolated autosomal dominant congenital cataract. <i>Ophthalmic Genetics</i> , 2020, 41, 131-134.	1.2	8
13	Brittle cornea syndrome: Disease-causing mutations in ZNF469 and two novel variants identified in a patient followed for 26 years. <i>Biomedical Papers of the Medical Faculty of the University Palacky&#x0301;, Olomouc, Czechoslovakia</i> , 2020, 164, 183-188.	0.6	6
14	IPSC-Derived Corneal Endothelial-like Cells Act as an Appropriate Model System to Assess the Impact of <i>SLC4A11</i> Variants on Pre-mRNA Splicing. , 2019, 60, 3084.		18
15	Paraproteinemic keratopathy associated with monoclonal gammopathy of undetermined significance (<sc>MGUS</sc>): clinical findings in twelve patients including recurrence after keratoplasty. <i>Acta Ophthalmologica</i> , 2019, 97, e987-e992.	1.1	13
16	The utility of massively parallel sequencing for posterior polymorphous corneal dystrophy type 3 molecular diagnosis. <i>Experimental Eye Research</i> , 2019, 182, 160-166.	2.6	8
17	Coincidental Occurrence of Schnyder Corneal Dystrophy and Posterior Polymorphous Corneal Dystrophy Type 3. <i>Cornea</i> , 2019, 38, 758-760.	1.7	3
18	Congenital fibrosis of the extraocular muscles in a Czech family and its molecular genetic cause. <i>Ceska A Slovenska Neurologie A Neurochirurgie</i> , 2019, 82/115, 561-566.	0.1	0

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19	Molecular Genetic Cause of Achromatopsia in Two Patients of Czech Origin. <i>Ceska A Slovenska Oftalmologie</i> , 2019, 75, 272-276.	0.2	0
20	Antisense Therapy for a Common Corneal Dystrophy Ameliorates TCF4 Repeat Expansion-Mediated Toxicity. <i>American Journal of Human Genetics</i> , 2018, 102, 528-539.	6.2	59
21	Ectopic GRHL2 Expression Due to Non-coding Mutations Promotes Cell State Transition and Causes Posterior Polymorphous Corneal Dystrophy 4. <i>American Journal of Human Genetics</i> , 2018, 102, 447-459.	6.2	45
22	SD-OCT imaging as a valuable tool to support molecular genetic diagnostics of Usher syndrome type 1. <i>Journal of AAPOS</i> , 2018, 22, 312-314.e3.	0.3	2
23	Familial Limbal Stem Cell Deficiency: Clinical, Cytological and Genetic Characterization. <i>Stem Cell Reviews and Reports</i> , 2018, 14, 148-151.	5.6	4
24	Analysis of <i>KERA</i> in four families with cornea plana identifies two novel mutations. <i>Acta Ophthalmologica</i> , 2018, 96, e87-e91.	1.1	4
25	Schnyder corneal dystrophy and associated phenotypes caused by novel and recurrent mutations in the <i>UBIAD1</i> gene. <i>BMC Ophthalmology</i> , 2018, 18, 250.	1.4	9
26	<i>OPA1</i> analysis in an international series of probands with bilateral optic atrophy. <i>Acta Ophthalmologica</i> , 2017, 95, 363-369.	1.1	7
27	Segregation of a novel p.(Ser270Tyr) MAF mutation and p.(Tyr56âˆ’) CRYGD variant in a family with dominantly inherited congenital cataracts. <i>Molecular Biology Reports</i> , 2017, 44, 435-440.	2.3	9
28	Copper in Keratoconic Corneas. <i>Cornea</i> , 2017, 36, e14-e14.	1.7	5
29	Replication of SNP associations with keratoconus in a Czech cohort. <i>PLoS ONE</i> , 2017, 12, e0172365.	2.5	22
30	Active transforming growth factor- β 2 in the aqueous humor of posterior polymorphous corneal dystrophy patients. <i>PLoS ONE</i> , 2017, 12, e0175509.	2.5	5
31	Leber Hereditary Optic Neuropathy. <i>Ceska A Slovenska Neurologie A Neurochirurgie</i> , 2017, 80/113, 534-544.	0.1	2
32	Early detection of bilateral cataracts in utero may represent a manifestation of severe congenital disease. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1843-1848.	1.2	6
33	Novel <i>TGFBI</i> mutation p.(Leu558Arg) in a lattice corneal dystrophy patient. <i>Ophthalmic Genetics</i> , 2016, 37, 473-474.	1.2	10
34	Phenotypic features of <i>CRB1</i> -associated early-onset severe retinal dystrophy and the different molecular approaches to identifying the disease-causing variants. <i>Graefe's Archive for Clinical and Experimental Ophthalmology</i> , 2016, 254, 1833-1839.	1.9	19
35	Autosomal-Dominant Corneal Endothelial Dystrophies <i>CHED1</i> and <i>PPCD1</i> Are Allelic Disorders Caused by Non-coding Mutations in the Promoter of <i>OVOL2</i> . <i>American Journal of Human Genetics</i> , 2016, 98, 75-89.	6.2	70
36	Heterozygous deletions at the <i>ZEB1</i> locus verify haploinsufficiency as the mechanism of disease for posterior polymorphous corneal dystrophy type 3. <i>European Journal of Human Genetics</i> , 2016, 24, 985-991.	2.8	33

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37	The presence of lysyl oxidase-like enzymes in human control and keratoconic corneas. <i>Histology and Histopathology</i> , 2016, 31, 63-71.	0.7	11
38	Detailed Assessment of Renal Function in a Proband with Harboyan Syndrome Caused by a Novel Homozygous <i>SLC4A11</i> Nonsense Mutation. <i>Ophthalmic Research</i> , 2015, 53, 30-35.	1.9	9
39	Is copper imbalance an environmental factor influencing keratoconus development?. <i>Medical Hypotheses</i> , 2015, 84, 518-524.	1.5	10
40	Validation of rs2956540:G>C and rs3735520:G>A association with keratoconus in a population of European descent. <i>European Journal of Human Genetics</i> , 2015, 23, 1581-1583.	2.8	34
41	Identification of Six Novel Mutations in <i>ZEB1</i> and Description of the Associated Phenotypes in Patients with Posterior Polymorphous Corneal Dystrophy 3. <i>Annals of Human Genetics</i> , 2015, 79, 1-9.	0.8	29
42	Corneal Endothelial Findings in a Czech Patient with Compound Heterozygous Mutations in <i>KERA</i> . <i>Ophthalmic Genetics</i> , 2014, 35, 252-254.	1.2	8
43	Macular corneal dystrophy and associated corneal thinning. <i>Eye</i> , 2014, 28, 1201-1205.	2.1	11
44	The impairment of lysyl oxidase in keratoconus and in keratoconus-associated disorders. <i>Journal of Neural Transmission</i> , 2013, 120, 977-982.	2.8	33
45	Changes in lysyl oxidase (LOX) distribution and its decreased activity in keratoconus corneas. <i>Experimental Eye Research</i> , 2012, 104, 74-81.	2.6	83
46	The OV-TL 12/30 Clone of Anti-cytokeratin 7 Antibody as a New Marker of Corneal Conjunctivalization in Patients with Limbal Stem Cell Deficiency. , 2011, 52, 5892.		31