Yelena Bykhovskaya

List of Publications by Year
in descending order

37	2,776	257450 24	395702 33
papers	citations	h-index	g-index

39	39	39	2755
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Update on the genetics of keratoconus. Experimental Eye Research, 2021, 202, 108398.	2.6	36
2	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
3	Association of Genetic Variation With Keratoconus. JAMA Ophthalmology, 2020, 138, 174.	2.5	34
4	PPIP5K2 and PCSK1 are Candidate Genetic Contributors to Familial Keratoconus. Scientific Reports, 2019, 9, 19406.	3.3	34
5	Corneal Perforation After Corneal Cross-Linking in Keratoconus Associated With Potentially Pathogenic ZNF469 Mutations. Cornea, 2019, 38, 1033-1039.	1.7	13
6	Differential Expression of Coding and Long Noncoding RNAs in Keratoconus-Affected Corneas. , 2018, 59, 2717.		45
7	Cross-ancestry genome-wide association analysis of corneal thickness strengthens link between complex and Mendelian eye diseases. Nature Communications, 2018, 9, 1864.	12.8	63
8	TSC1 Mutations in Keratoconus Patients With or Without Tuberous Sclerosis. , 2017, 58, 6462.		10
9	Linkage Analysis of High-density SNPs Confirms Keratoconus Locus at 5q Chromosomal Region. Ophthalmic Genetics, 2016, 37, 1-2.	1.2	18
10	Abnormal Regulation of Extracellular Matrix and Adhesion Molecules in Corneas of Patients with Keratoconus. International Journal of Keratoconus and Ectatic Corneal Diseases, 2016, 5, 63-70.	0.5	25
11	Genetics in Keratoconus: where are we?. Eye and Vision (London, England), 2016, 3, 16.	3.0	78
12	Pseudouridine synthase 1 deficient mice, a model for Mitochondrial Myopathy with Sideroblastic Anemia, exhibit muscle morphology and physiology alterations. Scientific Reports, 2016, 6, 26202.	3.3	26
13	Independent Origin of c.57 Câ€‰›â€‰T Mutation in MIR184 Associated with Inherited Corneal and Lens Abnormalities. Ophthalmic Genetics, 2015, 36, 95-97.	1.2	6
14	C.57 C > T Mutation in MIR 184 is Responsible for Congenital Cataracts and Corneal Abnormalities in a Five-generation Family from Galicia, Spain. Ophthalmic Genetics, 2015, 36, 244-247.	1.2	37
15	Genome-wide association analyses identify multiple loci associated with central corneal thickness and keratoconus. Nature Genetics, 2013, 45, 155-163.	21.4	269
16	An Association Between the Calpastatin (CAST) Gene and Keratoconus. Cornea, 2013, 32, 696-701.	1.7	38
17	Genetic Association ofCOL5A1Variants in Keratoconus Patients Suggests a Complex Connection between Corneal Thinning and Keratoconus. , 2013, 54, 2696.		73
18	Variation in the Lysyl Oxidase (LOX) Gene Is Associated with Keratoconus in Family-Based and Case-Control Studies. , 2012, 53, 4152.		116

#	Article	IF	Citations
19	A genome-wide association study identifies a potential novel gene locus for keratoconus, one of the commonest causes for corneal transplantation in developed countries. Human Molecular Genetics, 2012, 21, 421-429.	2.9	108
20	Association of Polymorphisms in the Hepatocyte Growth Factor Gene Promoter with Keratoconus. , 2011, 52, 8514.		114
21	Phenotypic expression of maternally inherited deafness is affected by RNA modification and cytoplasmic ribosomal proteins. Molecular Genetics and Metabolism, 2009, 97, 297-304.	1.1	9
22	Pleiotropic effects and compensation mechanisms determine tissue specificity in mitochondrial myopathy and sideroblastic anemia (MLASA). Molecular Genetics and Metabolism, 2007, 91, 148-156.	1.1	18
23	Mutation in TRMU Related to Transfer RNA Modification Modulates the Phenotypic Expression of the Deafness-Associated Mitochondrial 12S Ribosomal RNA Mutations. American Journal of Human Genetics, 2006, 79, 291-302.	6.2	212
24	Human TRMU encoding the mitochondrial 5-methylaminomethyl-2-thiouridylate-methyltransferase is a putative nuclear modifier gene for the phenotypic expression of the deafness-associated 12S rRNA mutations. Biochemical and Biophysical Research Communications, 2006, 342, 1130-1136.	2.1	31
25	Mitochondrial Myopathy, Sideroblastic Anemia, and Lactic Acidosis: An Autosomal Recessive Syndrome in Persian Jews Caused by a Mutation in the PUS1 Gene. Journal of Child Neurology, 2005, 20, 449-452.	1.4	67
26	Mitochondrial Myopathy and Sideroblastic Anemia (MLASA). Journal of Biological Chemistry, 2005, 280, 19823-19828.	3.4	118
27	Gene responsible for mitochondrial myopathy and sideroblastic anemia (MSA) maps to chromosome 12q24.33. American Journal of Medical Genetics Part A, 2004, 127A, 44-49.	2.4	19
28	Missense Mutation in Pseudouridine Synthase 1 (PUS1) Causes Mitochondrial Myopathy and Sideroblastic Anemia (MLASA). American Journal of Human Genetics, 2004, 74, 1303-1308.	6.2	274
29	Human mitochondrial transcription factor B1 as a modifier gene for hearing loss associated with the mitochondrial A1555G mutation. Molecular Genetics and Metabolism, 2004, 82, 27-32.	1.1	75
30	Phenotype of non-syndromic deafness associated with the mitochondrial A1555G mutation is modulated by mitochondrial RNA modifying enzymes MTO1 and GTPBP3. Molecular Genetics and Metabolism, 2004, 83, 199-206.	1.1	64
31	A nuclear-mitochondrial DNA interaction affecting hearing impairment in mice. Nature Genetics, 2001, 27, 191-194.	21.4	153
32	Modifier locus for mitochondrial DNA disease: Linkage and linkage disequilibrium mapping of a nuclear modifier gene for maternally inherited deafness. Genetics in Medicine, 2001, 3, 177-180.	2.4	56
33	Candidate Locus for a Nuclear Modifier Gene for Maternally Inherited Deafness. American Journal of Human Genetics, 2000, 66, 1905-1910.	6.2	103
34	Mitochondrial A7445G mutation in two pedigrees with palmoplantar keratoderma and deafness. American Journal of Medical Genetics Part A, 1998, 75, 179-185.	2.4	142
35	Evidence for complex nuclear inheritance in a pedigree with nonsyndromic deafness due to a homoplasmic mitochondrial mutation. American Journal of Medical Genetics Part A, 1998, 77, 421-426.	2.4	71
36	Hearing loss due to the mitochondrial A1555G mutation in Italian families. American Journal of Medical Genetics Part A, 1998, 79, 388-391.	2.4	81

#	Article	IF	Citations
37	Temporal bone analysis of patients with presbycusis reveals high frequency of mitochondrial mutations. Hearing Research, 1997, 110, 147-154.	2.0	102