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	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
2	Genome-wide association analyses identify multiple loci associated with central corneal thickness and keratoconus. Nature Genetics, 2013, 45, 155-163.	21.4	269
3	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
4	A nuclear-mitochondrial DNA interaction affecting hearing impairment in mice. Nature Genetics, 2001, 27, 191-194.	21.4	153
5	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
6	Mitochondrial Myopathy and Sideroblastic Anemia (MLASA). Journal of Biological Chemistry, 2005, 280, 19823-19828.	3.4	118
7	Variation in the Lysyl Oxidase (LOX) Gene Is Associated with Keratoconus in Family-Based and Case-Control Studies. , 2012, 53, 4152.		116
8	Association of Polymorphisms in the Hepatocyte Growth Factor Gene Promoter with Keratoconus. , 2011, 52, 8514.		114
9	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
10	Candidate Locus for a Nuclear Modifier Gene for Maternally Inherited Deafness. American Journal of Human Genetics, 2000, 66, 1905-1910.	6.2	103
11	Temporal bone analysis of patients with presbycusis reveals high frequency of mitochondrial mutations. Hearing Research, 1997, 110, 147-154.	2.0	102
12	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
13	Genetics in Keratoconus: where are we?. Eye and Vision (London, England), 2016, 3, 16.	3.0	78
14	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
15	Genetic Association ofCOL5A1Variants in Keratoconus Patients Suggests a Complex Connection between Corneal Thinning and Keratoconus. , 2013, 54, 2696.		73
16	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
17	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
18	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

#	Article	IF	Citations
19	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
20	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
21	Differential Expression of Coding and Long Noncoding RNAs in Keratoconus-Affected Corneas. , 2018, 59, 2717.		45
22	An Association Between the Calpastatin (CAST) Gene and Keratoconus. Cornea, 2013, 32, 696-701.	1.7	38
23	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
24	Update on the genetics of keratoconus. Experimental Eye Research, 2021, 202, 108398.	2.6	36
25	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
26	PPIP5K2 and PCSK1 are Candidate Genetic Contributors to Familial Keratoconus. Scientific Reports, 2019, 9, 19406.	3.3	34
27	Association of Genetic Variation With Keratoconus. JAMA Ophthalmology, 2020, 138, 174.	2.5	34
28	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
29	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
30	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
31	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
32	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
33	Linkage Analysis of High-density SNPs Confirms Keratoconus Locus at 5q Chromosomal Region. Ophthalmic Genetics, 2016, 37, 1-2.	1.2	18
34	Corneal Perforation After Corneal Cross-Linking in Keratoconus Associated With Potentially Pathogenic ZNF469 Mutations. Cornea, 2019, 38, 1033-1039.	1.7	13
35	TSC1 Mutations in Keratoconus Patients With or Without Tuberous Sclerosis. , 2017, 58, 6462.		10
36	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

#	Article	IF	Citations
37	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