Elena A Bliznetz

List of Publications by Year
in descending order

19	219	1163117 8	1058476 14
papers	citations	h-index	g-index

28	28	28	324
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genetic analysis of autosomal recessive osteopetrosis in Chuvashiya: the unique splice site mutation in TCIRG1 gene spread by the founder effect. European Journal of Human Genetics, 2009, 17, 664-672.	2.8	35
2	Update of the GJB2/DFNB1 mutation spectrum in Russia: a founder Ingush mutation del(GJB2-D13S175) is the most frequent among other large deletions. Journal of Human Genetics, 2017, 62, 789-795.	2.3	29
3	Population distribution and ancestry of the cancer protective MDM2 SNP285 (rs117039649). Oncotarget, 2014, 5, 8223-8234.	1.8	22
4	Changes in the connexin 26 gene (GJB2) in Russian patients with hearing loss: Results of long-term molecular diagnostics of hereditary nonsyndromic hearing loss. Russian Journal of Genetics, 2012, 48, 101-112.	0.6	18
5	Molecular-genetic causes for the high frequency of phenylketonuria in the population from the North Caucasus. PLoS ONE, 2018, 13, e0201489.	2.5	15
6	Clinical features of hearing loss caused by STRC gene deletions/mutations in Russian population. International Journal of Pediatric Otorhinolaryngology, 2020, 138, 110247.	1.0	12
7	New recurrent large deletion, encompassing both GJB2 and GB6 genes, results in isolated sensorineural hearing impairment with autosomal recessive mode of inheritance. Russian Journal of Genetics, 2014, 50, 415-420.	0.6	9
8	Hidden X Chromosomal Mosaicism in a 46,XX Male. Sexual Development, 2009, 3, 183-187.	2.0	8
9	Molecular Genetic Causes and Clinical Description of Branchio-Oto-renal Syndrome. Russian Journal of Genetics, 2019, 55, 630-638.	0.6	6
10	Hereditary angioedema. Clinical guidelines. (D84.1). Russian Journal of Allergy, 2021, 18, 77-114.	0.2	4
11	GENETIC FACTORS OF MALE INFERTILITY, THEIR COMBINATIONS AND THE SPERMATOLOGICAL CHARACTERISTICS OF MEN WITH FERTILITY FAILURES. Andrologia I Genital'naa Hirurgia, 2018, 19, 40-51.	0.2	4
12	A common founder effect of the splice site variant c.-23â€‰+â€‰1Gâ€‰>â€‰A in GJB2 gene causing autosomal recessive deafness 1A (DFNB1A) in Eurasia. Human Genetics, 2022, 141, 697-707.	3.8	4
13	Spectrum of the GJB2 mutations in Belarussian patients with hearing loss. Findings of pilot genetic screening of hearing impairment in newborns. Russian Journal of Genetics, 2014, 50, 191-197.	0.6	3
14	Phenotype in a patient with p.D50N mutation in GJB2 gene resemble both KID and Clouston syndromes. International Journal of Pediatric Otorhinolaryngology, 2016, 81, 10-14.	1.0	3
15	Results of molecular genetic testing in Russian patients with Pendred syndrome and allelic disorders. Russian Journal of Genetics, 2017, 53, 128-138.	0.6	2
16	Hereditary Angioedema (HAE) with a mutation in the plasminogen gene: a retrospective study of a cohort of 14 patients from Russia. Russian Journal of Allergy, 2021, 18, 5-19.	0.2	1
17	Genetic examination of children with hearing impairment in the astrakhan region. Russian Otorhinolaryngology, 2020, 19, 44-50.	0.5	1
18	DNA copy number analysis of the DFNB1 hereditary hearing loss locus. Russian Journal of Genetics, 2017, 53, 795-803.	0.6	0

#	Article	IF	Citations
19	Beals syndrome (congenital contractural arachnodactyly) in children: Clinical symptoms, diagnosis, treatment, and prevention. Rossiyskiy Vestnik Perinatologii I Pediatrii, 2016, 61, 47-51.	0.3	0