

Elena A Bliznetz

List of Publications by Year in descending order

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Version: 2024-02-01

19
papers

219
citations

1163117

8
h-index

1058476

14
g-index

28
all docs

28
docs citations

28
times ranked

324
citing authors

#	ARTICLE	IF	CITATIONS
1	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. <i>Human Genetics</i> , 2022, 141, 697-707.	3.8	4
2	Hereditary Angioedema (HAE) with a mutation in the plasminogen gene: a retrospective study of a cohort of 14 patients from Russia. <i>Russian Journal of Allergy</i> , 2021, 18, 5-19.	0.2	1
3	Hereditary angioedema. Clinical guidelines. (D84.1). <i>Russian Journal of Allergy</i> , 2021, 18, 77-114.	0.2	4
4	Clinical features of hearing loss caused by STRC gene deletions/mutations in Russian population. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2020, 138, 110247.	1.0	12
5	Genetic examination of children with hearing impairment in the astrakhan region. <i>Russian Otorhinolaryngology</i> , 2020, 19, 44-50.	0.5	1
6	Molecular Genetic Causes and Clinical Description of Branchio-Oto-renal Syndrome. <i>Russian Journal of Genetics</i> , 2019, 55, 630-638.	0.6	6
7	Molecular-genetic causes for the high frequency of phenylketonuria in the population from the North Caucasus. <i>PLoS ONE</i> , 2018, 13, e0201489.	2.5	15
8	GENETIC FACTORS OF MALE INFERTILITY, THEIR COMBINATIONS AND THE SPERMATOLOGICAL CHARACTERISTICS OF MEN WITH FERTILITY FAILURES. <i>Andrologia I Genital'naa Hirurgia</i> , 2018, 19, 40-51.	0.2	4
9	Update of the GJB2/DFNB1 mutation spectrum in Russia: a founder Ingush mutation del(GJB2-D13S175) is the most frequent among other large deletions. <i>Journal of Human Genetics</i> , 2017, 62, 789-795.	2.3	29
10	Results of molecular genetic testing in Russian patients with Pendred syndrome and allelic disorders. <i>Russian Journal of Genetics</i> , 2017, 53, 128-138.	0.6	2
11	DNA copy number analysis of the DFNB1 hereditary hearing loss locus. <i>Russian Journal of Genetics</i> , 2017, 53, 795-803.	0.6	0
12	Phenotype in a patient with p.D50N mutation in GJB2 gene resemble both KID and Clouston syndromes. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2016, 81, 10-14.	1.0	3
13	Beals syndrome (congenital contractural arachnodactyly) in children: Clinical symptoms, diagnosis, treatment, and prevention. <i>Rossiyskiy Vestnik Perinatologii I Pediatrii</i> , 2016, 61, 47-51.	0.3	0
14	Spectrum of the GJB2 mutations in Belarussian patients with hearing loss. Findings of pilot genetic screening of hearing impairment in newborns. <i>Russian Journal of Genetics</i> , 2014, 50, 191-197.	0.6	3
15	New recurrent large deletion, encompassing both GJB2 and GB6 genes, results in isolated sensorineural hearing impairment with autosomal recessive mode of inheritance. <i>Russian Journal of Genetics</i> , 2014, 50, 415-420.	0.6	9
16	Population distribution and ancestry of the cancer protective MDM2 SNP285 (rs117039649). <i>Oncotarget</i> , 2014, 5, 8223-8234.	1.8	22
17	Changes in the connexin 26 gene (GJB2) in Russian patients with hearing loss: Results of long-term molecular diagnostics of hereditary nonsyndromic hearing loss. <i>Russian Journal of Genetics</i> , 2012, 48, 101-112.	0.6	18
18	Genetic analysis of autosomal recessive osteopetrosis in Chuvashiya: the unique splice site mutation in TCIRG1 gene spread by the founder effect. <i>European Journal of Human Genetics</i> , 2009, 17, 664-672.	2.8	35

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19	Hidden X Chromosomal Mosaicism in a 46,XX Male. <i>Sexual Development</i> , 2009, 3, 183-187.	2.0	8