BÃ©la Melegh

List of Publications by Year
in descending order

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Version: 2024-02-01

64 papers	3,476 citations	430874 18 h-index	175258 52 g-index
72 all docs	72 docs citations	72 times ranked	6335 citing authors

#	Article	IF	Citations
1	Clinical and genetic spectrum of a large cohort of patients with Î´-sarcoglycan muscular dystrophy. Brain, 2022, 145, 596-606.	7.6	11
2	The need for recognition of core professional groups in genetics healthcare services in Europe. European Journal of Human Genetics, 2022, 30, 639-640.	2.8	2
3	Genome-Wide Marker Data-Based Comparative Population Analysis of Szeklers From Korond, Transylvania, and From Transylvania Living Non-Szekler Hungarians. Frontiers in Genetics, 2022, 13, 841769.	2.3	1
4	Mapping genomic loci implicates genes and synaptic biology in schizophrenia. Nature, 2022, 604, 502-508.	27.8	929
5	TUBB4B gene mutation in Leber phenotype of congenital amaurosis syndrome associated with early-onset deafness. European Journal of Medical Genetics, 2022, 65, 104471.	1.3	3
6	Ãœber die Notwendigkeit der Anerkennung von sog. Kernberufsgruppen innerhalb der genetischen Gesundheitsversorgung in Europa. Medizinische Genetik, 2022, 34, 81-83.	0.2	1
7	Erratum zu: Ãœber die Notwendigkeit der Anerkennung von sog. Kernberufsgruppen innerhalb der genetischen Gesundheitsversorgung in Europa. Medizinische Genetik, 2022, 34, 189-191.	0.2	0
8	Xp11.2 Duplication in Females: Unique Features of a Rare Copy Number Variation. Frontiers in Genetics, 2021, 12, 635458.	2.3	2
9	Genotype-Phenotype Associations in Patients With Type-1, Type-2, and Atypical NF1 Microdeletions. Frontiers in Genetics, 2021, 12, 673025.	2.3	11
10	Genome-wide autosomal, mtDNA, and Y chromosome analysis of King Bela III of the Hungarian Arpad dynasty. Scientific Reports, 2021, 11, 19210.	3.3	9
11	Whole Exome Sequencing in a Series of Patients with a Clinical Diagnosis of Tuberous Sclerosis Not Confirmed by Targeted TSC1/TSC2 Sequencing. Genes, 2021, 12, 1401.	2.4	4
12	Intrafamilial variability of limb-girdle muscular dystrophy, LGMD1D type. European Journal of Medical Genetics, 2020, 63, 103655.	1.3	10
13	Mutation spectrum of the SCN1A gene in a Hungarian population with epilepsy. Seizure: the Journal of the British Epilepsy Association, 2020, 74, 8-13.	2.0	8
14	Investigating the genetic characteristics of the Csangos, a traditionally Hungarian speaking ethnic group residing in Romania. Journal of Human Genetics, 2020, 65, 1093-1103.	2.3	2
15	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	7.6	45
16	Conversion of individuals at risk for spinocerebellar ataxia types 1, 2, 3, and 6 to manifest ataxia (RISCA): a longitudinal cohort study. Lancet Neurology, The, 2020, 19, 738-747.	10.2	41
17	Mass Spectrometric Analysis of L-carnitine and its Esters: Potential Biomarkers of Disturbances in Carnitine Homeostasis. Current Molecular Medicine, 2020, 20, 336-354.	1.3	15
18	Revealing the Genetic Impact of the Ottoman Occupation on Ethnic Groups of East-Central Europe and on the Roma Population of the Area. Frontiers in Genetics, 2019, 10, 558.	2.3	9

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19	Possible Phenotypic Consequences of Structural Differences in Idic(15) in a Small Cohort of Patients. International Journal of Molecular Sciences, 2019, 20, 4935.	4.1	1
20	Prediction of Survival With Longâ€Term Disease Progression in Most Common Spinocerebellar Ataxia. Movement Disorders, 2019, 34, 1220-1227.	3.9	14
21	A novel pathogenic variant in TNPO3 in a Hungarian family with limb-girdle muscular dystrophy 1F. European Journal of Medical Genetics, 2019, 62, 103662.	1.3	15
22	Hearing impairment locus heterogeneity and identification of PLS1 as a new autosomal dominant gene in Hungarian Roma. European Journal of Human Genetics, 2019, 27, 869-878.	2.8	10
23	DNA profiling of Hungarian King BÃ©la III and other skeletal remains originating from the Royal Basilica of SzÃ©kesfehÃ©rvÃ¡r. Archaeological and Anthropological Sciences, 2019, 11, 1345-1357.	1.8	24
24	Age-Related Hearing Impairment Associated NAT2, GRM7, GRHL2 Susceptibility Gene Polymorphisms and Haplotypes in Roma and Hungarian Populations. Pathology and Oncology Research, 2019, 25, 1349-1355.	1.9	5
25	Role of carnitine and its derivatives in the development and management of type 2 diabetes. Nutrition and Diabetes, 2018, 8, 8.	3.2	121
26	Survival in patients with spinocerebellar ataxia types 1, 2, 3, and 6 (EUROSCA): a longitudinal cohort study. Lancet Neurology, The, 2018, 17, 327-334.	10.2	69
27	Small supernumerary marker chromosome 15 and a ring chromosome 15 associated with a 15q26.3 deletion excluding the IGF1R gene. American Journal of Medical Genetics, Part A, 2018, 176, 443-449.	1.2	7
28	Revealing the impact of the Caucasus region on the genetic legacy of Romani people from genome-wide data. PLoS ONE, 2018, 13, e0202890.	2.5	5
29	Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. Journal of Neurology, 2018, 265, 2040-2051.	3.6	34
30	Marked Differences of Haplotype Tagging SNP Distribution, Linkage, and Haplotype Profile of APOA5 Gene in Roma Population Samples. Pathology and Oncology Research, 2017, 23, 853-861.	1.9	10
31	CYP2C9 and VKORC1 in therapeutic dosing and safety of acenocoumarol treatment: implication for clinical practice in Hungary. Environmental Toxicology and Pharmacology, 2017, 56, 282-289.	4.0	4
32	Novel phenotypic variant in the MYH7 spectrum due to a stop-loss mutation in the C-terminal region: a case report. BMC Medical Genetics, 2017, 18, 105.	2.1	8
33	On the distribution of intranuclear and cytoplasmic aggregates in the brainstem of patients with spinocerebellar ataxia type 2 and 3. Brain Pathology, 2017, 27, 345-355.	4.1	36
34	Refining the South Asian Origin of the Romani people. BMC Genetics, 2017, 18, 82.	2.7	26
35	Initiating an undiagnosed diseases program in the Western Australian public health system. Orphanet Journal of Rare Diseases, 2017, 12, 83.	2.7	24
36	Kleefstra syndrome in Hungarian patients: additional symptoms besides the classic phenotype. Molecular Cytogenetics, 2016, 9, 22.	0.9	14

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37	Evidence for Genetic Overlap Between Schizophrenia and Age at First Birth in Women. JAMA Psychiatry, 2016, 73, 497.	11.0	51
38	Cytochrome P450 Drug Metabolizing Enzymes in Roma Population Samples: Systematic Review of the Literature. Current Medicinal Chemistry, 2016, 23, 3632-3652.	2.4	7
39	Partial tetrasomy of the proximal long arm of chromosome 15 in two patients: the significance of the gene dosage in terms of phenotype. Molecular Cytogenetics, 2015, 8, 41.	0.9	3
40	Marked differences in frequencies of statin therapy relevant SLCO1B1 variants and haplotypes between Roma and Hungarian populations. BMC Genetics, 2015, 16, 108.	2.7	10
41	Genome Sequences of Three Turkey Orthoreovirus Strains Isolated in Hungary. Genome Announcements, 2015, 3, .	0.8	3
42	MARVELD2 (DFNB49) Mutations in the Hearing Impaired Central European Roma Population - Prevalence, Clinical Impact and the Common Origin. PLoS ONE, 2015, 10, e0124232.	2.5	17
43	Identification ofSLC22A5Gene Mutation in a Family with Carnitine Uptake Defect. Case Reports in Genetics, 2015, 2015, 1-5.	0.2	14
44	Towards a European consensus for reporting incidental findings during clinical NGS testing. European Journal of Human Genetics, 2015, 23, 1601-1606.	2.8	85
45	Extreme differences in SLCO1B3 functional polymorphisms in Roma and Hungarian populations. Environmental Toxicology and Pharmacology, 2015, 39, 1246-1251.	4.0	4
46	Undiagnosed Diseases Network International (UDNI): White paper for global actions to meet patient needs. Molecular Genetics and Metabolism, 2015, 116, 223-225.	1.1	69
47	Interethnic variability of CYP4F2 (V433M) in admixed population of Roma and Hungarians. Environmental Toxicology and Pharmacology, 2015, 40, 280-283.	4.0	2
48	Interethnic differences of cytochrome P450 gene polymorphisms may influence outcome of taxane therapy in Roma and Hungarian populations. Drug Metabolism and Pharmacokinetics, 2015, 30, 453-456.	2.2	3
49	Increased prevalence of functional minor allele variants of drug metabolizing CYP2B6 and CYP2D6 genes in Roma population samples. Pharmacological Reports, 2015, 67, 460-464.	3.3	14
50	Phenotypic variability in a Hungarian patient with the 4q21 microdeletion syndrome. Molecular Cytogenetics, 2015, 8, 16.	0.9	11
51	Long-term disease progression in spinocerebellar ataxia types 1, 2, 3, and 6: a longitudinal cohort study. Lancet Neurology, The, 2015, 14, 1101-1108.	10.2	213
52	Interleukins and interleukin receptors in rheumatoid arthritis: Research, diagnostics and clinical implications. World Journal of Orthopedics, 2014, 5, 516.	1.8	60
53	Genetic polymorphisms in promoter and intronic regions of CYP1A2 gene in Roma and Hungarian population samples. Environmental Toxicology and Pharmacology, 2014, 38, 814-820.	4.0	7
54	Deletion of 4q28.3-31.23 in the background of multiple malformations with pulmonary hypertension. Molecular Cytogenetics, 2014, 7, 36.	0.9	15

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55	Ancient human genomes suggest three ancestral populations for present-day Europeans. Nature, 2014, 513, 409-413.	27.8	1,179
56	Marked differences of haplotype tagging SNP distribution, linkage, and haplotype profile of IL23 receptor gene in Roma and Hungarian population samples. Cytokine, 2014, 65, 148-152.	3.2	4
57	Exome sequencing identifies Laing distal myopathy MYH7 mutation in a Roma family previously diagnosed with distal neuronopathy. Neuromuscular Disorders, 2014, 24, 156-161.	0.6	17
58	Interleukin and interleukin receptor gene polymorphisms in inflammatory bowel diseases susceptibility. World Journal of Gastroenterology, 2014, 20, 3208.	3.3	23
59	Reconstructing Roma History from Genome-Wide Data. PLoS ONE, 2013, 8, e58633.	2.5	61
60	Non-syndromic Hearing Impairment in a Hungarian Family with the m.7510T>C Mutation of Mitochondrial tRNASer(UCN) and Review of Published Cases. JIMD Reports, 2012, 9, 105-111.	1.5	3
61	Analysis of mtDNA A3243G mutation frequency in Hungary. Open Medicine (Poland), 2010, 5, 322-328.	1.3	3
62	Tel Hashomer camptodactyly syndrome: 12-year follow-up of a Hungarian patient and review. American Journal of Medical Genetics, Part A, 2005, 135A, 320-323.	1.2	4
63	Molecular epidemiology of human P[8],G9 rotaviruses in Hungary between 1998 and 2001. Journal of Medical Microbiology, 2004, 53, 791-801.	1.8	24
64	Phenotypic manifestations of the OCTN2 V295X mutation: Sudden infant death and carnitine-responsive cardiomyopathy in Roma families. American Journal of Medical Genetics Part A, 2004, 131A, 121-126.	2.4	34