Istvan Balogh

List of Publications by Year in descending order

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84 papers	1,689 citations	361413 20 h-index	330143 37 g-index

95 all docs	95 docs citations	95 times ranked	2538 citing authors

#	Article	IF	Citations
1	Establishing the Mutational Spectrum of Hungarian Patients with Familial Hypercholesterolemia. Genes, 2022, 13, 153.	2.4	4
2	The Role of Exosomes in Cancer Progression. International Journal of Molecular Sciences, 2022, 23, 8.	4.1	23
3	Clinical Aspects of Genetic and Non-Genetic Cardiovascular Risk Factors in Familial Hypercholesterolemia. Genes, 2022, 13, 1158.	2.4	5
4	Enhanced Expression of Human Epididymis Protein 4 (HE4) Reflecting Pro-Inflammatory Status Is Regulated by CFTR in Cystic Fibrosis Bronchial Epithelial Cells. Frontiers in Pharmacology, 2021, 12, 592184.	3.5	10
5	A Comprehensive Analysis of Hungarian MODY Patientsâ€”Part II: Glucokinase MODY Is the Most Prevalent Subtype Responsible for about 70% of Confirmed Cases. Life, 2021, 11, 771.	2.4	2
6	Hereditary and non-hereditary etiologies associated with extensive brain calcification: case series. Metabolic Brain Disease, 2021, 36, 2131-2139.	2.9	1
7	A Comprehensive Analysis of Hungarian MODY Patientsâ€”Part I: Gene Panel Sequencing Reveals Pathogenic Mutations in HNF1A, HNF1B, HNF4A, ABCC8 and INS Genes. Life, 2021, 11, 755.	2.4	4
8	Biochemical and Clinical Effects of Vitamin E Supplementation in Hungarian Smith-Lemli-Opitz Syndrome Patients. Biomolecules, 2021, 11, 1228.	4.0	2
9	Four New Cases of Hypomyelinating Leukodystrophy Associated with the UFM1 c.-155_-153delTCA Founder Mutation in Pediatric Patients of Roma Descent in Hungary. Genes, 2021, 12, 1331.	2.4	5
10	MED13L-related intellectual disability due to paternal germinal mosaicism. Journal of Physical Education and Sports Management, 2021, , mcs.a006124.	1.2	2
11	Non-lupus full-house nephropathyâ€”immune dysregulation as a rare cause of pediatric nephrotic syndrome: Answers. Pediatric Nephrology, 2021, , 1.	1.7	0
12	Non-lupus full-house nephropathyâ€”immune dysregulation as a rare cause of pediatric nephrotic syndrome: Questions. Pediatric Nephrology, 2021, , 1.	1.7	2
13	Altered microRNAs expression levels of sperm and seminal plasma in patients with infertile ejaculates compared with normozoospermic males. Human Fertility, 2020, 23, 246-255.	1.7	19
14	Laboratory biomarkers for lung disease severity and progression in cystic fibrosis. Clinica Chimica Acta, 2020, 508, 277-286.	1.1	11
15	The importance of the multiplex ligation-dependent probe amplification in the identification of a novel two-exon deletion of the NR5A1 gene in a patient with 46,XY differences of sex development. Molecular Biology Reports, 2019, 46, 5595-5601.	2.3	5
16	FBN1 gene mutations in 26 Hungarian patients with suspected Marfan syndrome or related fibrillinopathies. Journal of Biotechnology, 2019, 301, 105-111.	3.8	2
17	Copy number variants detection by microarray and multiplex ligation-dependent probe amplification in congenital heart diseases. Journal of Biotechnology, 2019, 299, 86-95.	3.8	11
18	Work-related neck and upper limb disorders â€“ quantitative exposureâ€“response relationships adjusted for personal characteristics and psychosocial conditions. BMC Musculoskeletal Disorders, 2019, 20, 139.	1.9	65

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19	Subcellular localization of sterol biosynthesis enzymes. Journal of Molecular Histology, 2019, 50, 63-73.	2.2	10
20	Human epididymis protein 4 (HE4) levels inversely correlate with lung function improvement (delta) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5 271-277.	0.7	18
21	Monogenic Forms of Diabetes Mellitus. Experientia Supplementum (2012), 2019, 111, 385-416.	0.9	15
22	A novel point mutation affecting Asn76 of dystrophin protein leads to dystrophinopathy. Neuromuscular Disorders, 2018, 28, 129-136.	0.6	9
23	Analytical parameters and validation of homopolymer detection in a pyrosequencing-based next generation sequencing system. BMC Genomics, 2018, 19, 158.	2.8	18
24	Comprehensive genetic testing in children with a clinical diagnosis of ARPKD identifies phenocopies. Pediatric Nephrology, 2018, 33, 1713-1721.	1.7	25
25	Interfering effect of maternal cell contamination on invasive prenatal molecular genetic testing. Prenatal Diagnosis, 2018, 38, 713-719.	2.3	1
26	Relative anterior microphthalmos in oculodentodigital dysplasia. Indian Journal of Ophthalmology, 2018, 66, 334.	1.1	2
27	Examination of Y-chromosomal microdeletions and partial microdeletions in idiopathic infertility in East Hungarian patients. Journal of Human Reproductive Sciences, 2018, 11, 329.	0.9	3
28	Vulnerability of DHCR7+/â˜' mutation carriers to aripiprazole and trazodone exposure. Journal of Lipid Research, 2017, 58, 2139-2146.	4.2	16
29	Different phenotypes in identical twins with cerebrotendinous xanthomatosis: case series. Neurological Sciences, 2017, 38, 481-483.	1.9	18
30	Myopia and Late-Onset Progressive Cone Dystrophy Associate to LVAVA/MVAVA Exon 3 Interchange Haplotypes of Opsin Genes on Chromosome X. , 2017, 58, 1834.		21
31	Congenital Hyperinsulinism Caused by a Mutation in the ABCC8 Gene - A Case Report. Electronic Journal of the International Federation of Clinical Chemistry and Laboratory Medicine, 2017, 28, 85-91.	0.7	1
32	A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. PLoS ONE, 2016, 11, e0162866.	2.5	96
33	Factors affecting thrombosis risk during pregnancy and in the postpartum period among factor V Leiden carriers: indications for selective prophylaxis. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2016, 206, 260.	1.1	0
34	Human Epididymis Protein 4: A Novel Serum Inflammatory Biomarker in CysticÂFibrosis. Chest, 2016, 150, 661-672.	0.8	45
35	The Effect of Small Molecules on Sterol Homeostasis: Measuring 7-Dehydrocholesterol in Dhcr7-Deficient Neuro2a Cells and Human Fibroblasts. Journal of Medicinal Chemistry, 2016, 59, 1102-1115.	6.4	48
36	Exposureâ€"response relationships for work-related neck and shoulder musculoskeletal disorders â€" Analyses of pooled uniform data sets. Applied Ergonomics, 2016, 55, 70-84.	3.1	112

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37	Significantly impaired shoulder function in the first years of rheumatoid arthritis: a controlled study. Arthritis Research and Therapy, 2015, 17, 261.	3.5	13
38	Ophthalmological phenotype associated with homozygous null mutation in the NEUROD1 gene. Molecular Vision, 2015, 21, 124-30.	1.1	4
39	Molecular Analysis of Cystic Fibrosis Patients in Hungary â€“ An Update to the Mutational Spectrum/Molekularna Analiza Obolelih Od CistiÄne Fibroze U MaÄ‘arskoj â€“ Dopune Spektru Mutacija. Journal of Medical Biochemistry, 2014, 34, 46-51.	1.7	9
40	<scp>ADAMTS</scp>9 locus associates with increased risk of wet <scp>AMD</scp>. Acta Ophthalmologica, 2014, 92, e410.	1.1	13
41	Cerebrotendinous xanthomatosis with the c.379<scp>C</scp>><scp>T</scp> (p.<scp>R</scp>127<scp>W</scp>) mutation in the <scp>CYP27A1</scp> gene associated with premature ageâ€associated limbic tauopathy. Neuropathology and Applied Neurobiology, 2014, 40, 345-350.	3.2	14
42	Complete recovery from psychosis upon miglustat treatment in a juvenile Niemannâ€“Pick C patient. European Journal of Paediatric Neurology, 2014, 18, 75-78.	1.6	19
43	Effect of the -133 a>g npc1l1 gene polymorphism on the efficacy of ezetimibe monotherapy. Atherosclerosis, 2014, 235, e75.	0.8	0
44	Novel dedicator of cytokinesis 8 mutations identified by multiplex ligationâ€dependent probe amplification. European Journal of Haematology, 2013, 91, 369-375.	2.2	4
45	Relation between biomarkers and clinical severity in patients with Smithâ€“Lemliâ€“Opitz syndrome. European Journal of Pediatrics, 2013, 172, 623-630.	2.7	19
46	Effect of Apolipoprotein E genotypes on the efficacy of ezetimibe monotherapy in patients with statin induced adverse effects. International Journal of Clinical Pharmacology and Therapeutics, 2013, 51, 746-752.	0.6	2
47	Sulfonylurea Use During Entire Pregnancy in Diabetes Because of KCNJ11 Mutation: A Report of Two Cases. Diabetes Care, 2012, 35, e40-e40.	8.6	13
48	Mutational Spectrum of Smith-Lemli-Opitz Syndrome Patients in Hungary. Molecular Syndromology, 2012, 3, 215-222.	0.8	14
49	Endocrine and anatomical findings in a case of Solitary Median Maxillary Central Incisor Syndrome. European Journal of Medical Genetics, 2012, 55, 109-111.	1.3	10
50	Effect of the Gas6 c.834+7G>A Polymorphism and the Interaction of Known Risk Factors on AMD Pathogenesis in Hungarian Patients. PLoS ONE, 2012, 7, e50181.	2.5	6
51	Molecular Diagnostic Challenges and Complex Management of Consecutive Twin Pregnancies in a Family with CD40 Ligand Deficiency. Scandinavian Journal of Immunology, 2012, 75, 227-230.	2.7	3
52	Distribution of CFTR mutations in Eastern Hungarians: Relevance to genetic testing and to the introduction of newborn screening for cystic fibrosis. Journal of Cystic Fibrosis, 2011, 10, 217-220.	0.7	11
53	Analysis of complement factor H Y402H, LOC387715, HTRA1 polymorphisms and ApoE alleles with susceptibility to ageâ€related macular degeneration in Hungarian patients. Acta Ophthalmologica, 2011, 89, 255-262.	1.1	19
54	Scientific correspondence. Neuropathology and Applied Neurobiology, 2011, 37, 428-430.	3.2	54

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55	Detection of mutations by flow cytometric melting point analysis of PCR products. Cytometry Part A: the Journal of the International Society for Analytical Cytology, 2011, 79A, 720-726.	1.5	1
56	Umbilical cord plasma levels of growth-arrest specific protein 6 in intrauterine growth restriction. Acta Obstetricia Et Gynecologica Scandinavica, 2010, 89, 22-26.	2.8	4
57	Personality trait scores among occupationally active bullied persons and witnesses to bullying. Motivation and Emotion, 2009, 33, 387-399.	1.3	39
58	Novel and recurrent STAT3 mutations in hyper-IgE syndrome patients from different ethnic groups. Molecular Immunology, 2008, 46, 202-206.	2.2	82
59	Investigation of Thr715Pro P-selectin gene polymorphism and soluble P-selectin levels in type 2 diabetes mellitus. Thrombosis and Haemostasis, 2007, 98, 186-191.	3.4	15
60	TGFBI (BIGH3) gene mutations in Hungary--report of the novel F547S mutation associated with polymorphic corneal amyloidosis. Molecular Vision, 2007, 13, 1976-83.	1.1	13
61	Analysis of the population heterogeneity in Hungary using fifteen forensically informative STR markers. Forensic Science International, 2006, 158, 244-249.	2.2	25
62	Severe Shwachman-Diamond syndrome phenotype caused by compound heterozygous missense mutations in the SBDS gene. Experimental Hematology, 2006, 34, 1517-1521.	0.4	27
63	Variable effect of prothrombotic factors on fetomaternal circulation. American Journal of Obstetrics and Gynecology, 2005, 193, 2180-2181.	1.3	0
64	A novel homozygous mutation (1619delC) in GPIIb gene associated with Glanzmann thrombasthenia, the decay of GPIIb-mRNA and the synthesis of a truncated GPIIb unable to form complex with GPIIIa. Thrombosis and Haemostasis, 2005, 93, 904-909.	3.4	7
65	Analysis of Gas6 in Human Platelets and Plasma. Arteriosclerosis, Thrombosis, and Vascular Biology, 2005, 25, 1280-1286.	2.4	114
66	Frequencies of two common mutations (c.35delG and c.167delT) of the connexin 26 gene in different populations of Hungary. International Journal of Molecular Medicine, 2004, 14, 1105.	4.0	1
67	Activated protein C resistance in anterior ischaemic optic neuropathy. Acta Ophthalmologica, 2004, 82, 140-143.	0.3	20
68	Frequencies of two common mutations (c.35delG and c.167delT) of the connexin 26 gene in different populations of Hungary. International Journal of Molecular Medicine, 2004, 14, 1105-8.	4.0	14
69	Anti-factor V auto-antibody in the plasma and platelets of a patient with repeated gastrointestinal bleeding. Journal of Thrombosis and Haemostasis, 2003, 1, 943-949.	3.8	34
70	Population genetic analysis in Hungarian populations using the Powerplexâ„¢ 16 system. International Congress Series, 2003, 1239, 121-122.	0.2	0
71	Severe coagulation factor V deficiency caused by 2 novel frameshift mutations: 2952delT in exon 13 and 5493insG in exon 16 of factor 5 gene. Blood, 2002, 99, 702-705.	1.4	34
72	The P28T Mutation in the GALK1 Gene Accounts for Galactokinase Deficiency in Roma (Gypsy) Patients across Europe. Pediatric Research, 2002, 51, 602-606.	2.3	40

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73	Factor V Leiden as a risk factor for miscarriage and reduced fertility. Australian and New Zealand Journal of Obstetrics and Gynaecology, 2000, 40, 186-190.	1.0	37
74	Val34Leu polymorphism of plasma factor XIII: biochemistry and epidemiology in familial thrombophilia. Blood, 2000, 96, 2479-2486.	1.4	120
75	State-of-the-Art-Review : The Leiden Mutation of Coagulation Factor V in Hungarian SLE Patients. Clinical and Applied Thrombosis/Hemostasis, 2000, 6, 41-45.	1.7	12
76	Hypercoagulability in various autoimmune diseases: no association with factor V Leiden mutation. Haematologia, 2000, 30, 35-39.	0.3	6
77	Muscular rest and gap frequency as EMG measures of physical exposure: the impact of work tasks and individual related. Ergonomics, 2000, 43, 1904-1919.	2.1	99
78	Val34Leu polymorphism of plasma factor XIII: biochemistry and epidemiology in familial thrombophilia. Blood, 2000, 96, 2479-86.	1.4	23
79	The frequency of the haemochromatosis C282Y mutation in the ethnic Hungarian and Romany populations of eastern Hungary. British Journal of Haematology, 1999, 107, 464-465.	2.5	12
80	High Prevalence of Factor V Leiden Mutation and 20210A Prothrombin Variant in Hungary. Thrombosis and Haemostasis, 1999, 81, 660-661.	3.4	20
81	High Frequency of Factor V Leiden Mutation and Prothrombin 20210A Variant in Romanies of Eastern Hungary. Thrombosis and Haemostasis, 1999, 82, 1555-1556.	3.4	10
82	Public health approach to activated protein C resistance assay. American Journal of Obstetrics and Gynecology, 1997, 177, 1271-1272.	1.3	2
83	Knee moment at work: validation of a questionnaire based on knee moment in working life. International Archives of Occupational and Environmental Health, 1996, 68, 321-324.	2.3	2
84	Knee moment at work: validation of a questionnaire based on knee moment in working life. International Archives of Occupational and Environmental Health, 1996, 68, 321-324.	2.3	1