Milan Macek

List of Publications by Year in descending order

50 papers	3,458 citations	394421 19 h-index	254184 43 g-index
51 all docs	51 docs citations	51 times ranked	6622 citing authors

#	Article	IF	Citations
1	Response to elexacaftor/tezacaftor/ivacaftor in intestinal organoids derived from people with cystic fibrosis. Journal of Cystic Fibrosis, 2022, 21, 243-245.	0.7	10
2	Early-onset pulmonary and cutaneous vasculitis driven by constitutively active SRC-family kinase HCK. Journal of Allergy and Clinical Immunology, 2022, 149, 1464-1472.e3.	2.9	10
3	<scp>Zimmermannâ€“Laband</scp> syndrome in monozygotic twins with a mild neurobehavioral phenotype lacking gingival overgrowthâ€”A case report of a novel <scp> KCNN3 </scp> gene variant. American Journal of Medical Genetics, Part A, 2022, 188, 1083-1087.	1.2	2
4	Co-designing models for the communication of genomic results for rare diseases: a comparative study in the Czech Republic and the United Kingdom. Journal of Community Genetics, 2022, 13, 313-327.	1.2	3
5	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	2.8	48
6	Opportunistic genomic screening. Recommendations of the European Society of Human Genetics. European Journal of Human Genetics, 2021, 29, 365-377.	2.8	76
7	Outcomes of post mortem genetic diagnosis in SCD victims and primary prevention of cardiac arrest in relatives: a nationwide multidisciplinary and multicentric collaboration in the Czechia. European Journal of Preventive Cardiology, 2021, 28, .	1.8	0
8	Concealed cardiomyopathy as a frequent cause of idiopathic ventricular fibrillation in a representative Czech cohort of survivors of sudden cardiac arrest (SCA). Europace, 2021, 23, .	1.7	0
9	Novel LOX Variants in Five Families with Aortic/Arterial Aneurysm and Dissection with Variable Connective Tissue Findings. International Journal of Molecular Sciences, 2021, 22, 7111.	4.1	7
10	Demographic characteristics, clinical and laboratory features, and the distribution of pathogenic variants in the CFTR gene in the Cypriot cystic fibrosis (CF) population demonstrate the utility of a national CF patient registry. Orphanet Journal of Rare Diseases, 2021, 16, 409.	2.7	6
11	The future of cystic fibrosis care: a global perspective. Lancet Respiratory Medicine,the, 2020, 8, 65-124.	10.7	573
12	The impact of type 1 diabetes mellitus on male sexual functions and sex hormone levels. Endocrine Journal, 2020, 67, 59-71.	1.6	11
13	Monitoring of Early Changes of Circulating Tumor DNA in the Plasma of Rectal Cancer Patients Receiving Neoadjuvant Concomitant Chemoradiotherapy: Evaluation for Prognosis and Prediction of Therapeutic Response. Frontiers in Oncology, 2020, 10, 1028.	2.8	25
14	P1107First results of molecular autopsy examinations in sudden cardiac death drawn from nationwide multidisciplinary and multicentric collaboration in the Czech Republic. Europace, 2020, 22, .	1.7	0
15	Patients with hypertrophic obstructive cardiomyopathy after alcohol septal ablation have favorable long-term outcome irrespective of their genetic background. Cardiovascular Diagnosis and Therapy, 2020, 10, 193-200.	1.7	3
16	Laboratory biomarkers for lung disease severity and progression in cystic fibrosis. Clinica Chimica Acta, 2020, 508, 277-286.	1.1	11
17	The Key Role of Purine Metabolism in the Folate-Dependent Phenotype of Autism Spectrum Disorders: An In Silico Analysis. Metabolites, 2020, 10, 184.	2.9	7
18	Assisted Reproductive Techniques and Pregnancy Results in Women with Mayer-Rokitansky-KÃ¼ster-Hauser Syndrome Undergoing Uterus Transplantation: the Czech Experience. Journal of Pediatric and Adolescent Gynecology, 2020, 33, 410-414.	0.7	18

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19	Desminopathy: Novel Desmin Variants, a New Cardiac Phenotype, and Further Evidence for Secondary Mitochondrial Dysfunction. Journal of Clinical Medicine, 2020, 9, 937.	2.4	24
20	Comparison of variant detection rate in genes between two cohorts of Czech living patients versus victims of sudden cardiac death with clinical / post mortem diagnosis of non-ischemic cardiomyopathy. European Heart Journal, 2020, 41, .	2.2	0
21	The Gen-Equip Project: evaluation and impact of genetics e-learning resources for primary care in six European languages. Genetics in Medicine, 2019, 21, 718-726.	2.4	17
22	The utility of the Mayo Score for predicting the yield of genetic testing in patients with hypertrophic cardiomyopathy. Archives of Medical Science, 2019, 15, 641-649.	0.9	20
23	5162Novel insights into desminopathy in the era of next generation sequencing. European Heart Journal, 2019, 40, .	2.2	0
24	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
25	Epidemiology of rare diseases detected by newborn screening in the Czech Republic. Central European Journal of Public Health, 2019, 27, 153-159.	1.1	16
26	Analytical parameters and validation of homopolymer detection in a pyrosequencing-based next generation sequencing system. BMC Genomics, 2018, 19, 158.	2.8	18
27	Genome-wide association study identifies inversion in the CTRB1-CTRB2 locus to modify risk for alcoholic and non-alcoholic chronic pancreatitis. Gut, 2018, 67, 1855-1863.	12.1	97
28	Recent developments in genetics and medically assisted reproduction: from research to clinical applications. European Journal of Human Genetics, 2018, 26, 12-33.	2.8	76
29	Modeling ageâ€specific facial development in Williamsâ€“Beurenâ€, Noonanâ€, and 22q11.2 deletion syndromes in cohorts of Czech patients aged 3â€“18â€‰years: A crossâ€sectional threeâ€dimensional geometric morphometry analysis of their facial gestalt. American Journal of Medical Genetics, Part A, 2018, 176, 2604-2613.	1.2	2
30	Estimating the age of p.(Phe508del) with family studies of geographically distinct European populations and the early spread of cystic fibrosis. European Journal of Human Genetics, 2018, 26, 1832-1839.	2.8	45
31	Cost-of-illness analysis and regression modeling in cystic fibrosis: a retrospective prevalence-based study. European Journal of Health Economics, 2017, 18, 73-82.	2.8	10
32	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	6.2	305
33	Implementing genetic education in primary care: the Gen-Equip programme. Journal of Community Genetics, 2017, 8, 147-150.	1.2	16
34	Identification of likely associations between cerebral folate deficiency and complex geneticâ€•and metabolic pathogenesis of autism spectrum disorders by utilization of a pilot interaction modeling approach. Autism Research, 2017, 10, 1424-1435.	3.8	15
35	From Mendel to Medical Genetics. European Journal of Human Genetics, 2017, 25, S53-S59.	2.8	6
36	A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. PLoS ONE, 2016, 11, e0162866.	2.5	96

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37	Molecular genetic analysis in 14 Czech Kabuki syndrome patients is confirming the utility of phenotypic scoring. Clinical Genetics, 2016, 90, 230-237.	2.0	16
38	Human Epididymis Protein 4: A Novel Serum Inflammatory Biomarker in CysticÂFibrosis. Chest, 2016, 150, 661-672.	0.8	45
39	A product of immunoreactive trypsinogen and pancreatitis-associated protein as second-tier strategy in cystic fibrosis newborn screening. Journal of Cystic Fibrosis, 2016, 15, 752-758.	0.7	14
40	Comparison of different IRT-PAP protocols to screen newborns for cystic fibrosis in three central European populations. Journal of Cystic Fibrosis, 2014, 13, 15-23.	0.7	39
41	Bridging genomics research between developed and developing countries: the Genomic Medicine Alliance. Personalized Medicine, 2014, 11, 615-623.	1.5	22
42	European Cystic Fibrosis Society Standards of Care: Framework for the Cystic Fibrosis Centre. Journal of Cystic Fibrosis, 2014, 13, S3-S22.	0.7	153
43	Current issues in medically assisted reproduction and genetics in Europe: research, clinical practice, ethics, legal issues and policy. European Journal of Human Genetics, 2013, 21, S1-S21.	2.8	120
44	Prospective and parallel assessments of cystic fibrosis newborn screening protocols in the Czech Republic: IRT/DNA/IRT versus IRT/PAP and IRT/PAP/DNA. European Journal of Pediatrics, 2012, 171, 1223-1229.	2.7	29
45	Genetical realtions of social loneliness in geriatric patients. European Psychiatry, 2011, 26, 808-808.	0.2	0
46	Diagnostic method validation: High resolution melting (HRM) of small amplicons genotyping for the most common variants in the MTHFR gene. Clinical Biochemistry, 2009, 42, 1308-1316.	1.9	46
47	Correlation between Genetic and Geographic Structure in Europe. Current Biology, 2008, 18, 1241-1248.	3.9	449
48	Cystic fibrosis: A worldwide analysis ofCFTR mutations?correlation with incidence data and application to screening. Human Mutation, 2002, 19, 575-606.	2.5	904
49	Pancreatic insufficiency and pulmonary disease in German and Slavic cystic fibrosis patients with the R347P mutation. Human Mutation, 1995, 6, 219-225.	2.5	11
50	First-trimester prenatal diagnosis of the nijmegen breakage syndrome and ataxia telangiectasia using an assay of radioresistant dna synthesis. Prenatal Diagnosis, 1990, 10, 667-674.	2.3	19