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	Cystic fibrosis: A worldwide analysis ofCFTR mutations?correlation with incidence data and application to screening. Human Mutation, 2002, 19, 575-606.	2.5	904
2	The future of cystic fibrosis care: a global perspective. Lancet Respiratory Medicine,the, 2020, 8, 65-124.	10.7	573
3	Correlation between Genetic and Geographic Structure in Europe. Current Biology, 2008, 18, 1241-1248.	3.9	449
4	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	6.2	305
5	European Cystic Fibrosis Society Standards of Care: Framework for the Cystic Fibrosis Centre. Journal of Cystic Fibrosis, 2014, 13, S3-S22.	0.7	153
6	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
7	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
8	A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. PLoS ONE, 2016, 11, e0162866.	2.5	96
9	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
10	Opportunistic genomic screening. Recommendations of the European Society of Human Genetics. European Journal of Human Genetics, 2021, 29, 365-377.	2.8	76
11	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	2.8	48
12	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
13	Human Epididymis Protein 4: A Novel Serum Inflammatory Biomarker in CysticÂFibrosis. Chest, 2016, 150, 661-672.	0.8	45
14	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
15	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
16	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
17	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
18	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

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19	Bridging genomics research between developed and developing countries: the Genomic Medicine Alliance. Personalized Medicine, 2014, 11, 615-623.	1.5	22
20	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
21	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
22	Analytical parameters and validation of homopolymer detection in a pyrosequencing-based next generation sequencing system. BMC Genomics, 2018, 19, 158.	2.8	18
23	Human epididymis protein 4 (HE4) levels inversely correlate with lung function improvement (delta) Tj ETQq1 1 0.784314 rgBT /Overl 271-277.	0.7	18
24	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
25	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
26	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
27	Implementing genetic education in primary care: the Gen-Equip programme. Journal of Community Genetics, 2017, 8, 147-150.	1.2	16
28	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
29	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
30	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
31	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
32	The impact of type 1 diabetes mellitus on male sexual functions and sex hormone levels. Endocrine Journal, 2020, 67, 59-71.	1.6	11
33	Laboratory biomarkers for lung disease severity and progression in cystic fibrosis. Clinica Chimica Acta, 2020, 508, 277-286.	1.1	11
34	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
35	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
36	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

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37	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
38	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
39	From Mendel to Medical Genetics. European Journal of Human Genetics, 2017, 25, S53-S59.	2.8	6
40	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
41	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
42	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
43	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
44	<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
45	Genetical realtions of social loneliness in geriatric patients. European Psychiatry, 2011, 26, 808-808.	0.2	0
46	5162Novel insights into desminopathy in the era of next generation sequencing. European Heart Journal, 2019, 40, .	2.2	0
47	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
48	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
49	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
50	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