

Gabor Matyas

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

60
papers

2,597
citations

25
h-index

50
g-index

61
ext. papers

2,899
ext. citations

5.3
avg, IF

4.38
L-index

#	Paper	IF	Citations
60	Potential predictors of severe cardiovascular involvement in Marfan syndrome: the emphasized role of genotype-phenotype correlations in improving risk stratification-a literature review. <i>Orphanet Journal of Rare Diseases</i> , 2021 , 16, 245	4.2	1
59	Interdisziplinäre Betreuung von Menschen mit Marfan-Syndrom IPharmakologie, Schwangerschaft, Auge, Skelett und organisatorische Aspekte. <i>Zeitschrift Fur Herz-, Thorax- Und Gefasschirurgie</i> , 2021 , 35, 232-241	0.1	
58	Potential of whole-genome sequencing-based pharmacogenetic profiling. <i>Pharmacogenomics</i> , 2021 , 22, 177-190	2.6	5
57	Celiprolol but not losartan improves the biomechanical integrity of the aorta in a mouse model of vascular Ehlers-Danlos syndrome. <i>Cardiovascular Research</i> , 2020 , 116, 457-465	9.9	15
56	Interdisziplinäre Betreuung von Menschen mit Marfan-Syndrom IIGenetik, bildgebende Untersuchung und kardiovaskuläres System. <i>Zeitschrift Fur Herz-, Thorax- Und Gefasschirurgie</i> , 2020 , 34, 115-126	0.1	1
55	Hereditäre Bindegewebskrankheiten. <i>Springer Reference Medizin</i> , 2020 , 2835-2859	0	
54	Vascular Ehlers-Danlos syndrome: can the beneficial effect of celiprolol be extrapolated to bisoprolol?. <i>European Heart Journal - Cardiovascular Pharmacotherapy</i> , 2020 , 6, 199-200	6.4	4
53	Variant filtering, digenic variants, and other challenges in clinical sequencing: a lesson from fibrillinopathies. <i>Clinical Genetics</i> , 2020 , 97, 235-245	4	11
52	Optimising the mutation screening strategy in Marfan syndrome and identifying genotypes with more severe aortic involvement. <i>Orphanet Journal of Rare Diseases</i> , 2020 , 15, 290	4.2	5
51	Added Value of Clinical Sequencing: WGS-Based Profiling of Pharmacogenes. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	16
50	More Genes for Thoracic Aortic Aneurysms and Dissections. <i>Journal of the American College of Cardiology</i> , 2019 , 73, 528-529	15.1	2
49	Hereditäre Bindegewebskrankheiten bei Kindern und Jugendlichen. <i>Springer Reference Medizin</i> , 2019 , 1-25	0	1
48	Clinical sequencing: From raw data to diagnosis with lifetime value. <i>Clinical Genetics</i> , 2018 , 93, 508-519	4	53
47	Hungarian Marfan family with large deletion calls attention to copy number variation detection in the current NGS era. <i>Journal of Thoracic Disease</i> , 2018 , 10, 2456-2460	2.6	10
46	Response to: The genetics and pathogenesis of thoracic aortic aneurysm disorder and dissections. <i>Clinical Genetics</i> , 2017 , 91, 501	4	1
45	Need for speed in accurate whole-genome data analysis: GENALICE MAP challenges BWA/GATK more than PEMapper/PECaller and Isaac. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017 , 114, E8320-E8322	11.5	14
44	Clinical sequencing: is WGS the better WES?. <i>Human Genetics</i> , 2016 , 135, 359-62	6.3	174

43	Birt-Hogg-Dubé syndrome: novel FLCN frameshift deletion in daughter and father with renal cell carcinomas. <i>Familial Cancer</i> , 2016 , 15, 127-32	3	1
42	New insights into the performance of human whole-exome capture platforms. <i>Nucleic Acids Research</i> , 2015 , 43, e76	20.1	80
41	Gene polymorphisms as risk factors for predicting the cardiovascular manifestations in Marfan syndrome. Role of folic acid metabolism enzyme gene polymorphisms in Marfan syndrome. <i>Thrombosis and Haemostasis</i> , 2015 , 114, 748-56	7	13
40	Recurrent spontaneous coronary dissections in a patient with a de novo fibrillin-1 mutation without Marfan syndrome. <i>Thrombosis and Haemostasis</i> , 2015 , 113, 668-70	7	3
39	Hereditäre Bindegewebskrankheiten bei Kindern und Jugendlichen 2015 , 1-24		
38	Medical treatment of aortic aneurysms in Marfan syndrome and other heritable conditions. <i>Current Cardiology Reviews</i> , 2014 , 10, 161-71	2.4	27
37	De novo mutation of the latency-associated peptide domain of TGFβ3 in a patient with overgrowth and Loeys-Dietz syndrome features. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 2141-3	2.5	26
36	Outcome of aortic surgery in patients with Loeys-Dietz syndrome primarily treated as having Marfan syndrome. <i>European Journal of Cardio-thoracic Surgery</i> , 2014 , 46, 444-9; discussion 449	3	10
35	Genetische Bindegewebskrankheiten 2014 , 1912-1925		1
34	Acute aortic dissection determines the fate of initially untreated aortic segments in Marfan syndrome. <i>Circulation</i> , 2013 , 127, 1569-75	16.7	67
33	Precise breakpoint localization of large genomic deletions using PacBio and Illumina next-generation sequencers. <i>BioTechniques</i> , 2013 , 54, 98-100	2.5	9
32	Barth syndrome in a female patient. <i>Molecular Genetics and Metabolism</i> , 2012 , 106, 115-20	3.7	37
31	Phenotypic spectrum of the SMAD3-related aneurysms-osteoarthritis syndrome. <i>Journal of Medical Genetics</i> , 2012 , 49, 47-57	5.8	189
30	Cardiovascular surgery in Marfan syndrome: implications of new molecular concepts in thoracic aortic disease. <i>Future Cardiology</i> , 2011 , 7, 557-69	1.3	6
29	Hemizygous deletion of COL3A1, COL5A2, and MSTN causes a complex phenotype with aortic dissection: a lesson for and from true haploinsufficiency. <i>European Journal of Human Genetics</i> , 2010 , 18, 1315-21	5.3	22
28	HMOX1 and GST variants modify attenuation of FEV25-75% decline due to PM10 reduction. <i>European Respiratory Journal</i> , 2010 , 35, 505-14	13.6	15
27	Proteomic analysis in aortic media of patients with Marfan syndrome reveals increased activity of calpain 2 in aortic aneurysms. <i>Circulation</i> , 2009 , 120, 983-91	16.7	44
26	Quantitative sequence analysis of FBN1 premature termination codons provides evidence for incomplete NMD in leukocytes. <i>Human Mutation</i> , 2009 , 30, 1355-64	4.7	21

25	A new sporadic case of early-onset Loews-Dietz syndrome due to the recurrent mutation p.R528C in the TGFBR2 gene substantiates interindividual clinical variability. <i>Journal of Applied Genetics</i> , 2009 , 50, 405-10	2.5	13
24	Severe phenotype with cis-acting heterozygous PMP22 mutations. <i>Clinical Genetics</i> , 2009 , 75, 286-9	4	3
23	Polar body biopsy for Curschmann-Steinert disease and successful pregnancy following embryo vitrification. <i>Reproductive BioMedicine Online</i> , 2009 , 18, 815-20	4	3
22	Re: Management of patients with aortic dissection. New insights. <i>Deutsches A&#x0308;rzteblatt International</i> , 2009 , 106, 171; author reply 171-2	2.5	
21	Novel mutations in the folliculin gene associated with spontaneous pneumothorax. <i>European Respiratory Journal</i> , 2008 , 32, 1316-20	13.6	44
20	Glutathione S-transferase genotypes modify lung function decline in the general population: SAPALDIA cohort study. <i>Respiratory Research</i> , 2007 , 8, 2	7.3	50
19	Large genomic fibrillin-1 (FBN1) gene deletions provide evidence for true haploinsufficiency in Marfan syndrome. <i>Human Genetics</i> , 2007 , 122, 23-32	6.3	62
18	A bioinformatics framework for genotype-phenotype correlation in humans with Marfan syndrome caused by FBN1 gene mutations. <i>Journal of Biomedical Informatics</i> , 2006 , 39, 171-83	10.2	7
17	Different patterns of aortic wall elasticity in patients with Marfan syndrome: a noninvasive follow-up study. <i>Journal of Thoracic and Cardiovascular Surgery</i> , 2006 , 132, 811-9	1.5	35
16	Identification and in silico analyses of novel TGFBR1 and TGFBR2 mutations in Marfan syndrome-related disorders. <i>Human Mutation</i> , 2006 , 27, 760-9	4.7	89
15	Mutations in CABP4, the gene encoding the Ca ²⁺ -binding protein 4, cause autosomal recessive night blindness. <i>American Journal of Human Genetics</i> , 2006 , 79, 657-67	11	134
14	Clinical profiles of four patients with Rett syndrome carrying a novel exon 1 mutation or genomic rearrangement in the MECP2 gene. <i>Clinical Genetics</i> , 2006 , 69, 319-26	4	13
13	The common G-allele of interleukin-18 single-nucleotide polymorphism is a genetic risk factor for atopic asthma. The SAPALDIA Cohort Study. <i>Clinical and Experimental Allergy</i> , 2006 , 36, 211-8	4.1	42
12	Diagnostic power of aortic elastic properties in young patients with Marfan syndrome. <i>Journal of Thoracic and Cardiovascular Surgery</i> , 2005 , 129, 730-9	1.5	61
11	Marfan Syndrome. <i>Methods of Information in Medicine</i> , 2005 , 44, 487-497	1.5	5
10	Mutations in GRM6 cause autosomal recessive congenital stationary night blindness with a distinctive scotopic 15-Hz flicker electroretinogram. <i>Investigative Ophthalmology and Visual Science</i> , 2005 , 46, 4328-35		116
9	Novel mutations in CACNA1F and NYX in Dutch families with X-linked congenital stationary night blindness. <i>Molecular Vision</i> , 2005 , 11, 179-83	2.3	25
8	Genetic variation of oaks (<i>Quercus</i> spp.) in Switzerland. 3. Lack of impact of postglacial recolonization history on nuclear gene loci. <i>Theoretical and Applied Genetics</i> , 2003 , 106, 346-52	6	21

7	Update of the UMD-FBN1 mutation database and creation of an FBN1 polymorphism database. <i>Human Mutation</i> , 2003 , 22, 199-208	4-7	251
6	Quantification of single nucleotide polymorphisms: a novel method that combines primer extension assay and capillary electrophoresis. <i>Human Mutation</i> , 2002 , 19, 58-68	4-7	29
5	Evaluation and application of denaturing HPLC for mutation detection in Marfan syndrome: Identification of 20 novel mutations and two novel polymorphisms in the FBN1 gene. <i>Human Mutation</i> , 2002 , 19, 443-56	4-7	51
4	Identification of refugia and post-glacial colonisation routes of European white oaks based on chloroplast DNA and fossil pollen evidence. <i>Forest Ecology and Management</i> , 2002 , 156, 49-74	3-9	482
3	Chloroplast DNA variation of white oaks in the alpine region. <i>Forest Ecology and Management</i> , 2002 , 156, 131-145	3-9	33
2	Chloroplast DNA polymorphisms provide evidence for postglacial re-colonisation of oaks (<i>Quercus</i> spp.) across the Swiss Alps. <i>Theoretical and Applied Genetics</i> , 2001 , 102, 12-20	6	49
1	Tandem repeats in plant mitochondrial genomes: application to the analysis of population differentiation in the conifer Norway spruce. <i>Molecular Ecology</i> , 2001 , 10, 257-63	5-7	95