Gabor Matyas

List of Publications by Citations

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	Identification of refugia and post-glacial colonisation routes of European white oaks based on chloroplast DNA and fossil pollen evidence. *Forest Ecology and Management*, **2002**, 156, 49-74	3.9	482
59	Update of the UMD-FBN1 mutation database and creation of an FBN1 polymorphism database. *Human Mutation*, **2003**, 22, 199-208	4.7	251
58	Phenotypic spectrum of the SMAD3-related aneurysms-osteoarthritis syndrome. *Journal of Medical Genetics*, **2012**, 49, 47-57	5.8	189
57	Clinical sequencing: is WGS the better WES?. *Human Genetics*, **2016**, 135, 359-62	6.3	174
56	Mutations in CABP4, the gene encoding the Ca2+-binding protein 4, cause autosomal recessive night blindness. *American Journal of Human Genetics*, **2006**, 79, 657-67	11	134
55	Mutations in GRM6 cause autosomal recessive congenital stationary night blindness with a distinctive scotopic 15-Hz flicker electroretinogram. *Investigative Ophthalmology and Visual Science*, **2005**, 46, 4328-35		116
54	Tandem repeats in plant mitochondrial genomes: application to the analysis of population differentiation in the conifer Norway spruce. *Molecular Ecology*, **2001**, 10, 257-63	5.7	95
53	Identification and in silico analyses of novel TGFBR1 and TGFBR2 mutations in Marfan syndrome-related disorders. *Human Mutation*, **2006**, 27, 760-9	4.7	89
52	New insights into the performance of human whole-exome capture platforms. *Nucleic Acids Research*, **2015**, 43, e76	20.1	80
51	Acute aortic dissection determines the fate of initially untreated aortic segments in Marfan syndrome. *Circulation*, **2013**, 127, 1569-75	16.7	67
50	Large genomic fibrillin-1 (FBN1) gene deletions provide evidence for true haploinsufficiency in Marfan syndrome. *Human Genetics*, **2007**, 122, 23-32	6.3	62
49	Diagnostic power of aortic elastic properties in young patients with Marfan syndrome. *Journal of Thoracic and Cardiovascular Surgery*, **2005**, 129, 730-9	1.5	61
48	Clinical sequencing: From raw data to diagnosis with lifetime value. *Clinical Genetics*, **2018**, 93, 508-519	4	53
47	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. *Human Mutation*, **2002**, 19, 443-56	4.7	51
46	Glutathione S-transferase genotypes modify lung function decline in the general population: SAPALDIA cohort study. *Respiratory Research*, **2007**, 8, 2	7.3	50
45	Chloroplast DNA polymorphisms provide evidence for postglacial re-colonisation of oaks (Quercus spp.) across the Swiss Alps. *Theoretical and Applied Genetics*, **2001**, 102, 12-20	6	49
44	Proteomic analysis in aortic media of patients with Marfan syndrome reveals increased activity of calpain 2 in aortic aneurysms. *Circulation*, **2009**, 120, 983-91	16.7	44

43	Novel mutations in the folliculin gene associated with spontaneous pneumothorax. *European Respiratory Journal*, **2008**, 32, 1316-20	13.6	44
42	The common G-allele of interleukin-18 single-nucleotide polymorphism is a genetic risk factor for atopic asthma. The SAPALDIA Cohort Study. *Clinical and Experimental Allergy*, **2006**, 36, 211-8	4.1	42
41	Barth syndrome in a female patient. *Molecular Genetics and Metabolism*, **2012**, 106, 115-20	3.7	37
40	Different patterns of aortic wall elasticity in patients with Marfan syndrome: a noninvasive follow-up study. *Journal of Thoracic and Cardiovascular Surgery*, **2006**, 132, 811-9	1.5	35
39	Chloroplast DNA variation of white oaks in the alpine region. *Forest Ecology and Management*, **2002**, 156, 131-145	3.9	33
38	Quantification of single nucleotide polymorphisms: a novel method that combines primer extension assay and capillary electrophoresis. *Human Mutation*, **2002**, 19, 58-68	4.7	29
37	Medical treatment of aortic aneurysms in Marfan syndrome and other heritable conditions. *Current Cardiology Reviews*, **2014**, 10, 161-71	2.4	27
36	De novo mutation of the latency-associated peptide domain of TGFB3 in a patient with overgrowth and Loeys-Dietz syndrome features. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2141-3	2.5	26
35	Novel mutations in CACNA1F and NYX in Dutch families with X-linked congenital stationary night blindness. *Molecular Vision*, **2005**, 11, 179-83	2.3	25
34	Hemizygous deletion of COL3A1, COL5A2, and MSTN causes a complex phenotype with aortic dissection: a lesson for and from true haploinsufficiency. *European Journal of Human Genetics*, **2010**, 18, 1315-21	5.3	22
33	Quantitative sequence analysis of FBN1 premature termination codons provides evidence for incomplete NMD in leukocytes. *Human Mutation*, **2009**, 30, 1355-64	4.7	21
32	Genetic variation of oaks (Quercus spp.) in Switzerland. 3. Lack of impact of postglacial recolonization history on nuclear gene loci. *Theoretical and Applied Genetics*, **2003**, 106, 346-52	6	21
31	Added Value of Clinical Sequencing: WGS-Based Profiling of Pharmacogenes. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	16
30	Celiprolol but not losartan improves the biomechanical integrity of the aorta in a mouse model of vascular Ehlers-Danlos syndrome. *Cardiovascular Research*, **2020**, 116, 457-465	9.9	15
29	HMOX1 and GST variants modify attenuation of FEF25-75% decline due to PM10 reduction. *European Respiratory Journal*, **2010**, 35, 505-14	13.6	15
28	Need for speed in accurate whole-genome data analysis: GENALICE MAP challenges BWA/GATK more than PEMapper/PECaller and Isaac. *Proceedings of the National Academy of Sciences of the United States of America*, **2017**, 114, E8320-E8322	11.5	14
27	Gene polymorphisms as risk factors for predicting the cardiovascular manifestations in Marfan syndrome. Role of folic acid metabolism enzyme gene polymorphisms in Marfan syndrome. *Thrombosis and Haemostasis*, **2015**, 114, 748-56	7	13
26	A new sporadic case of early-onset Loeys-Dietz syndrome due to the recurrent mutation p.R528C in the TGFBR2 gene substantiates interindividual clinical variability. *Journal of Applied Genetics*, **2009**, 50, 405-10	2.5	13

25	Clinical profiles of four patients with Rett syndrome carrying a novel exon 1 mutation or genomic rearrangement in the MECP2 gene. *Clinical Genetics*, **2006**, 69, 319-26	4	13
24	Variant filtering, digenic variants, and other challenges in clinical sequencing: a lesson from fibrillinopathies. *Clinical Genetics*, **2020**, 97, 235-245	4	11
23	Outcome of aortic surgery in patients with Loeys-Dietz syndrome primarily treated as having Marfan syndrome. *European Journal of Cardio-thoracic Surgery*, **2014**, 46, 444-9; discussion 449	3	10
22	Hungarian Marfan family with large deletion calls attention to copy number variation detection in the current NGS era. *Journal of Thoracic Disease*, **2018**, 10, 2456-2460	2.6	10
21	Precise breakpoint localization of large genomic deletions using PacBio and Illumina next-generation sequencers. *BioTechniques*, **2013**, 54, 98-100	2.5	9
20	A bioinformatics framework for genotype-phenotype correlation in humans with Marfan syndrome caused by FBN1 gene mutations. *Journal of Biomedical Informatics*, **2006**, 39, 171-83	10.2	7
19	Cardiovascular surgery in Marfan syndrome: implications of new molecular concepts in thoracic aortic disease. *Future Cardiology*, **2011**, 7, 557-69	1.3	6
18	Marfan Syndrome. *Methods of Information in Medicine*, **2005**, 44, 487-497	1.5	5
17	Optimising the mutation screening strategy in Marfan syndrome and identifying genotypes with more severe aortic involvement. *Orphanet Journal of Rare Diseases*, **2020**, 15, 290	4.2	5
16	Potential of whole-genome´sequencing-based pharmacogenetic profiling. *Pharmacogenomics*, **2021**, 22, 177-190	2.6	5
15	Vascular Ehlers-Danlos syndrome: can the beneficial effect of celiprolol be extrapolated to bisoprolol?. *European Heart Journal - Cardiovascular Pharmacotherapy*, **2020**, 6, 199-200	6.4	4
14	Recurrent spontaneous coronary dissections in a patient with a de novo fibrillin-1 mutation without Marfan syndrome. *Thrombosis and Haemostasis*, **2015**, 113, 668-70	7	3
13	Severe phenotype with cis-acting heterozygous PMP22 mutations. *Clinical Genetics*, **2009**, 75, 286-9	4	3
12	Polar body biopsy for Curschmann-Steinert disease and successful pregnancy following embryo vitrification. *Reproductive BioMedicine Online*, **2009**, 18, 815-20	4	3
11	More Genes for Thoracic Aortic Aneurysms and´Dissections. *Journal of the American College of Cardiology*, **2019**, 73, 528-529	15.1	2
10	Response to: The genetics and pathogenesis of thoracic aortic aneurysm disorder and dissections. *Clinical Genetics*, **2017**, 91, 501	4	1
9	Interdisziplinäre Betreuung von Menschen mit Marfan-Syndrom – Genetik, bildgebende Untersuchung und kardiovaskuläres System. *Zeitschrift Fur Herz-, Thorax- Und Gefasschirurgie*, **2020**, 34, 115-126	0.1	1
8	Birt-Hogg-Dubé syndrome: novel FLCN frameshift deletion in daughter and father with renal cell carcinomas. *Familial Cancer*, **2016**, 15, 127-32	3	1

7	Genetische Bindegewebskrankheiten **2014**, 1912-1925		1
6	Hereditäre Bindegewebskrankheiten bei Kindern und Jugendlichen. *Springer Reference Medizin*, **2019**, 1-25	0	1
5	Potential predictors of severe cardiovascular involvement in Marfan syndrome: the emphasized role of genotype-phenotype correlations in improving risk stratification-a literature review. *Orphanet Journal of Rare Diseases*, **2021**, 16, 245	4.2	1
4	Hereditäre Bindegewebskrankheiten. *Springer Reference Medizin*, **2020**, 2835-2859	0	
3	Hereditäre Bindegewebskrankheiten bei Kindern und Jugendlichen **2015**, 1-24		
2	Re: Management of patients with aortic dissection. New insights. *Deutsches Ärzteblatt International*, **2009**, 106, 171; author reply 171-2	2.5	
1	Interdisziplinäre Betreuung von Menschen mit Marfan-Syndrom – Pharmakologie, Schwangerschaft, Auge, Skelett und organisatorische Aspekte. *Zeitschrift Fur Herz-, Thorax- Und Gefasschirurgie*, **2021**, 35, 232-241	0.1	