Viktor Stranecky

List of Publications by Year in Descending Order

70 papers	1,797 citations	22 h-index	41 g-index
78 ext. papers	2,216 ext. citations	6.9 avg, IF	3.77 L-index

#	Paper	IF	Citations
70	Biallelic loss-of-function variants in PLD1 cause congenital right-sided cardiac valve defects and neonatal cardiomyopathy. *Journal of Clinical Investigation*, **2021**, 131,	15.9	4
69	Genetic heterogeneity of neuronal intranuclear inclusion disease: What about the infantile variant?. *Annals of Clinical and Translational Neurology*, **2021**, 8, 994-1001	5.3	1
68	Non-Penetrance for Ocular Phenotype in Two Individuals Carrying Heterozygous Loss-of-Function Alleles. *Genes*, **2021**, 12,	4.2	0
67	Re: ERCC3, a new ovarian cancer susceptibility gene?. *European Journal of Cancer*, **2021**, 150, 278-280	7.5	
66	GPD1 Deficiency - Underdiagnosed Cause of Liver Disease. *Indian Journal of Pediatrics*, **2021**, 88, 80-81	3	2
65	POLRMT mutations impair mitochondrial transcription causing neurological disease. *Nature Communications*, **2021**, 12, 1135	17.4	3
64	A mutation in the SAA1 promoter causes hereditary amyloid A amyloidosis. *Kidney International*, **2021**,	9.9	3
63	Pseudodominant Nanophthalmos in a Roma Family Caused by a Novel Variant. *Journal of Ophthalmology*, **2020**, 2020, 6807809	2	1
62	Severe phenotype of ATP6AP1-CDG in two siblings with a novel mutation leading to a differential tissue-specific ATP6AP1 protein pattern, cellular oxidative stress and hepatic copper accumulation. *Journal of Inherited Metabolic Disease*, **2020**, 43, 694-700	5.4	8
61	Fatal neonatal nephrocutaneous syndrome in 18 Roma children with EGFR deficiency. *Journal of Dermatology*, **2020**, 47, 663-668	1.6	2
60	Multigene Panel Germline Testing of 1333 Czech Patients with Ovarian Cancer. *Cancers*, **2020**, 12,	6.6	8
59	Spinal muscular atrophy caused by a novel Alu-mediated deletion of exons 2a-5 in SMN1 undetectable with routine genetic testing. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1238	2.3	7
58	Desminopathy: Novel Desmin Variants, a New Cardiac Phenotype, and Further Evidence for Secondary Mitochondrial Dysfunction. *Journal of Clinical Medicine*, **2020**, 9,	5.1	7
57	Autosomal-dominant adult neuronal ceroid lipofuscinosis caused by duplication in DNAJC5 initially missed by Sanger and whole-exome sequencing. *European Journal of Human Genetics*, **2020**, 28, 783-789	5.3	3
56	Peritoneal dialysis induces alterations in the transcriptome of peritoneal cells before detectible peritoneal functional changes. *American Journal of Physiology - Renal Physiology*, **2020**, 318, F229-F237	4.3	7
55	Identification of Germline Mutations in Melanoma Patients with Early Onset, Double Primary Tumors, or Family Cancer History by NGS Analysis of 217 Genes. *Biomedicines*, **2020**, 8,	4.8	4
54	LncRNA Profiling Reveals That the Deregulation of H19, WT1-AS, TCL6, and LEF1-AS1 Is Associated with Higher-Risk Myelodysplastic Syndrome. *Cancers*, **2020**, 12,	6.6	6

53	Molecular Fingerprints of Borderline Changes in Kidney Allografts Are Influenced by Donor Category. *Frontiers in Immunology*, **2020**, 11, 423	8.4	1
52	Identification of deleterious germline CHEK2 mutations and their association with breast and ovarian cancer. *International Journal of Cancer*, **2019**, 145, 1782-1797	7.5	22
51	Parental gonadal but not somatic mosaicism leading to de novo NFIX variants shared by two brothers with Malan syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2119-2123	2.5	4
50	A novel variant of C12orf4 in a consanguineous Armenian family confirms the etiology of autosomal recessive intellectual disability type 66 with delineation of the phenotype. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e865	2.3	2
49	POLR3B-associated leukodystrophy: clinical, neuroimaging and molecular-genetic analyses in four patients: clinical heterogeneity and novel mutations in POLR3B gene. *Neurologia I Neurochirurgia Polska*, **2019**, 53, 369-376	1	
48	Genetic architecture of recent-onset dilated cardiomyopathy in Moravian region assessed by whole-exome sequencing and its clinical correlates. *Biomedical Papers of the Medical Faculty of the University Palacký, Olomouc, Czechoslovakia*, **2019**, 163, 309-317	1.7	1
47	Molecular Patterns Discriminate Accommodation and Subclinical Antibody-mediated Rejection in Kidney Transplantation. *Transplantation*, **2019**, 103, 909-917	1.8	7
46	Sideroblastic anemia associated with multisystem mitochondrial disorders. *Pediatric Blood and Cancer*, **2019**, 66, e27591	3	11
45	Rare copy number variation in extremely impulsively violent males. *Genes, Brain and Behavior*, **2019**, 18, e12536	3.6	3
44	Ectopic GRHL2 Expression Due to Non-coding Mutations Promotes Cell State Transition and Causes Posterior Polymorphous Corneal Dystrophy 4. *American Journal of Human Genetics*, **2018**, 102, 447-459	11	25
43	Mutations in Vps15 perturb neuronal migration in mice and are associated with neurodevelopmental disease in humans. *Nature Neuroscience*, **2018**, 21, 207-217	25.5	24
42	Clinical manifestations and molecular aspects of phosphoribosylpyrophosphate synthetase superactivity in females. *Rheumatology*, **2018**, 57, 1180-1185	3.9	6
41	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. *PLoS Genetics*, **2018**, 14, e1007671	6	11
40	Early isolated V-lesion may not truly represent rejection of the kidney allograft. *Clinical Science*, **2018**, 132, 2269-2284	6.5	8
39	LAMP2 exon-copy number variations in Danon disease heterozygote female probands: Infrequent or underdetected?. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2430-2434	2.5	5
38	Noninvasive Immunohistochemical Diagnosis and Novel Mutations Causing Autosomal Dominant Tubulointerstitial Kidney Disease. *Journal of the American Society of Nephrology: JASN*, **2018**, 29, 2418-2431	12.7	28
37	Validation of CZECANCA (CZEch CAncer paNel for Clinical Application) for targeted NGS-based analysis of hereditary cancer syndromes. *PLoS ONE*, **2018**, 13, e0195761	3.7	15
36	Thymidine kinase 2 and alanyl-tRNA synthetase 2 deficiencies cause lethal mitochondrial cardiomyopathy: case reports and review of the literature. *Cardiology in the Young*, **2017**, 27, 936-944	1	21

35	Segregation of a novel p.(Ser270Tyr) MAF mutation and p.(Tyr56*) CRYGD variant in a family with dominantly inherited congenital cataracts. *Molecular Biology Reports*, **2017**, 44, 435-440	2.8	5
34	Multiplex PCR and NGS-based identification of mRNA splicing variants: Analysis of BRCA1 splicing pattern as a model. *Gene*, **2017**, 637, 41-49	3.8	8
33	Analysis of 31-year-old patient with SYNGAP1 gene defect points to importance of variants in broader splice regions and reveals developmental trajectory of SYNGAP1-associated phenotype: case report. *BMC Medical Genetics*, **2017**, 18, 62	2.1	15
32	Acadian variant of Fanconi syndrome is caused by mitochondrial respiratory chain complex I deficiency due to a non-coding mutation in complex I assembly factor NDUFAF6. *Human Molecular Genetics*, **2016**, 25, 4062-4079	5.6	35
31	Rare variants in known and novel candidate genes predisposing to statin-associated myopathy. *Pharmacogenomics*, **2016**, 17, 1405-14	2.6	15
30	Autosomal-Dominant Corneal Endothelial Dystrophies CHED1 and PPCD1 Are Allelic Disorders Caused by Non-coding Mutations in the Promoter of OVOL2. *American Journal of Human Genetics*, **2016**, 98, 75-89	11	53
29	Heterozygous deletions at the ZEB1 locus verify haploinsufficiency as the mechanism of disease for posterior polymorphous corneal dystrophy type 3. *European Journal of Human Genetics*, **2016**, 24, 985-91	5.3	24
28	Molecular patterns of diffuse and nodular parathyroid hyperplasia in long-term hemodialysis. *American Journal of Physiology - Endocrinology and Metabolism*, **2016**, 311, E720-E729	6	4
27	Heterozygous Loss-of-Function SEC61A1 Mutations Cause Autosomal-Dominant Tubulo-Interstitial and Glomerulocystic Kidney Disease with Anemia. *American Journal of Human Genetics*, **2016**, 99, 174-87	11	87
26	Hereditary truncating mutations of DNA repair and other genes in BRCA1/BRCA2/PALB2-negatively tested breast cancer patients. *Clinical Genetics*, **2016**, 90, 324-33	4	29
25	Validation of rs2956540:G>C and rs3735520:G>A association with keratoconus in a population of European descent. *European Journal of Human Genetics*, **2015**, 23, 1581-3	5.3	27
24	Molecular diagnostics identifies risks for graft dysfunction despite borderline histologic changes. *Kidney International*, **2015**, 88, 785-95	9.9	11
23	A patient showing features of both SBBYSS and GPS supports the concept of a KAT6B-related disease spectrum, with mutations in mid-exon 18 possibly leading to combined phenotypes. *European Journal of Medical Genetics*, **2015**, 58, 550-5	2.6	22
22	Hereditary haemochromatosis caused by homozygous HJV mutation evolved through paternal disomy. *Clinical Genetics*, **2015**, 87, 96-8	4	1
21	Mutations in PNPLA6 are linked to photoreceptor degeneration and various forms of childhood blindness. *Nature Communications*, **2015**, 6, 5614	17.4	56
20	Mutation of Nogo-B receptor, a subunit of cis-prenyltransferase, causes a congenital disorder of glycosylation. *Cell Metabolism*, **2014**, 20, 448-57	24.6	72
19	Large copy number variations in combination with point mutations in the TYMP and SCO2 genes found in two patients with mitochondrial disorders. *European Journal of Human Genetics*, **2014**, 22, 431-4	5.3	8
18	Cerebellar dysfunction in a family harboring the PSEN1 mutation co-segregating with a cathepsin D variant p.A58V. *Journal of the Neurological Sciences*, **2013**, 326, 75-82	3.2	15

List of Publications

17	Bioinformatic perspectives in the neuronal ceroid lipofuscinoses. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2013**, 1832, 1831-41	6.9	8
16	Mutations in ANTXR1 cause GAPO syndrome. *American Journal of Human Genetics*, **2013**, 92, 792-9	11	60
15	Isolated X-linked hypertrophic cardiomyopathy caused by a novel mutation of the four-and-a-half LIM domain 1 gene. *Circulation: Cardiovascular Genetics*, **2013**, 6, 543-51		33
14	Ornithine carbamoyltransferase deficiency: molecular characterization of 29 families. *Clinical Genetics*, **2013**, 84, 552-9	4	5
13	Compensatory upregulation of respiratory chain complexes III and IV in isolated deficiency of ATP synthase due to TMEM70 mutation. *Biochimica Et Biophysica Acta - Bioenergetics*, **2012**, 1817, 1037-43	4.6	26
12	Large heterozygous deletions on 22q13.33 in combination with TYMP or SCO2 point mutations in two patients with mitochondrial disorders. *Biochimica Et Biophysica Acta - Bioenergetics*, **2012**, 1817, S143	4.6	
11	Adaptation of respiratory chain biogenesis to cytochrome c oxidase deficiency caused by SURF1 gene mutations. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2012**, 1822, 1114-24	6.9	25
10	Complete OATP1B1 and OATP1B3 deficiency causes human Rotor syndrome by interrupting conjugated bilirubin reuptake into the liver. *Journal of Clinical Investigation*, **2012**, 122, 519-28	15.9	265
9	DNA damage response and inflammatory signaling limit the MLL-ENL-induced leukemogenesis in vivo. *Cancer Cell*, **2012**, 21, 517-31	24.3	48
8	Mutations in DNAJC5, encoding cysteine-string protein alpha, cause autosomal-dominant adult-onset neuronal ceroid lipofuscinosis. *American Journal of Human Genetics*, **2011**, 89, 241-52	11	192
7	Mutations in DNAJC5, Encoding Cysteine-String Protein Alpha, Cause Autosomal-Dominant Adult-Onset Neuronal Ceroid Lipofuscinosis. *American Journal of Human Genetics*, **2011**, 89, 589	11	4
6	Dominant renin gene mutations associated with early-onset hyperuricemia, anemia, and chronic kidney failure. *American Journal of Human Genetics*, **2009**, 85, 204-13	11	119
5	HEPATIC GENE EXPRESSION IN PRAGUE HEREDITARY HYPERCHOLESTEROLEMIC (PHHC) RAT. *Atherosclerosis*, **2009**, 207, e12	3.1	
4	TMEM70 mutations cause isolated ATP synthase deficiency and neonatal mitochondrial encephalocardiomyopathy. *Nature Genetics*, **2008**, 40, 1288-90	36.3	161
3	Development of a human mitochondrial oligonucleotide microarray (h-MitoArray) and gene expression analysis of fibroblast cell lines from 13 patients with isolated F1Fo ATP synthase deficiency. *BMC Genomics*, **2008**, 9, 38	4.5	21
2	Rotor-type hyperbilirubinaemia has no defect in the canalicular bilirubin export pump. *Liver International*, **2007**, 27, 485-91	7.9	19
1	Mutations in TMEM76* cause mucopolysaccharidosis IIIC (Sanfilippo C syndrome). *American Journal of Human Genetics*, **2006**, 79, 807-19	11	61