

Viktor Stranecky

List of Publications by Citations

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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.

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	Complete OATP1B1 and OATP1B3 deficiency causes human Rotor syndrome by interrupting conjugated bilirubin reuptake into the liver. <i>Journal of Clinical Investigation</i> , 2012 , 122, 519-28	15.9	265
69	Mutations in DNAJC5, encoding cysteine-string protein alpha, cause autosomal-dominant adult-onset neuronal ceroid lipofuscinosis. <i>American Journal of Human Genetics</i> , 2011 , 89, 241-52	11	192
68	TMEM70 mutations cause isolated ATP synthase deficiency and neonatal mitochondrial encephalomyopathy. <i>Nature Genetics</i> , 2008 , 40, 1288-90	36.3	161
67	Dominant renin gene mutations associated with early-onset hyperuricemia, anemia, and chronic kidney failure. <i>American Journal of Human Genetics</i> , 2009 , 85, 204-13	11	119
66	Heterozygous Loss-of-Function SEC61A1 Mutations Cause Autosomal-Dominant Tubulo-Interstitial and Glomerulocystic Kidney Disease with Anemia. <i>American Journal of Human Genetics</i> , 2016 , 99, 174-87 ¹¹	11	87
65	Mutation of Nogo-B receptor, a subunit of cis-prenyltransferase, causes a congenital disorder of glycosylation. <i>Cell Metabolism</i> , 2014 , 20, 448-57	24.6	72
64	Mutations in TMEM76* cause mucopolysaccharidosis IIIC (Sanfilippo C syndrome). <i>American Journal of Human Genetics</i> , 2006 , 79, 807-19	11	61
63	Mutations in ANTXR1 cause GAPO syndrome. <i>American Journal of Human Genetics</i> , 2013 , 92, 792-9	11	60
62	Mutations in PNPLA6 are linked to photoreceptor degeneration and various forms of childhood blindness. <i>Nature Communications</i> , 2015 , 6, 5614	17.4	56
61	Autosomal-Dominant Corneal Endothelial Dystrophies CHED1 and PPCD1 Are Allelic Disorders Caused by Non-coding Mutations in the Promoter of OVOL2. <i>American Journal of Human Genetics</i> , 2016 , 98, 75-89	11	53
60	DNA damage response and inflammatory signaling limit the MLL-ENL-induced leukemogenesis in vivo. <i>Cancer Cell</i> , 2012 , 21, 517-31	24.3	48
59	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. <i>Human Molecular Genetics</i> , 2016 , 25, 4062-4079	5.6	35
58	Isolated X-linked hypertrophic cardiomyopathy caused by a novel mutation of the four-and-a-half LIM domain 1 gene. <i>Circulation: Cardiovascular Genetics</i> , 2013 , 6, 543-51		33
57	Hereditary truncating mutations of DNA repair and other genes in BRCA1/BRCA2/PALB2-negatively tested breast cancer patients. <i>Clinical Genetics</i> , 2016 , 90, 324-33	4	29
56	Noninvasive Immunohistochemical Diagnosis and Novel Mutations Causing Autosomal Dominant Tubulointerstitial Kidney Disease. <i>Journal of the American Society of Nephrology: JASN</i> , 2018 , 29, 2418-2431 ^{12,7}	12.7	28
55	Validation of rs2956540:G>C and rs3735520:G>A association with keratoconus in a population of European descent. <i>European Journal of Human Genetics</i> , 2015 , 23, 1581-3	5.3	27
54	Compensatory upregulation of respiratory chain complexes III and IV in isolated deficiency of ATP synthase due to TMEM70 mutation. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2012 , 1817, 1037-43	4.6	26

53	Ectopic GRHL2 Expression Due to Non-coding Mutations Promotes Cell State Transition and Causes Posterior Polymorphous Corneal Dystrophy 4. <i>American Journal of Human Genetics</i> , 2018 , 102, 447-459	11	25
52	Adaptation of respiratory chain biogenesis to cytochrome c oxidase deficiency caused by SURF1 gene mutations. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2012 , 1822, 1114-24	6.9	25
51	Mutations in Vps15 perturb neuronal migration in mice and are associated with neurodevelopmental disease in humans. <i>Nature Neuroscience</i> , 2018 , 21, 207-217	25.5	24
50	Heterozygous deletions at the ZEB1 locus verify haploinsufficiency as the mechanism of disease for posterior polymorphous corneal dystrophy type 3. <i>European Journal of Human Genetics</i> , 2016 , 24, 985-993	5.3	24
49	Identification of deleterious germline CHEK2 mutations and their association with breast and ovarian cancer. <i>International Journal of Cancer</i> , 2019 , 145, 1782-1797	7.5	22
48	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. <i>European Journal of Medical Genetics</i> , 2015 , 58, 550-5	2.6	22
47	Thymidine kinase 2 and alanyl-tRNA synthetase 2 deficiencies cause lethal mitochondrial cardiomyopathy: case reports and review of the literature. <i>Cardiology in the Young</i> , 2017 , 27, 936-944	1	21
46	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. <i>BMC Genomics</i> , 2008 , 9, 38	4.5	21
45	Rotor-type hyperbilirubinaemia has no defect in the canalicular bilirubin export pump. <i>Liver International</i> , 2007 , 27, 485-91	7.9	19
44	Rare variants in known and novel candidate genes predisposing to statin-associated myopathy. <i>Pharmacogenomics</i> , 2016 , 17, 1405-14	2.6	15
43	Cerebellar dysfunction in a family harboring the PSEN1 mutation co-segregating with a cathepsin D variant p.A58V. <i>Journal of the Neurological Sciences</i> , 2013 , 326, 75-82	3.2	15
42	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. <i>BMC Medical Genetics</i> , 2017 , 18, 62	2.1	15
41	Validation of CZE CANCA (CZEch CANcer paNel for Clinical Application) for targeted NGS-based analysis of hereditary cancer syndromes. <i>PLoS ONE</i> , 2018 , 13, e0195761	3.7	15
40	Molecular diagnostics identifies risks for graft dysfunction despite borderline histologic changes. <i>Kidney International</i> , 2015 , 88, 785-95	9.9	11
39	Sideroblastic anemia associated with multisystem mitochondrial disorders. <i>Pediatric Blood and Cancer</i> , 2019 , 66, e27591	3	11
38	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. <i>PLoS Genetics</i> , 2018 , 14, e1007671	6	11
37	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. <i>Journal of Inherited Metabolic Disease</i> , 2020 , 43, 694-700	5.4	8
36	Multigene Panel Germline Testing of 1333 Czech Patients with Ovarian Cancer. <i>Cancers</i> , 2020 , 12,	6.6	8

35	Bioinformatic perspectives in the neuronal ceroid lipofuscinoses. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2013 , 1832, 1831-41	6.9	8
34	Multiplex PCR and NGS-based identification of mRNA splicing variants: Analysis of BRCA1 splicing pattern as a model. <i>Gene</i> , 2017 , 637, 41-49	3.8	8
33	Large copy number variations in combination with point mutations in the TYMP and SCO2 genes found in two patients with mitochondrial disorders. <i>European Journal of Human Genetics</i> , 2014 , 22, 431-4	5.3	8
32	Early isolated V-lesion may not truly represent rejection of the kidney allograft. <i>Clinical Science</i> , 2018 , 132, 2269-2284	6.5	8
31	Spinal muscular atrophy caused by a novel Alu-mediated deletion of exons 2a-5 in SMN1 undetectable with routine genetic testing. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1238	2.3	7
30	Desminopathy: Novel Desmin Variants, a New Cardiac Phenotype, and Further Evidence for Secondary Mitochondrial Dysfunction. <i>Journal of Clinical Medicine</i> , 2020 , 9,	5.1	7
29	Peritoneal dialysis induces alterations in the transcriptome of peritoneal cells before detectible peritoneal functional changes. <i>American Journal of Physiology - Renal Physiology</i> , 2020 , 318, F229-F237	4.3	7
28	Molecular Patterns Discriminate Accommodation and Subclinical Antibody-mediated Rejection in Kidney Transplantation. <i>Transplantation</i> , 2019 , 103, 909-917	1.8	7
27	Clinical manifestations and molecular aspects of phosphoribosylpyrophosphate synthetase superactivity in females. <i>Rheumatology</i> , 2018 , 57, 1180-1185	3.9	6
26	LncRNA Profiling Reveals That the Dereglulation of H19, WT1-AS, TCL6, and LEF1-AS1 Is Associated with Higher-Risk Myelodysplastic Syndrome. <i>Cancers</i> , 2020 , 12,	6.6	6
25	Segregation of a novel p.(Ser270Tyr) MAF mutation and p.(Tyr56*) CRYGD variant in a family with dominantly inherited congenital cataracts. <i>Molecular Biology Reports</i> , 2017 , 44, 435-440	2.8	5
24	Ornithine carbamoyltransferase deficiency: molecular characterization of 29 families. <i>Clinical Genetics</i> , 2013 , 84, 552-9	4	5
23	LAMP2 exon-copy number variations in Danon disease heterozygote female probands: Infrequent or underdetected?. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 2430-2434	2.5	5
22	Parental gonadal but not somatic mosaicism leading to de novo NFIX variants shared by two brothers with Malan syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 2119-2123	2.5	4
21	Mutations in DNAJC5, Encoding Cysteine-String Protein Alpha, Cause Autosomal-Dominant Adult-Onset Neuronal Ceroid Lipofuscinosis. <i>American Journal of Human Genetics</i> , 2011 , 89, 589	11	4
20	Molecular patterns of diffuse and nodular parathyroid hyperplasia in long-term hemodialysis. <i>American Journal of Physiology - Endocrinology and Metabolism</i> , 2016 , 311, E720-E729	6	4
19	Identification of Germline Mutations in Melanoma Patients with Early Onset, Double Primary Tumors, or Family Cancer History by NGS Analysis of 217 Genes. <i>Biomedicine</i> , 2020 , 8,	4.8	4
18	Biallelic loss-of-function variants in PLD1 cause congenital right-sided cardiac valve defects and neonatal cardiomyopathy. <i>Journal of Clinical Investigation</i> , 2021 , 131,	15.9	4

17	Autosomal-dominant adult neuronal ceroid lipofuscinosis caused by duplication in DNAJC5 initially missed by Sanger and whole-exome sequencing. <i>European Journal of Human Genetics</i> , 2020 , 28, 783-789	5.3	3
16	Rare copy number variation in extremely impulsively violent males. <i>Genes, Brain and Behavior</i> , 2019 , 18, e12536	3.6	3
15	POLRMT mutations impair mitochondrial transcription causing neurological disease. <i>Nature Communications</i> , 2021 , 12, 1135	17.4	3
14	A mutation in the SAA1 promoter causes hereditary amyloid A amyloidosis. <i>Kidney International</i> , 2021 ,	9.9	3
13	Fatal neonatal nephrocutaneous syndrome in 18 Roma children with EGFR deficiency. <i>Journal of Dermatology</i> , 2020 , 47, 663-668	1.6	2
12	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. <i>Molecular Genetics & Genomic Medicine</i> , 2019 , 7, e865	2.3	2
11	GPD1 Deficiency - Underdiagnosed Cause of Liver Disease. <i>Indian Journal of Pediatrics</i> , 2021 , 88, 80-81	3	2
10	Pseudodominant Nanophthalmos in a Roma Family Caused by a Novel Variant. <i>Journal of Ophthalmology</i> , 2020 , 2020, 6807809	2	1
9	Hereditary haemochromatosis caused by homozygous HJV mutation evolved through paternal disomy. <i>Clinical Genetics</i> , 2015 , 87, 96-8	4	1
8	Genetic architecture of recent-onset dilated cardiomyopathy in Moravian region assessed by whole-exome sequencing and its clinical correlates. <i>Biomedical Papers of the Medical Faculty of the University Palacky&#x0301;, Olomouc, Czechoslovakia</i> , 2019 , 163, 309-317	1.7	1
7	Genetic heterogeneity of neuronal intranuclear inclusion disease: What about the infantile variant?. <i>Annals of Clinical and Translational Neurology</i> , 2021 , 8, 994-1001	5.3	1
6	Molecular Fingerprints of Borderline Changes in Kidney Allografts Are Influenced by Donor Category. <i>Frontiers in Immunology</i> , 2020 , 11, 423	8.4	1
5	Non-Penetrance for Ocular Phenotype in Two Individuals Carrying Heterozygous Loss-of-Function Alleles. <i>Genes</i> , 2021 , 12,	4.2	0
4	Large heterozygous deletions on 22q13.33 in combination with TYMP or SCO2 point mutations in two patients with mitochondrial disorders. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2012 , 1817, S143	4.6	
3	HEPATIC GENE EXPRESSION IN PRAGUE HEREDITARY HYPERCHOLESTEROLEMIC (PHHC) RAT. <i>Atherosclerosis</i> , 2009 , 207, e12	3.1	
2	POLR3B-associated leukodystrophy: clinical, neuroimaging and molecular-genetic analyses in four patients: clinical heterogeneity and novel mutations in POLR3B gene. <i>Neurologia I Neurochirurgia Polska</i> , 2019 , 53, 369-376	1	
1	Re: ERCC3, a new ovarian cancer susceptibility gene?. <i>European Journal of Cancer</i> , 2021 , 150, 278-280	7.5	