Viktor Stranecky

List of Publications by Year in descending order

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73 papers	2,518 citations	236612 25 h-index	205818 48 g-index
78 all docs	78 docs citations	78 times ranked	4955 citing authors

#	Article	IF	Citations
1	Complete OATP1B1 and OATP1B3 deficiency causes human Rotor syndrome by interrupting conjugated bilirubin reuptake into the liver. Journal of Clinical Investigation, 2012, 122, 519-528.	3.9	321
2	Mutations in DNAJC5, Encoding Cysteine-String Protein Alpha, Cause Autosomal-Dominant Adult-Onset Neuronal Ceroid Lipofuscinosis. American Journal of Human Genetics, 2011, 89, 241-252.	2.6	236
3	TMEM70 mutations cause isolated ATP synthase deficiency and neonatal mitochondrial encephalocardiomyopathy. Nature Genetics, 2008, 40, 1288-1290.	9.4	183
4	Dominant Renin Gene Mutations Associated with Early-Onset Hyperuricemia, Anemia, and Chronic Kidney Failure. American Journal of Human Genetics, 2009, 85, 204-213.	2.6	146
5	Heterozygous Loss-of-Function SEC61A1 Mutations Cause Autosomal-Dominant Tubulo-Interstitial and Glomerulocystic Kidney Disease with Anemia. American Journal of Human Genetics, 2016, 99, 174-187.	2.6	124
6	Mutation of Nogo-B Receptor, a Subunit of cis-Prenyltransferase, Causes a Congenital Disorder of Glycosylation. Cell Metabolism, 2014, 20, 448-457.	7.2	104
7	Mutations in TMEM76* Cause Mucopolysaccharidosis IIIC (Sanfilippo C Syndrome). American Journal of Human Genetics, 2006, 79, 807-819.	2.6	77
8	Mutations in PNPLA6 are linked to photoreceptor degeneration and various forms of childhood blindness. Nature Communications, 2015, 6, 5614.	5.8	77
9	Mutations in ANTXR1 Cause GAPO Syndrome. American Journal of Human Genetics, 2013, 92, 792-799.	2.6	73
10	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.	2.6	70
11	Identification of deleterious germline CHEK2 mutations and their association with breast and ovarian cancer. International Journal of Cancer, 2019, 145, 1782-1797.	2.3	62
12	DNA Damage Response and Inflammatory Signaling Limit the MLL-ENL-Induced Leukemogenesis InÂVivo. Cancer Cell, 2012, 21, 517-531.	7.7	56
13	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.	1.4	55
14	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.	2.6	45
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, 543-551.	5.1	43
16	Multiplex PCR and NGS-based identification of mRNA splicing variants: Analysis of BRCA1 splicing pattern as a model. Gene, 2017, 637, 41-49.	1.0	43
17	Hereditary truncating mutations of <scp>DNA</scp> repair and other genes in <scp>BRCA1</scp>/<scp>BRCA2</scp>/<scp>PALB2</scp>â€negatively tested breast cancer patients. Clinical Genetics, 2016, 90, 324-333.	1.0	38
18	Noninvasive Immunohistochemical Diagnosis and Novel MUC1 Mutations Causing Autosomal Dominant Tubulointerstitial Kidney Disease. Journal of the American Society of Nephrology: JASN, 2018, 29, 2418-2431.	3.0	38

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19	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, 1581-1583.	1.4	34
20	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, 985-991.	1.4	33
21	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, 1037-1043.	0.5	32
22	Validation of CZECANCA (CZEch CAncer paNel for Clinical Application) for targeted NGS-based analysis of hereditary cancer syndromes. PLoS ONE, 2018, 13, e0195761.	1.1	31
23	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, 1114-1124.	1.8	30
24	Mutations in Vps15 perturb neuronal migration in mice and are associated with neurodevelopmental disease in humans. Nature Neuroscience, 2018, 21, 207-217.	7.1	30
25	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.	0.4	28
26	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, 550-555.	0.7	25
27	Desminopathy: Novel Desmin Variants, a New Cardiac Phenotype, and Further Evidence for Secondary Mitochondrial Dysfunction. Journal of Clinical Medicine, 2020, 9, 937.	1.0	24
28	Rotor-type hyperbilirubinaemia has no defect in the canalicular bilirubin export pump. Liver International, 2007, 27, 485-491.	1.9	22
29	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.	1.2	22
30	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	22
31	Molecular diagnostics identifies risks for graft dysfunction despite borderline histologic changes. Kidney International, 2015, 88, 785-795.	2.6	21
32	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	5.8	21
33	DNAJC30 defect: a frequent cause of recessive Leber hereditary optic neuropathy and Leigh syndrome. Brain, 2022, 145, 1624-1631.	3.7	21
34	Multigene Panel Germline Testing of 1333 Czech Patients with Ovarian Cancer. Cancers, 2020, 12, 956.	1.7	19
35	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.	0.3	18
36	Rare variants in known and novel candidate genes predisposing to statin-associated myopathy. Pharmacogenomics, 2016, 17, 1405-1414.	0.6	17

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37	LncRNA Profiling Reveals That the Deregulation of H19, WT1-AS, TCL6, and LEF1-AS1 Is Associated with Higher-Risk Myelodysplastic Syndrome. Cancers, 2020, 12, 2726.	1.7	17
38	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.	1.5	16
39	Biallelic loss-of-function variants in PLD1 cause congenital right-sided cardiac valve defects and neonatal cardiomyopathy. Journal of Clinical Investigation, 2021, 131, .	3.9	16
40	Severe phenotype of <scp>ATP6AP1â€CDG</scp> in two siblings with a novel mutation leading to a differential tissueâ€specific <scp>ATP6AP1</scp> protein pattern, cellular oxidative stress and hepatic copper accumulation. Journal of Inherited Metabolic Disease, 2020, 43, 694-700.	1.7	14
41	Early isolated V-lesion may not truly represent rejection of the kidney allograft. Clinical Science, 2018, 132, 2269-2284.	1.8	13
42	Sideroblastic anemia associated with multisystem mitochondrial disorders. Pediatric Blood and Cancer, 2019, 66, e27591.	0.8	13
43	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.	1.3	13
44	Clinical manifestations and molecular aspects of phosphoribosylpyrophosphate synthetase superactivity in females. Rheumatology, 2018, 57, 1180-1185.	0.9	12
45	Molecular Patterns Discriminate Accommodation and Subclinical Antibody-mediated Rejection in Kidney Transplantation. Transplantation, 2019, 103, 909-917.	0.5	12
46	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, 431-434.	1.4	11
47	Bioinformatic perspectives in the neuronal ceroid lipofuscinoses. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2013, 1832, 1831-1841.	1.8	10
48	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.	0.7	10
49	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.	1.4	10
50	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, 404.	1.4	10
51	Spinal muscular atrophy caused by a novel Aluâ€mediated deletion of exons 2aâ€5 in SMN1 undetectable with routine genetic testing. Molecular Genetics & Genomic Medicine, 2020, 8, e1238.	0.6	10
52	A mutation in the SAA1 promoter causes hereditary amyloid A amyloidosis. Kidney International, 2022, 101, 349-359.	2.6	10
53	Segregation of aÂnovel p.(Ser270Tyr) MAF mutation and p.(Tyr56âˆ—) CRYGD variant in a family with dominantly inherited congenital cataracts. Molecular Biology Reports, 2017, 44, 435-440.	1.0	9
54	LAMP2 exonâ€copy number variations in Danon disease heterozygote female probands: Infrequent or underdetected?. American Journal of Medical Genetics, Part A, 2018, 176, 2430-2434.	0.7	9

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55	Rare copy number variation in extremely impulsively violent males. Genes, Brain and Behavior, 2019, 18, e12536.	1.1	9
56	Molecular patterns of diffuse and nodular parathyroid hyperplasia in long-term hemodialysis. American Journal of Physiology - Endocrinology and Metabolism, 2016, 311, E720-E729.	1.8	8
57	Ornithine carbamoyltransferase deficiency: molecular characterization of 29 families. Clinical Genetics, 2013, 84, 552-559.	1.0	6
58	Fatal neonatal nephrocutaneous syndrome in 18 Roma children with EGFR deficiency. Journal of Dermatology, 2020, 47, 663-668.	0.6	6
59	Molecular Fingerprints of Borderline Changes in Kidney Allografts Are Influenced by Donor Category. Frontiers in Immunology, 2020, 11, 423.	2.2	5
60	Mutations in DNAJC5, Encoding Cysteine-String Protein Alpha, Cause Autosomal-Dominant Adult-Onset Neuronal Ceroid Lipofuscinosis. American Journal of Human Genetics, 2011, 89, 589.	2.6	4
61	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.	0.6	4
62	GPD1 Deficiency â€“ Underdiagnosed Cause of Liver Disease. Indian Journal of Pediatrics, 2021, 88, 80-81.	0.3	4
63	Pseudodominant Nanophthalmos in a Roma Family Caused by a Novel PRSS56 Variant. Journal of Ophthalmology, 2020, 2020, 1-9.	0.6	3
64	Non-Penetrance for Ocular Phenotype in Two Individuals Carrying Heterozygous Loss-of-Function ZEB1 Alleles. Genes, 2021, 12, 677.	1.0	3
65	Genetic heterogeneity of neuronal intranuclear inclusion disease: What about the infantile variant?. Annals of Clinical and Translational Neurology, 2021, 8, 994-1001.	1.7	2
66	Germline CHEK2 Gene Mutations in Hereditary Breast Cancer Predisposition â€“ Mutation Types and their Biological and Clinical Relevance. Klinicka Onkologie, 2019, 32, 36-50.	0.1	2
67	Hereditary haemochromatosis caused by homozygous <scp>HJV</scp> mutation evolved through paternal disomy. Clinical Genetics, 2015, 87, 96-98.	1.0	1
68	Re: ERCC3, a new ovarian cancer susceptibility gene?. European Journal of Cancer, 2021, 150, 278-280.	1.3	1
69	Contribution of Massive Parallel Sequencing to Diagnosis of Hereditary Ovarian Cancer in the Czech Republic. Klinicka Onkologie, 2019, 32, 72-78.	0.1	1
70	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.	0.2	1
71	HEPATIC GENE EXPRESSION IN PRAGUE HEREDITARY HYPERCHOLESTEROLEMIC (PHHC) RAT. Atherosclerosis, 2009, 207, e12.	0.4	0
72	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.	0.5	0

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73	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.	0.6	0