Stanislav Kmoch

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5382065/publications.pdf

Version: 2024-02-01

100 papers	4,478 citations	109321 35 h-index	118850 62 g-index
103 all docs	103 docs citations	103 times ranked	6328 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.	8.2	321
2	Autosomal dominant tubulointerstitial kidney disease: diagnosis, classification, and managementâ€"A KDIGO consensus report. Kidney International, 2015, 88, 676-683.	5.2	276
3	Mutations causing medullary cystic kidney disease type 1 lie in a large VNTR in MUC1 missed by massively parallel sequencing. Nature Genetics, 2013, 45, 299-303.	21.4	237
4	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.	6.2	236
5	TMEM70 mutations cause isolated ATP synthase deficiency and neonatal mitochondrial encephalocardiomyopathy. Nature Genetics, 2008, 40, 1288-1290.	21.4	183
6	Dominant Renin Gene Mutations Associated with Early-Onset Hyperuricemia, Anemia, and Chronic Kidney Failure. American Journal of Human Genetics, 2009, 85, 204-213.	6.2	146
7	Autosomal dominant tubulointerstitial kidney disease. Nature Reviews Disease Primers, 2019, 5, 60.	30.5	139
8	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.	6.2	124
9	Small Molecule Targets TMED9 and Promotes Lysosomal Degradation to Reverse Proteinopathy. Cell, 2019, 178, 521-535.e23.	28.9	124
10	Adenylosuccinate lyase deficiency. Journal of Inherited Metabolic Disease, 2015, 38, 231-242.	3.6	119
11	Mutation of Nogo-B Receptor, a Subunit of cis-Prenyltransferase, Causes a Congenital Disorder of Glycosylation. Cell Metabolism, 2014, 20, 448-457.	16.2	104
12	HIF and reactive oxygen species regulate oxidative phosphorylation in cancer. Carcinogenesis, 2008, 29, 1528-1537.	2.8	84
13	Mutations in TMEM76* Cause Mucopolysaccharidosis IIIC (Sanfilippo C Syndrome). American Journal of Human Genetics, 2006, 79, 807-819.	6.2	77
14	Clinical and genetic spectra of autosomal dominant tubulointerstitial kidney disease due to mutationsÂin UMOD and MUC1. Kidney International, 2020, 98, 717-731.	5.2	75
15	Mutations in ANTXR1 Cause GAPO Syndrome. American Journal of Human Genetics, 2013, 92, 792-799.	6.2	73
16	Mitochondrial encephalocardio-myopathy with early neonatal onset due to TMEM70 mutation. Archives of Disease in Childhood, 2010, 95, 296-301.	1.9	72
17	Uromodulin Biology and Pathophysiology â€“ An Update. Kidney and Blood Pressure Research, 2010, 33, 456-475.	2.0	71
18	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.	6.2	70

#	Article	IF	Citations
19	Posterior Polymorphous Corneal Dystrophy in Czech Families Maps to Chromosome 20 and Excludes theVSX1Gene. , 2005, 46, 4480.		67
20	Variable Clinical Presentation of an MUC1 Mutation Causing Medullary Cystic Kidney Disease Type 1. Clinical Journal of the American Society of Nephrology: CJASN, 2014, 9, 527-535.	4.5	65
21	Clinical, biochemical and molecular findings in seven Polish patients with adenylosuccinate lyase deficiency. Molecular Genetics and Metabolism, 2008, 94, 435-442.	1.1	62
22	Mutations of ATIC and ADSL affect purinosome assembly in cultured skin fibroblasts from patients with AICA-ribosiduria and ADSL deficiency. Human Molecular Genetics, 2012, 21, 1534-1543.	2.9	62
23	Identification of deleterious germline CHEK2 mutations and their association with breast and ovarian cancer. International Journal of Cancer, 2019, 145, 1782-1797.	5.1	62
24	Autosomal Dominant Tubulointerstitial Kidney Disease. Advances in Chronic Kidney Disease, 2017, 24, 86-93.	1.4	60
25	A Survey of the Newborn Populations in Belgium, Germany, Poland, Czech Republic, Hungary, Bulgaria, Spain, Turkey, and Japan for the G985 Variant Allele with Haplotype Analysis at the Medium Chain Acyl-CoA. Pediatric Research, 1997, 41, 201-209.	2.3	60
26	Lethal Fetal and Early Neonatal Presentation of Adenylosuccinate Lyase Deficiency: Observation of 6 Patients in 4 Families. Journal of Pediatrics, 2007, 150, 57-61.e2.	1.8	58
27	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.	2.9	55
28	Familial Juvenile Hyperuricemic Nephropathy: Localization of the Gene on Chromosome 16p11.2â€"and Evidence for Genetic Heterogeneity. American Journal of Human Genetics, 2000, 66, 1989-1994.	6.2	51
29	Sanfilippo syndrome type C: mutation spectrum in the heparan sulfate acetyl-CoA: Î±-glucosaminide N-acetyltransferase (HGSNAT) gene. Human Mutation, 2009, 30, 918-925.	2.5	51
30	Biochemical and structural analysis of 14 mutant adsl enzyme complexes and correlation to phenotypic heterogeneity of adenylosuccinate lyase deficiency. Human Mutation, 2010, 31, 445-455.	2.5	45
31	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.	6.2	45
32	Uromodulin-Associated Kidney Disease. Nephron Clinical Practice, 2010, 118, c31-c36.	2.3	44
33	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
34	Autosomal dominant tubulointerstitial kidney disease-UMOD is the most frequent non polycystic genetic kidney disease. BMC Nephrology, 2018, 19, 301.	1.8	39
35	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.	6.1	38
36	TMEM70 protein â€" A novel ancillary factor of mammalian ATP synthase. Biochimica Et Biophysica Acta - Bioenergetics, 2009, 1787, 529-532.	1.0	37

#	Article	IF	Citations
37	Adhesion of osteoblasts on chemically patterned nanocrystalline diamonds. Physica Status Solidi (B): Basic Research, 2008, 245, 2124-2127.	1.5	36
38	Hereditary Interstitial Kidney Disease. Seminars in Nephrology, 2010, 30, 366-373.	1.6	36
39	The clinical, biochemical and genetic features associated withRMND1-related mitochondrial disease. Journal of Medical Genetics, 2016, 53, 768-775.	3.2	35
40	CRISPR-Cas9 induced mutations along de novo purine synthesis in HeLa cells result in accumulation of individual enzyme substrates and affect purinosome formation. Molecular Genetics and Metabolism, 2016, 119, 270-277.	1.1	33
41	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.	1.0	32
42	Elevated urinary CRELD2 is associated with endoplasmic reticulum stressâ€“mediated kidney disease. JCI Insight, 2017, 2, .	5.0	32
43	Autosomal dominant tubulointerstitial kidney disease: of names and genes. Kidney International, 2014, 86, 459-461.	5.2	31
44	Validation of CZECANCA (CZEch CAncer paNel for Clinical Application) for targeted NGS-based analysis of hereditary cancer syndromes. PLoS ONE, 2018, 13, e0195761.	2.5	31
45	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.	3.8	30
46	Genetic and Clinical Predictors of Age of ESKD in Individuals With Autosomal Dominant Tubulointerstitial Kidney Disease Due to UMOD Mutations. Kidney International Reports, 2020, 5, 1472-1485.	0.8	30
47	Mapping of a new candidate locus for uromodulin-associated kidney disease (UAKD) to chromosome 1q41. Kidney International, 2005, 68, 1472-1482.	5.2	28
48	Diagnosis and misdiagnosis of adult neuronal ceroid lipofuscinosis (Kufs disease). Neurology, 2016, 87, 579-584.	1.1	28
49	An international cohort study of autosomal dominant tubulointerstitial kidney disease due to mutations identifies distinct clinical subtypes. Kidney International, 2020, 98, 1589-1604.	5.2	27
50	Expression and processing of the TMEM70 protein. Biochimica Et Biophysica Acta - Bioenergetics, 2011, 1807, 144-149.	1.0	26
51	Familial juvenile hyperuricaemic nephropathy (FJHN): linkage analysis in 15 families, physical and transcriptional characterisation of the FJHN critical region on chromosome 16p11.2 and the analysis of seven candidate genes. European Journal of Human Genetics, 2003, 11, 145-154.	2.8	25
52	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.	1.3	25
53	Development and Validation of a Mass Spectrometryâ€“Based Assay for the Molecular Diagnosis of Mucin-1 Kidney Disease. Journal of Molecular Diagnostics, 2016, 18, 566-571.	2.8	25
54	Desminopathy: Novel Desmin Variants, a New Cardiac Phenotype, and Further Evidence for Secondary Mitochondrial Dysfunction. Journal of Clinical Medicine, 2020, 9, 937.	2.4	24

#	Article	IF	Citations
55	Rotor-type hyperbilirubinaemia has no defect in the canalicular bilirubin export pump. Liver International, 2007, 27, 485-491.	3.9	22
56	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.	2.8	22
57	PAICS deficiency, a new defect of de novo purine synthesis resulting in multiple congenital anomalies and fatal outcome. Human Molecular Genetics, 2019, 28, 3805-3814.	2.9	22
58	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
59	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	12.8	21
60	Genetic Etiologies for Chronic Kidney Disease Revealed through Next-Generation Renal Gene Panel. American Journal of Nephrology, 2022, 53, 297-306.	3.1	21
61	Identification and determination of succinyladenosine in human cerebrospinal fluid. Biomedical Applications, 1999, 726, 53-58.	1.7	19
62	Multigene Panel Germline Testing of 1333 Czech Patients with Ovarian Cancer. Cancers, 2020, 12, 956.	3.7	19
63	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.	0.6	18
64	Tamm Horsfall Glycoprotein and Uromodulin. Clinical Journal of the American Society of Nephrology: CJASN, 2016, 11, 6-8.	4.5	18
65	Rare variants in known and novel candidate genes predisposing to statin-associated myopathy. Pharmacogenomics, 2016, 17, 1405-1414.	1.3	17
66	Biallelic loss-of-function variants in PLD1 cause congenital right-sided cardiac valve defects and neonatal cardiomyopathy. Journal of Clinical Investigation, 2021, 131, .	8.2	16
67	Genetic and metabolomic analysis of AdeD and AdeI mutants of de novo purine biosynthesis: Cellular models of de novo purine biosynthesis deficiency disorders. Molecular Genetics and Metabolism, 2013, 108, 178-189.	1.1	14
68	Ntrk1 mutation co-segregating with bipolar disorder and inherited kidney disease in a multiplex family causes defects in neuronal growth and depression-like behavior in mice. Translational Psychiatry, 2020, 10, 407.	4.8	14
69	The utility of a genetic kidney disease clinic employing a broad range of genomic testing platforms: experience of the Irish Kidney Gene Project. Journal of Nephrology, 2022, 35, 1655-1665.	2.0	14
70	Detection of viral infections by an oligonucleotide microarray. Journal of Virological Methods, 2010, 165, 64-70.	2.1	12
71	Clinical manifestations and molecular aspects of phosphoribosylpyrophosphate synthetase superactivity in females. Rheumatology, 2018, 57, 1180-1185.	1.9	12
72	Outcomes of patient self-referral for the diagnosis of several rare inherited kidney diseases. Genetics in Medicine, 2020, 22, 142-149.	2.4	11

#	Article	IF	Citations
73	Bioinformatic perspectives in the neuronal ceroid lipofuscinoses. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2013, 1832, 1831-1841.	3.8	10
74	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.	2.8	10
75	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.	1.2	10
76	A mutation in the SAA1 promoter causes hereditary amyloid A amyloidosis. Kidney International, 2022, 101, 349-359.	5.2	10
77	Molecular characterization of the AdeI mutant of Chinese hamster ovary cells: A cellular model of adenylosuccinate lyase deficiency. Molecular Genetics and Metabolism, 2011, 102, 61-68.	1.1	9
78	Teenage-onset progressive myoclonic epilepsy due to a familial C9orf72 repeat expansion. Neurology, 2018, 90, e658-e663.	1.1	9
79	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.	1.2	9
80	Rare copy number variation in extremely impulsively violent males. Genes, Brain and Behavior, 2019, 18, e12536.	2.2	9
81	Alu â€mediated Xq24 deletion encompassing CUL4B , LAMP2 , ATP1B4 , TMEM255A , and ZBTB33 genes causes Danon disease in a female patient. American Journal of Medical Genetics, Part A, 2020, 182, 219-223.	1.2	9
82	Phenylbutyrate rescues the transport defect of the Sec61Î± mutations V67G and T185A for renin. Life Science Alliance, 2022, 5, e202101150.	2.8	9
83	Study of purinosome assembly in cell-based model systems with de novo purine synthesis and salvage pathway deficiencies. PLoS ONE, 2018, 13, e0201432.	2.5	8
84	Autosomal dominant tubulointerstitial kidney disease: more than just HNF1Î². Pediatric Nephrology, 2021, , 1.	1.7	8
85	The need for vigilance: False-negative screening for adenylosuccinate lyase deficiency caused by deribosylation of urinary biomarkers. Clinical Biochemistry, 2013, 46, 1899-1901.	1.9	7
86	Screening for adenylosuccinate lyase deficiency using tandem mass spectrometry analysis of succinylpurines in neonatal dried blood spots. Clinical Biochemistry, 2015, 48, 2-7.	1.9	7
87	Ultrabright plasmonic fluor nanolabel-enabled detection of a urinary ER stress biomarker in autosomal dominant tubulointerstitial kidney disease. American Journal of Physiology - Renal Physiology, 2021, 321, F236-F244.	2.7	5
88	Mitochondriopathy Manifesting as Inherited Tubulointerstitial Nephropathy Without Symptomatic Other Organ Involvement. Kidney International Reports, 2021, 6, 2514-2518.	0.8	5
89	Mutations in DNAJC5, Encoding Cysteine-String Protein Alpha, Cause Autosomal-Dominant Adult-Onset Neuronal Ceroid Lipofuscinosis. American Journal of Human Genetics, 2011, 89, 589.	6.2	4
90	Plasma Mucin-1 (CA15-3) Levels in Autosomal Dominant Tubulointerstitial Kidney Disease due to <i>MUC1</i> Mutations. American Journal of Nephrology, 2021, 52, 378-387.	3.1	4

#	Article	IF	Citations
91	The Varied Clinical Presentation of Autosomal Dominant Tubulointerstitial Kidney Disease Due to HNF1Î² Mutations. Kidney International Reports, 2020, 5, 2133-2135.	0.8	3
92	Non-Penetrance for Ocular Phenotype in Two Individuals Carrying Heterozygous Loss-of-Function ZEB1 Alleles. Genes, 2021, 12, 677.	2.4	3
93	Urinary Pterins in Lesch-Nyhan Syndrome. Advances in Experimental Medicine and Biology, 1991, 309B, 261-264.	1.6	3
94	Renal transplant outcomes in patients with autosomal dominant tubulointerstitial kidney disease. Clinical Transplantation, 2020, 34, e13783.	1.6	2
95	Genetic disorders of mitochondrial ATP synthase. Biochimica Et Biophysica Acta - Bioenergetics, 2010, 1797, 47-48.	1.0	1
96	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.	0.6	1
97	Gout: A Step Forward. Advances in Chronic Kidney Disease, 2012, 19, 356-357.	1.4	0
98	Elucidation of a novel mechanism for faulty protein retention and a therapeutic strategy for facilitated lysosomal removal. FASEB Journal, 2021, 35, .	0.5	0
99	Biochemicalâ€structuralâ€phenotypic correlation of adenylosuccinate lyase deficiency based on analysis of mutant enzyme complexes. FASEB Journal, 2010, 24, 889.7.	0.5	0
100	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.	1.2	0