Hana Hartmannova

List of Publications by Year
in descending order

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37	1,873	331670 21	330143 37
papers	citations	h-index	g-index

38	38	38	3838
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A mutation in the SAA1 promoter causes hereditary amyloid A amyloidosis. Kidney International, 2022, 101, 349-359.	5.2	10
2	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	12.8	21
3	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
4	Outcomes of patient self-referral for the diagnosis of several rare inherited kidney diseases. Genetics in Medicine, 2020, 22, 142-149.	2.4	11
5	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
6	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
7	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
8	Rare copy number variation in extremely impulsively violent males. Genes, Brain and Behavior, 2019, 18, e12536.	2.2	9
9	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
10	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
11	Clinical manifestations and molecular aspects of phosphoribosylpyrophosphate synthetase superactivity in females. Rheumatology, 2018, 57, 1180-1185.	1.9	12
12	Multiplex PCR and NGS-based identification of mRNA splicing variants: Analysis of BRCA1 splicing pattern as a model. Gene, 2017, 637, 41-49.	2.2	43
13	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
14	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.	2.0	38
15	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
16	Rare variants in known and novel candidate genes predisposing to statin-associated myopathy. Pharmacogenomics, 2016, 17, 1405-1414.	1.3	17
17	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
18	Mutations in PNPLA6 are linked to photoreceptor degeneration and various forms of childhood blindness. Nature Communications, 2015, 6, 5614.	12.8	77

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19	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
20	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
21	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
22	Mutations in ANTXR1 Cause GAPO Syndrome. American Journal of Human Genetics, 2013, 92, 792-799.	6.2	73
23	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
24	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
25	Expression and processing of the TMEM70 protein. Biochimica Et Biophysica Acta - Bioenergetics, 2011, 1807, 144-149.	1.0	26
26	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
27	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
28	Detection of viral infections by an oligonucleotide microarray. Journal of Virological Methods, 2010, 165, 64-70.	2.1	12
29	Assembly of osteoblastic cell micro-arrays on diamond guided by protein pre-adsorption. Diamond and Related Materials, 2010, 19, 153-157.	3.9	18
30	TMEM70 mutations cause isolated ATP synthase deficiency and neonatal mitochondrial encephalocardiomyopathy. Nature Genetics, 2008, 40, 1288-1290.	21.4	183
31	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
32	The RF plasma surface chemical modification of nanodiamond films grown on glass and silicon at low temperature. Diamond and Related Materials, 2007, 16, 671-674.	3.9	21
33	Rotor-type hyperbilirubinaemia has no defect in the canalicular bilirubin export pump. Liver International, 2007, 27, 485-491.	3.9	22
34	Mutations in TMEM76* Cause Mucopolysaccharidosis IIIC (Sanfilippo C Syndrome). American Journal of Human Genetics, 2006, 79, 807-819.	6.2	77
35	Preparation of 5-amino-4-imidazole-N-succinocarboxamide ribotide, 5-amino-4-imidazole-N-succinocarboxamide riboside and succinyladenosine, compounds usable in diagnosis and research of adenylosuccinate lyase deficiency. Journal of Inherited Metabolic Disease, 2005, 28, 493-499.	3.6	22
36	Identification and determination of succinyladenosine in human cerebrospinal fluid. Biomedical Applications, 1999, 726, 53-58.	1.7	19

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37	Adenylosuccinase deficiency: Clinical and biochemical findings in 5 Czech patients. Journal of Inherited Metabolic Disease, 1997, 20, 343-344.	3.6	20