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List of Publications by Year
in descending order

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38 papers	1,470 citations	304743 22 h-index	330143 37 g-index
38 all docs	38 docs citations	38 times ranked	2740 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	Phenylbutyrate rescues the transport defect of the Sec61Î± mutations V67G and T185A for renin. Life Science Alliance, 2022, 5, e202101150.	2.8	9
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	Mitochondriopathy Manifesting as Inherited Tubulointerstitial Nephropathy Without Symptomatic Other Organ Involvement. Kidney International Reports, 2021, 6, 2514-2518.	0.8	5
5	Outcomes of patient self-referral for the diagnosis of several rare inherited kidney diseases. Genetics in Medicine, 2020, 22, 142-149.	2.4	11
6	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
7	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
8	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
9	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
10	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
11	Rare copy number variation in extremely impulsively violent males. Genes, Brain and Behavior, 2019, 18, e12536.	2.2	9
12	Autosomal dominant tubulointerstitial kidney disease-UMOD is the most frequent non polycystic genetic kidney disease. BMC Nephrology, 2018, 19, 301.	1.8	39
13	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
14	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
15	Clinical manifestations and molecular aspects of phosphoribosylpyrophosphate synthetase superactivity in females. Rheumatology, 2018, 57, 1180-1185.	1.9	12
16	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
17	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
18	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

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19	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
20	Rare variants in known and novel candidate genes predisposing to statin-associated myopathy. Pharmacogenomics, 2016, 17, 1405-1414.	1.3	17
21	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
22	Mutations in PNPLA6 are linked to photoreceptor degeneration and various forms of childhood blindness. Nature Communications, 2015, 6, 5614.	12.8	77
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, 550-555.	1.3	25
24	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
25	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
26	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
27	Mutations in ANTXR1 Cause GAPO Syndrome. American Journal of Human Genetics, 2013, 92, 792-799.	6.2	73
28	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
29	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
30	Genetic and clinical features of patients with Gaucher disease in Hungary. Blood Cells, Molecules, and Diseases, 2007, 39, 119-123.	1.4	18
31	Alterations of uromodulin biology: a common denominator of the genetically heterogeneous FJHN/MCKD syndrome. Kidney International, 2006, 70, 1155-1169.	5.2	111
32	Mapping of a new candidate locus for uromodulin-associated kidney disease (UAKD) to chromosome 1q41. Kidney International, 2005, 68, 1472-1482.	5.2	28
33	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
34	Transient expression of wild-type and mutant glucocerebrosidases in hybrid vaccinia expression system. European Journal of Human Genetics, 2003, 11, 369-374.	2.8	9
35	Analysis of the Î²-Glucocerebrosidase Gene in Czech and Slovak Gaucher Patients: Mutation Profile and Description of Six Novel Mutant Alleles. Blood Cells, Molecules, and Diseases, 1999, 25, 287-298.	1.4	37
36	Interaction of a bZip Oligopeptide Model With Oligodeoxyribonucleotides Modelling DNA Binding Sites. The Effect of Flanking Sequences. Journal of Biomolecular Structure and Dynamics, 1997, 15, 587-596.	3.5	6

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37	Incorrect assignment of N370S mutation status by mismatched PCR/RFLP method in two Gaucher patients. Journal of Inherited Metabolic Disease, 1997, 20, 611-612.	3.6	4
38	A case of type I Gaucher disease with cardiopulmonary amyloidosis and chitotriosidase deficiency. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 1996, 429-429, 305-309.	2.8	14