

Moumita Chaki

List of Publications by Year in descending order

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Version: 2024-02-01

19
papers

2,289
citations

430442

18
h-index

794141

19
g-index

19
all docs

19
docs citations

19
times ranked

3554
citing authors

#	ARTICLE	IF	CITATIONS
1	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. <i>Cell</i> , 2012, 150, 533-548.	13.5	347
2	Candidate exome capture identifies mutation of SDCCAG8 as the cause of a retinal-renal ciliopathy. <i>Nature Genetics</i> , 2010, 42, 840-850.	9.4	295
3	FAN1 mutations cause karyomegalic interstitial nephritis, linking chronic kidney failure to defective DNA damage repair. <i>Nature Genetics</i> , 2012, 44, 910-915.	9.4	205
4	Identification of 99 novel mutations in a worldwide cohort of 1,056 patients with a nephronophthisis-related ciliopathy. <i>Human Genetics</i> , 2013, 132, 865-884.	1.8	199
5	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. <i>American Journal of Human Genetics</i> , 2013, 93, 672-686.	2.6	184
6	ZMYND10 Is Mutated in Primary Ciliary Dyskinesia and Interacts with LRRC6. <i>American Journal of Human Genetics</i> , 2013, 93, 336-345.	2.6	183
7	Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. <i>American Journal of Human Genetics</i> , 2013, 93, 711-720.	2.6	135
8	Mutation analysis of 18 nephronophthisis associated ciliopathy disease genes using a DNA pooling and next generation sequencing strategy. <i>Journal of Medical Genetics</i> , 2011, 48, 105-116.	1.5	123
9	High-throughput mutation analysis in patients with a nephronophthisis-associated ciliopathy applying multiplexed barcoded array-based PCR amplification and next-generation sequencing. <i>Journal of Medical Genetics</i> , 2012, 49, 756-767.	1.5	109
10	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. <i>Journal of Clinical Investigation</i> , 2010, 120, 791-802.	3.9	102
11	Genotype-phenotype correlation in 440 patients with NPHP-related ciliopathies. <i>Kidney International</i> , 2011, 80, 1239-1245.	2.6	99
12	Tyrosinase and ocular diseases: Some novel thoughts on the molecular basis of oculocutaneous albinism type 1. <i>Progress in Retinal and Eye Research</i> , 2007, 26, 323-358.	7.3	80
13	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. <i>Kidney International</i> , 2014, 85, 880-887.	2.6	67
14	Mutations in ANKS6 Cause a Nephronophthisis-Like Phenotype with ESRD. <i>Journal of the American Society of Nephrology: JASN</i> , 2014, 25, 1653-1661.	3.0	37
15	Loss of Glis2/NPHP7 causes kidney epithelial cell senescence and suppresses cyst growth in the Kif3a mouse model of cystic kidney disease. <i>Kidney International</i> , 2016, 89, 1307-1323.	2.6	33
16	Pseudodominant inheritance of nephronophthisis caused by a homozygous NPHP1 deletion. <i>Pediatric Nephrology</i> , 2011, 26, 967-971.	0.9	26
17	Determination of variants in the 3' region of the Tyrosinase gene requires locus specific amplification. <i>Human Mutation</i> , 2005, 26, 53-58.	1.1	24
18	Loss of diacylglycerol kinase epsilon in mice causes endothelial distress and impairs glomerular Cox-2 and PGE2 production. <i>American Journal of Physiology - Renal Physiology</i> , 2016, 310, F895-F908.	1.3	24

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19	Higher prevalence of OCA1 in an ethnic group of eastern India is due to a founder mutation in the tyrosinase gene. <i>Molecular Vision</i> , 2005, 11, 531-4.	1.1	17