Moumita Chaki

List of Publications by Year
in descending order

19 papers	2,289 citations	430874 18 h-index	794594 19 g-index
19 all docs	19 docs citations	19 times ranked	3554 citing authors

#	Article	IF	Citations
1	Loss of diacylglycerol kinase epsilon in mice causes endothelial distress and impairs glomerular Cox-2 and PGE2 production. American Journal of Physiology - Renal Physiology, 2016, 310, F895-F908.	2.7	24
2	Loss of Glis2/NPHP7 causes kidney epithelial cell senescence and suppresses cyst growth in the Kif3a mouse model of cystic kidney disease. Kidney International, 2016, 89, 1307-1323.	5.2	33
3	Mutations in ANKS6 Cause a Nephronophthisis-Like Phenotype with ESRD. Journal of the American Society of Nephrology: JASN, 2014, 25, 1653-1661.	6.1	37
4	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	5.2	67
5	Identification of 99 novel mutations in a worldwide cohort of 1,056 patients with a nephronophthisis-related ciliopathy. Human Genetics, 2013, 132, 865-884.	3.8	199
6	ZMYND10 Is Mutated in Primary Ciliary Dyskinesia and Interacts with LRRC6. American Journal of Human Genetics, 2013, 93, 336-345.	6.2	183
7	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 93, 672-686.	6.2	184
8	Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. American Journal of Human Genetics, 2013, 93, 711-720.	6.2	135
9	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. Cell, 2012, 150, 533-548.	28.9	347
10	High-throughput mutation analysis in patients with a nephronophthisis-associated ciliopathy applying multiplexed barcoded array-based PCR amplification and next-generation sequencing. Journal of Medical Genetics, 2012, 49, 756-767.	3.2	109
11	FAN1 mutations cause karyomegalic interstitial nephritis, linking chronic kidney failure to defective DNA damage repair. Nature Genetics, 2012, 44, 910-915.	21.4	205
12	Genotypeâ€“phenotype correlation in 440 patients with NPHP-related ciliopathies. Kidney International, 2011, 80, 1239-1245.	5.2	99
13	Mutation analysis of 18 nephronophthisis associated ciliopathy disease genes using a DNA pooling and next generation sequencing strategy. Journal of Medical Genetics, 2011, 48, 105-116.	3.2	123
14	Pseudodominant inheritance of nephronophthisis caused by a homozygous NPHP1 deletion. Pediatric Nephrology, 2011, 26, 967-971.	1.7	26
15	Candidate exome capture identifies mutation of SDCCAG8 as the cause of a retinal-renal ciliopathy. Nature Genetics, 2010, 42, 840-850.	21.4	295
16	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. Journal of Clinical Investigation, 2010, 120, 791-802.	8.2	102
17	Tyrosinase and ocular diseases: Some novel thoughts on the molecular basis of oculocutaneous albinism type 1. Progress in Retinal and Eye Research, 2007, 26, 323-358.	15.5	80
18	Determination of variants in the 3â€²-region of the Tyrosinase gene requires locus specific amplification. Human Mutation, 2005, 26, 53-58.	2.5	24

#	Article	IF	Citations
19	Higher prevalence of OCA1 in an ethnic group of eastern India is due to a founder mutation in the tyrosinase gene. Molecular Vision, 2005, 11, 531-4.	1.1	17