Katarzyna Szymanska

List of Publications by Year
in descending order

28	2,989	279798 23	477307 29
papers	citations	h-index	g-index

36	36	36	4531
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	21.4	326
2	Loss-of-function mutations in MICU1 cause a brain and muscle disorder linked to primary alterations in mitochondrial calcium signaling. Nature Genetics, 2014, 46, 188-193.	21.4	311
3	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	21.4	261
4	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	10.3	215
5	TMEM237 Is Mutated in Individuals with a Joubert Syndrome Related Disorder and Expands the Role of the TMEM Family at the Ciliary Transition Zone. American Journal of Human Genetics, 2011, 89, 713-730.	6.2	178
6	Disrupted alternative splicing for genes implicated in splicing and ciliogenesis causes PRPF31 retinitis pigmentosa. Nature Communications, 2018, 9, 4234.	12.8	158
7	TCTN3 Mutations Cause Mohr-Majewski Syndrome. American Journal of Human Genetics, 2012, 91, 372-378.	6.2	123
8	Nesprin-2 interacts with meckelin and mediates ciliogenesis via remodelling of the actin cytoskeleton. Journal of Cell Science, 2009, 122, 2716-2726.	2.0	119
9	Characterizing the morbid genome of ciliopathies. Genome Biology, 2016, 17, 242.	8.8	118
10	TMEM107 recruits ciliopathy proteins to subdomains of the ciliary transition zone and causes JoubertÂsyndrome. Nature Cell Biology, 2016, 18, 122-131.	10.3	118
11	The transition zone: an essential functional compartment of cilia. Cilia, 2012, 1, 10.	1.8	107
12	Meckelâ€“Gruber Syndrome: An Update on Diagnosis, Clinical Management, and Research Advances. Frontiers in Pediatrics, 2017, 5, 244.	1.9	107
13	CiliaCarta: An integrated and validated compendium of ciliary genes. PLoS ONE, 2019, 14, e0216705.	2.5	104
14	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. Nature Medicine, 2012, 18, 1423-1428.	30.7	103
15	A meckelinâ€“filamin A interaction mediates ciliogenesis. Human Molecular Genetics, 2012, 21, 1272-1286.	2.9	96
16	Variable expressivity of ciliopathy neurological phenotypes that encompass Meckelâ€“Gruber syndrome and Joubert syndrome is caused by complex de-regulated ciliogenesis, Shh and Wnt signalling defects. Human Molecular Genetics, 2013, 22, 1358-1372.	2.9	94
17	Mutations in MEGF10, a regulator of satellite cell myogenesis, cause early onset myopathy, areflexia, respiratory distress and dysphagia (EMARDD). Nature Genetics, 2011, 43, 1189-1192.	21.4	84
18	Mutations in CSPP1, Encoding a Core Centrosomal Protein, Cause a Range of Ciliopathy Phenotypes in Humans. American Journal of Human Genetics, 2014, 94, 73-79.	6.2	77

#	Article	IF	Citations
19	The kinetochore protein,CENPF, is mutated in human ciliopathy and microcephaly phenotypes. Journal of Medical Genetics, 2015, 52, 147-156.	3.2	75
20	DNAAF1 links heart laterality with the AAA+ ATPase RUVBL1 and ciliary intraflagellar transport. Human Molecular Genetics, 2018, 27, 529-545.	2.9	45
21	Founder mutations and genotype-phenotype correlations in Meckel-Gruber syndrome and associated ciliopathies. Cilia, 2012, 1, 18.	1.8	42
22	MKS3-Related Ciliopathy with Features of Autosomal Recessive Polycystic Kidney Disease, Nephronophthisis, and Joubert Syndrome. Journal of Pediatrics, 2009, 155, 386-392.e1.	1.8	35
23	Unraveling the genetics of Joubert and Meckel-Gruber syndromes. Journal of Pediatric Genetics, 2015, 03, 065-078.	0.7	35
24	Human Homolog of Drosophila Ariadne (HHARI) is a marker of cellular proliferation associated with nuclear bodies. Experimental Cell Research, 2013, 319, 161-172.	2.6	22
25	Screen-based identification and validation of four novel ion channels as regulators of renal ciliogenesis. Journal of Cell Science, 2015, 128, 4550-9.	2.0	15
26	Shadow autozygosity mapping by linkage exclusion (SAMPLE): a simple strategy to identify the genetic basis of lethal autosomal recessive disorders. Human Mutation, 2009, 30, 1642-1649.	2.5	5
27	Regulation of canonical Wnt signalling by the ciliopathy protein MKS1 and the E2 ubiquitin-conjugating enzyme UBE2E1. ELife, 2022, 11, .	6.0	4
28	Missense mutation of MAL causes a rare leukodystrophy similar to Pelizaeus-Merzbacher disease. European Journal of Human Genetics, 2022, 30, 860-864.	2.8	4