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	Regulation of canonical Wnt signalling by the ciliopathy protein MKS1 and the E2 ubiquitin-conjugating enzyme UBE2E1. ELife, 2022, 11, .	6.0	4
2	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
3	CiliaCarta: An integrated and validated compendium of ciliary genes. PLoS ONE, 2019, 14, e0216705.	2.5	104
4	DNAAF1 links heart laterality with the AAA+ ATPase RUVBL1 and ciliary intraflagellar transport. Human Molecular Genetics, 2018, 27, 529-545.	2.9	45
5	Disrupted alternative splicing for genes implicated in splicing and ciliogenesis causes PRPF31 retinitis pigmentosa. Nature Communications, 2018, 9, 4234.	12.8	158
6	Meckelâ€“Gruber Syndrome: An Update on Diagnosis, Clinical Management, and Research Advances. Frontiers in Pediatrics, 2017, 5, 244.	1.9	107
7	Characterizing the morbid genome of ciliopathies. Genome Biology, 2016, 17, 242.	8.8	118
8	TMEM107 recruits ciliopathy proteins to subdomains of the ciliary transition zone and causes JoubertÂsyndrome. Nature Cell Biology, 2016, 18, 122-131.	10.3	118
9	Unraveling the genetics of Joubert and Meckel-Gruber syndromes. Journal of Pediatric Genetics, 2015, 03, 065-078.	0.7	35
10	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	10.3	215
11	The kinetochore protein,CENPF, is mutated in human ciliopathy and microcephaly phenotypes. Journal of Medical Genetics, 2015, 52, 147-156.	3.2	75
12	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
13	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
14	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
15	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
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	A meckelinâ€“filamin A interaction mediates ciliogenesis. Human Molecular Genetics, 2012, 21, 1272-1286.	2.9	96
18	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. Nature Medicine, 2012, 18, 1423-1428.	30.7	103

#	Article	IF	Citations
19	The transition zone: an essential functional compartment of cilia. Cilia, 2012, 1, 10.	1.8	107
20	TCTN3 Mutations Cause Mohr-Majewski Syndrome. American Journal of Human Genetics, 2012, 91, 372-378.	6.2	123
21	Founder mutations and genotype-phenotype correlations in Meckel-Gruber syndrome and associated ciliopathies. Cilia, 2012, 1, 18.	1.8	42
22	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
23	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	21.4	326
24	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
25	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	21.4	261
26	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
27	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
28	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