Sophie Thomas

List of Publications by Year
in descending order

47	3,286	218677	233421
papers	citations	26 h-index	45 g-index

49	49	49	5813
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	2D and 3D Human Induced Pluripotent Stem Cell-Based Models to Dissect Primary Cilium Involvement during Neocortical Development. Journal of Visualized Experiments, 2022, , .	0.3	0
2	The first two <scp>nonâ€Finnish HYLS1</scp> variants: Expanding the phenotypic spectrum of hydrolethalus syndrome. Clinical Genetics, 2021, 100, 462-467.	2.0	0
3	TALPID3/KIAA0586 Regulates Multiple Aspects of Neuromuscular Patterning During Gastrointestinal Development in Animal Models and Human. Frontiers in Molecular Neuroscience, 2021, 14, 757646.	2.9	3
4	Fetal megacystisâ€microcolon: Genetic mutational spectrum and identification of <scp>PDCL3</scp> as a novel candidate gene. Clinical Genetics, 2020, 98, 261-273.	2.0	18
5	Bi-allelic Variations of SMO in Humans Cause a Broad Spectrum of Developmental Anomalies Due to Abnormal Hedgehog Signaling. American Journal of Human Genetics, 2020, 106, 779-792.	6.2	25
6	Cilia in hereditary cerebral anomalies. Biology of the Cell, 2019, 111, 217-231.	2.0	32
7	Altered GLI3 and FGF8 signaling underlies acrocallosal syndrome phenotypes inKif7depleted mice. Human Molecular Genetics, 2019, 28, 877-887.	2.9	15
8	Novel de novo ZBTB20 mutations in three cases with Primrose syndrome and constant corpus callosum anomalies. American Journal of Medical Genetics, Part A, 2018, 176, 1091-1098.	1.2	16
9	A neuropathological study of novel RTTN gene mutations causing a familial microcephaly with simplified gyral pattern. Birth Defects Research, 2018, 110, 598-602.	1.5	7
10	Whole exome sequencing diagnoses the first fetal case of <scp>B</scp>ainbridgeâ€<scp>R</scp>opers syndrome presenting as pontocerebellar hypoplasia type 1. Birth Defects Research, 2018, 110, 538-542.	1.5	10
11	Loss of function IFT27 variants associated with an unclassified lethal fetal ciliopathy with renal agenesis. American Journal of Medical Genetics, Part A, 2018, 176, 1610-1613.	1.2	16
12	Basal exon skipping and nonsense-associated altered splicing allows bypassing complete CEP290 loss-of-function in individuals with unusually mild retinal disease. Human Molecular Genetics, 2018, 27, 2689-2702.	2.9	31
13	Prenatal and postnatal presentations of corpus callosum agenesis with polymicrogyria caused by EGP5 mutation. American Journal of Medical Genetics, Part A, 2017, 173, 706-711.	1.2	12
14	WDR81 mutations cause extreme microcephaly and impair mitotic progression in human fibroblasts and Drosophila neural stem cells. Brain, 2017, 140, 2597-2609.	7.6	28
15	Heterogeneity of neuroblastoma cell identity defined by transcriptional circuitries. Nature Genetics, 2017, 49, 1408-1413.	21.4	331
16	Neuropathological Hallmarks of Brain Malformations in Extreme Phenotypes Related to DYNC1H1 Mutations. Journal of Neuropathology and Experimental Neurology, 2017, 76, 195-205.	1.7	15
17	Clinical, genetic and neuropathological findings in a series of 138 fetuses with a corpus callosum malformation. Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 36-46.	1.6	37
18	Novel NEK8 Mutations Cause Severe Syndromic Renal Cystic Dysplasia through YAP Dysregulation. PLoS Genetics, 2016, 12, e1005894.	3.5	77

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19	Mutations in KIAA0586 Cause Lethal Ciliopathies Ranging from a Hydrolethalus Phenotype to Short-Rib Polydactyly Syndrome. American Journal of Human Genetics, 2015, 97, 311-318.	6.2	82
20	TMEM231, mutated in orofaciodigital and Meckel syndromes, organizes the ciliary transition zone. Journal of Cell Biology, 2015, 209, 129-142.	5.2	95
21	IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. Journal of Medical Genetics, 2015, 52, 657-665.	3.2	32
22	Identification of a novel ARL13B variant in a Joubert syndrome-affected patient with retinal impairment and obesity. European Journal of Human Genetics, 2015, 23, 621-627.	2.8	48
23	A HomozygousPDE6DMutation in Joubert Syndrome Impairs Targeting of Farnesylated INPP5E Protein to the Primary Cilium. Human Mutation, 2014, 35, 137-146.	2.5	113
24	A Homozygous Missense Mutation in the Ciliary Gene TTC21B Causes Familial FSGS. Journal of the American Society of Nephrology: JASN, 2014, 25, 2435-2443.	6.1	86
25	Developmental molecular and functional cerebellar alterations induced by PCP4/PEP19 overexpression: Implications for Down syndrome. Neurobiology of Disease, 2014, 63, 92-106.	4.4	17
26	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	2.8	64
27	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	12.6	84
28	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. Nature Medicine, 2012, 18, 1423-1428.	30.7	103
29	TCTN3 Mutations Cause Mohr-Majewski Syndrome. American Journal of Human Genetics, 2012, 91, 372-378.	6.2	123
30	NovelKIF7mutations extend the phenotypic spectrum of acrocallosal syndrome. Journal of Medical Genetics, 2012, 49, 713-720.	3.2	28
31	Transcriptome profiling of genes involved in neural tube closure during human embryonic development using long serial analysis of gene expression (longâ€SAGE). Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 683-692.	1.6	18
32	KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes. Nature Genetics, 2011, 43, 601-606.	21.4	203
33	PCP4 (PEP19) overexpression induces premature neuronal differentiation associated with Ca²⁺/CalmodulinâEDependent kinase IIâ€ activation in mouse models of down syndrome. Journal of Comparative Neurology, 2011, 519, 2779-2802.	1.6	39
34	Disruption of a long distance regulatory region upstream of SOX9 in isolated disorders of sex development. Journal of Medical Genetics, 2011, 48, 825-830.	3.2	162
35	Distinct Effects of Allelic NFIX Mutations on Nonsense-Mediated mRNA Decay Engender Either a Sotos-like or a Marshall-Smith Syndrome. American Journal of Human Genetics, 2010, 87, 189-198.	6.2	131
36	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77

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37	High-throughput sequencing of a 4.1â€‰Mb linkage interval reveals FLVCR2 deletions and mutations in lethal cerebral vasculopathy. Human Mutation, 2010, 31, 1134-1141.	2.5	27
38	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	21.4	261
39	BBS10 mutations are common in 'Meckel'-type cystic kidneys. Journal of Medical Genetics, 2010, 47, 848-852.	3.2	25
40	Epistasis between RET and BBS mutations modulates enteric innervation and causes syndromic Hirschsprung disease. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 13921-13926.	7.1	51
41	CC2D2Amutations in Meckel and Joubert syndromes indicate a genotype-phenotype correlation. Human Mutation, 2009, 30, 1574-1582.	2.5	80
42	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. Nature Genetics, 2009, 41, 359-364.	21.4	364
43	Human neural crest cells display molecular and phenotypic hallmarks of stem cells. Human Molecular Genetics, 2008, 17, 3411-3425.	2.9	87
44	Methylation-associated PHOX2B gene silencing is a rare event in human neuroblastoma. European Journal of Cancer, 2007, 43, 2366-2372.	2.8	20
45	Matthew-Wood Syndrome Is Caused by Truncating Mutations in the Retinol-Binding Protein Receptor Gene STRA6. American Journal of Human Genetics, 2007, 80, 1179-1187.	6.2	174
46	Cytogenetic and histological features of a human embryo with homogeneous chromosome 8 trisomy. Prenatal Diagnosis, 2006, 26, 1201-1205.	2.3	5
47	PCP4 is highly expressed in ectoderm and particularly in neuroectoderm derivatives during mouse embryogenesis. Gene Expression Patterns, 2003, 3, 93-97.	0.8	14