

# Sophie Thomas

## List of Publications by Year in descending order

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47  
papers

3,286  
citations

218677

26  
h-index

233421

45  
g-index

49  
all docs

49  
docs citations

49  
times ranked

5813  
citing authors

#	ARTICLE	IF	CITATIONS
1	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. Nature Genetics, 2009, 41, 359-364.	21.4	364
2	Heterogeneity of neuroblastoma cell identity defined by transcriptional circuitries. Nature Genetics, 2017, 49, 1408-1413.	21.4	331
3	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	21.4	261
4	KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes. Nature Genetics, 2011, 43, 601-606.	21.4	203
5	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
6	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
7	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
8	TCTN3 Mutations Cause Mohr-Majewski Syndrome. American Journal of Human Genetics, 2012, 91, 372-378.	6.2	123
9	A Homozygous <i>PDE6D</i> Mutation in Joubert Syndrome Impairs Targeting of Farnesylated INPP5E Protein to the Primary Cilium. Human Mutation, 2014, 35, 137-146.	2.5	113
10	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. Nature Medicine, 2012, 18, 1423-1428.	30.7	103
11	TMEM231, mutated in orofacioidigital and Meckel syndromes, organizes the ciliary transition zone. Journal of Cell Biology, 2015, 209, 129-142.	5.2	95
12	Human neural crest cells display molecular and phenotypic hallmarks of stem cells. Human Molecular Genetics, 2008, 17, 3411-3425.	2.9	87
13	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
14	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	12.6	84
15	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
16	<i>CC2D2A</i> mutations in Meckel and Joubert syndromes indicate a genotype-phenotype correlation. Human Mutation, 2009, 30, 1574-1582.	2.5	80
17	Novel <i>TMEM67</i> mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77
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	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. <i>European Journal of Human Genetics</i> , 2013, 21, 1074-1078.	2.8	64
20	Epistasis between <i>RET</i> and <i>BBS</i> mutations modulates enteric innervation and causes syndromic Hirschsprung disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 13921-13926.	7.1	51
21	Identification of a novel <i>ARL13B</i> variant in a Joubert syndrome-affected patient with retinal impairment and obesity. <i>European Journal of Human Genetics</i> , 2015, 23, 621-627.	2.8	48
22	PCP4 (PEP19) overexpression induces premature neuronal differentiation associated with Ca <sup>2+</sup> /Calmodulin-Dependent kinase II' activation in mouse models of down syndrome. <i>Journal of Comparative Neurology</i> , 2011, 519, 2779-2802.	1.6	39
23	Clinical, genetic and neuropathological findings in a series of 138 fetuses with a corpus callosum malformation. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2016, 106, 36-46.	1.6	37
24	<i>IFT81</i> , encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. <i>Journal of Medical Genetics</i> , 2015, 52, 657-665.	3.2	32
25	Cilia in hereditary cerebral anomalies. <i>Biology of the Cell</i> , 2019, 111, 217-231.	2.0	32
26	Basal exon skipping and nonsense-associated altered splicing allows bypassing complete CEP290 loss-of-function in individuals with unusually mild retinal disease. <i>Human Molecular Genetics</i> , 2018, 27, 2689-2702.	2.9	31
27	Novel <i>KIF7</i> mutations extend the phenotypic spectrum of acrocallosal syndrome. <i>Journal of Medical Genetics</i> , 2012, 49, 713-720.	3.2	28
28	<i>WDR81</i> mutations cause extreme microcephaly and impair mitotic progression in human fibroblasts and <i>Drosophila</i> neural stem cells. <i>Brain</i> , 2017, 140, 2597-2609.	7.6	28
29	High-throughput sequencing of a 4.1 Mb linkage interval reveals <i>FLVCR2</i> deletions and mutations in lethal cerebral vasculopathy. <i>Human Mutation</i> , 2010, 31, 1134-1141.	2.5	27
30	<i>BBS10</i> mutations are common in 'Meckel'-type cystic kidneys. <i>Journal of Medical Genetics</i> , 2010, 47, 848-852.	3.2	25
31	Bi-allelic Variations of <i>SMO</i> in Humans Cause a Broad Spectrum of Developmental Anomalies Due to Abnormal Hedgehog Signaling. <i>American Journal of Human Genetics</i> , 2020, 106, 779-792.	6.2	25
32	Methylation-associated <i>PHOX2B</i> gene silencing is a rare event in human neuroblastoma. <i>European Journal of Cancer</i> , 2007, 43, 2366-2372.	2.8	20
33	Transcriptome profiling of genes involved in neural tube closure during human embryonic development using long serial analysis of gene expression (longSAGE). <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2012, 94, 683-692.	1.6	18
34	Fetal megacystis-microcolon: Genetic mutational spectrum and identification of <i>PDCL3</i> as a novel candidate gene. <i>Clinical Genetics</i> , 2020, 98, 261-273.	2.0	18
35	Developmental molecular and functional cerebellar alterations induced by PCP4/PEP19 overexpression: Implications for Down syndrome. <i>Neurobiology of Disease</i> , 2014, 63, 92-106.	4.4	17
36	Novel de novo <i>ZBTB20</i> mutations in three cases with Primrose syndrome and constant corpus callosum anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1091-1098.	1.2	16

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37	Loss of function IFT27 variants associated with an unclassified lethal fetal ciliopathy with renal agenesis. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1610-1613.	1.2	16
38	Neuropathological Hallmarks of Brain Malformations in Extreme Phenotypes Related to DYNC1H1 Mutations. <i>Journal of Neuropathology and Experimental Neurology</i> , 2017, 76, 195-205.	1.7	15
39	Altered GLI3 and FGF8 signaling underlies acrocallosal syndrome phenotypes in <i>Kif7</i> depleted mice. <i>Human Molecular Genetics</i> , 2019, 28, 877-887.	2.9	15
40	PCP4 is highly expressed in ectoderm and particularly in neuroectoderm derivatives during mouse embryogenesis. <i>Gene Expression Patterns</i> , 2003, 3, 93-97.	0.8	14
41	Prenatal and postnatal presentations of corpus callosum agenesis with polymicrogyria caused by <i>ECP5</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 706-711.	1.2	12
42	Whole exome sequencing diagnoses the first fetal case of <i>Bainbridge-Ropers</i> syndrome presenting as pontocerebellar hypoplasia type 1. <i>Birth Defects Research</i> , 2018, 110, 538-542.	1.5	10
43	A neuropathological study of novel <i>RTTN</i> gene mutations causing a familial microcephaly with simplified gyral pattern. <i>Birth Defects Research</i> , 2018, 110, 598-602.	1.5	7
44	Cytogenetic and histological features of a human embryo with homogeneous chromosome 8 trisomy. <i>Prenatal Diagnosis</i> , 2006, 26, 1201-1205.	2.3	5
45	TALPID3/KIAA0586 Regulates Multiple Aspects of Neuromuscular Patterning During Gastrointestinal Development in Animal Models and Human. <i>Frontiers in Molecular Neuroscience</i> , 2021, 14, 757646.	2.9	3
46	The first two non-Finnish <i>HYLS1</i> variants: Expanding the phenotypic spectrum of hydrolethalus syndrome. <i>Clinical Genetics</i> , 2021, 100, 462-467.	2.0	0
47	2D and 3D Human Induced Pluripotent Stem Cell-Based Models to Dissect Primary Cilium Involvement during Neocortical Development. <i>Journal of Visualized Experiments</i> , 2022, , .	0.3	0