Sophie Saunier

List of Publications by Citations

71 papers	5,768 citations	42 h-index	75 g-index
77 ext. papers	6,662 ext. citations	11.8 avg, IF	4.46 L-index

#	Paper	IF	Citations
71	The ciliary gene RPGRIP1L is mutated in cerebello-oculo-renal syndrome (Joubert syndrome type B) and Meckel syndrome. *Nature Genetics*, **2007**, 39, 875-81	36.3	379
70	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. *Nature Genetics*, **2011**, 43, 189-96	36.3	271
69	Exome capture reveals ZNF423 and CEP164 mutations, linking renal ciliopathies to DNA damage response signaling. *Cell*, **2012**, 150, 533-48	56.2	266
68	Candidate exome capture identifies mutation of SDCCAG8 as the cause of a retinal-renal ciliopathy. *Nature Genetics*, **2010**, 42, 840-50	36.3	257
67	Pleiotropic effects of CEP290 (NPHP6) mutations extend to Meckel syndrome. *American Journal of Human Genetics*, **2007**, 81, 170-9	11	221
66	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. *Nature Genetics*, **2010**, 42, 619-25	36.3	210
65	INF2 mutations in Charcot-Marie-Tooth disease with glomerulopathy. *New England Journal of Medicine*, **2011**, 365, 2377-88	59.2	200
64	The ciliary pocket: an endocytic membrane domain at the base of primary and motile cilia. *Journal of Cell Science*, **2010**, 123, 1785-95	5.3	197
63	The Meckel-Gruber syndrome gene, MKS3, is mutated in Joubert syndrome. *American Journal of Human Genetics*, **2007**, 80, 186-94	11	193
62	The gene mutated in juvenile nephronophthisis type 4 encodes a novel protein that interacts with nephrocystin. *Nature Genetics*, **2002**, 32, 300-5	36.3	186
61	Ciliopathies with skeletal anomalies and renal insufficiency due to mutations in the IFT-A gene WDR19. *American Journal of Human Genetics*, **2011**, 89, 634-43	11	180
60	Defects in the IFT-B component IFT172 cause Jeune and Mainzer-Saldino syndromes in humans. *American Journal of Human Genetics*, **2013**, 93, 915-25	11	155
59	ANKS6 is a central component of a nephronophthisis module linking NEK8 to INVS and NPHP3. *Nature Genetics*, **2013**, 45, 951-6	36.3	144
58	Mainzer-Saldino syndrome is a ciliopathy caused by IFT140 mutations. *American Journal of Human Genetics*, **2012**, 90, 864-70	11	134
57	Characterization of the nephrocystin/nephrocystin-4 complex and subcellular localization of nephrocystin-4 to primary cilia and centrosomes. *Human Molecular Genetics*, **2005**, 14, 645-56	5.6	130
56	High NPHP1 and NPHP6 mutation rate in patients with Joubert syndrome and nephronophthisis: potential epistatic effect of NPHP6 and AHI1 mutations in patients with NPHP1 mutations. *Journal of the American Society of Nephrology: JASN*, **2007**, 18, 1566-75	12.7	128
55	Nephronophthisis. *Pediatric Nephrology*, **2009**, 24, 2333-44	3.2	122

54	Mutation analysis of 18 nephronophthisis associated ciliopathy disease genes using a DNA pooling and next generation sequencing strategy. *Journal of Medical Genetics*, **2011**, 48, 105-16	5.8	112
53	Characterization of the NPHP1 locus: mutational mechanism involved in deletions in familial juvenile nephronophthisis. *American Journal of Human Genetics*, **2000**, 66, 778-89	11	103
52	TCTN3 mutations cause Mohr-Majewski syndrome. *American Journal of Human Genetics*, **2012**, 91, 372-8	11	96
51	Septins 2, 7 and 9 and MAP4 colocalize along the axoneme in the primary cilium and control ciliary length. *Journal of Cell Science*, **2013**, 126, 2583-94	5.3	93
50	Combined NGS approaches identify mutations in the intraflagellar transport gene IFT140 in skeletal ciliopathies with early progressive kidney Disease. *Human Mutation*, **2013**, 34, 714-24	4.7	89
49	The oral-facial-digital syndrome gene C2CD3 encodes a positive regulator of centriole elongation. *Nature Genetics*, **2014**, 46, 905-11	36.3	88
48	A homozygous PDE6D mutation in Joubert syndrome impairs targeting of farnesylated INPP5E protein to the primary cilium. *Human Mutation*, **2014**, 35, 137-46	4.7	88
47	Mutations of NPHP2 and NPHP3 in infantile nephronophthisis. *Kidney International*, **2009**, 75, 839-47	9.9	86
46	TMEM107 recruits ciliopathy proteins to subdomains of the ciliary transition zone and causes Joubert syndrome. *Nature Cell Biology*, **2016**, 18, 122-31	23.4	81
45	Integrin Alpha 8 Recessive Mutations Are Responsible for Bilateral Renal Agenesis in Humans. *American Journal of Human Genetics*, **2014**, 94, 799	11	78
44	Nephrocystin-1 and nephrocystin-4 are required for epithelial morphogenesis and associate with PALS1/PATJ and Par6. *Human Molecular Genetics*, **2009**, 18, 4711-23	5.6	76
43	TMEM231, mutated in orofaciodigital and Meckel syndromes, organizes the ciliary transition zone. *Journal of Cell Biology*, **2015**, 209, 129-42	7.3	71
42	Integrin alpha 8 recessive mutations are responsible for bilateral renal agenesis in humans. *American Journal of Human Genetics*, **2014**, 94, 288-94	11	71
41	Nephronophthisis. *Current Opinion in Genetics and Development*, **2005**, 15, 324-31	4.9	69
40	A homozygous missense mutation in the ciliary gene TTC21B causes familial FSGS. *Journal of the American Society of Nephrology: JASN*, **2014**, 25, 2435-43	12.7	68
39	DCDC2 mutations cause a renal-hepatic ciliopathy by disrupting Wnt signaling. *American Journal of Human Genetics*, **2015**, 96, 81-92	11	66
38	CC2D2A mutations in Meckel and Joubert syndromes indicate a genotype-phenotype correlation. *Human Mutation*, **2009**, 30, 1574-82	4.7	66
37	Mutations in TRAF3IP1/IFT54 reveal a new role for IFT proteins in microtubule stabilization. *Nature Communications*, **2015**, 6, 8666	17.4	65

36	Targeted Exome Sequencing Identifies as Involved in Monogenic Congenital Anomalies of the Kidney and Urinary Tract. *Journal of the American Society of Nephrology: JASN*, **2017**, 28, 2901-2914	12.7	59
35	Mutations of CEP83 cause infantile nephronophthisis and intellectual disability. *American Journal of Human Genetics*, **2014**, 94, 905-14	11	59
34	Fifteen years of research on oral-facial-digital syndromes: from 1 to 16 causal genes. *Journal of Medical Genetics*, **2017**, 54, 371-380	5.8	58
33	Mutations in KIAA0586 Cause Lethal Ciliopathies Ranging from a Hydrolethalus Phenotype to Short-Rib Polydactyly Syndrome. *American Journal of Human Genetics*, **2015**, 97, 311-8	11	57
32	Novel NEK8 Mutations Cause Severe Syndromic Renal Cystic Dysplasia through YAP Dysregulation. *PLoS Genetics*, **2016**, 12, e1005894	6	53
31	Dishevelled stabilization by the ciliopathy protein Rpgrip1l is essential for planar cell polarity. *Journal of Cell Biology*, **2012**, 198, 927-40	7.3	48
30	Control of the Wnt pathways by nephrocystin-4 is required for morphogenesis of the zebrafish pronephros. *Human Molecular Genetics*, **2011**, 20, 2611-27	5.6	45
29	Mutations in GREB1L Cause Bilateral Kidney Agenesis in Humans and Mice. *American Journal of Human Genetics*, **2017**, 101, 803-814	11	41
28	A human patient-derived cellular model of Joubert syndrome reveals ciliary defects which can be rescued with targeted therapies. *Human Molecular Genetics*, **2017**, 26, 4657-4667	5.6	40
27	Matrix metalloproteinase 13 (MMP13) and tissue inhibitor of matrix metalloproteinase 1 (TIMP1), regulated by the MAPK pathway, are both necessary for Madin-Darby canine kidney tubulogenesis. *Journal of Biological Chemistry*, **2008**, 283, 4272-82	5.4	40
26	KIF13B establishes a CAV1-enriched microdomain at the ciliary transition zone to promote Sonic hedgehog signalling. *Nature Communications*, **2017**, 8, 14177	17.4	37
25	Inversin/Nephrocystin-2 is required for fibroblast polarity and directional cell migration. *PLoS ONE*, **2013**, 8, e60193	3.7	37
24	Targeting of beta-arrestin2 to the centrosome and primary cilium: role in cell proliferation control. *PLoS ONE*, **2008**, 3, e3728	3.7	35
23	DCDC2 Mutations Cause Neonatal Sclerosing Cholangitis. *Human Mutation*, **2016**, 37, 1025-9	4.7	32
22	Cell type-specific regulation of ciliary transition zone assembly in vertebrates. *EMBO Journal*, **2018**, 37,	13	31
21	IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. *Journal of Medical Genetics*, **2015**, 52, 657-65	5.8	27
20	Targeted exon skipping of a mutation rescues Joubert syndrome phenotypes in vitro and in a murine model. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 12489-12494	11.5	27
19	Mutation of Growth Arrest Specific 8 Reveals a Role in Motile Cilia Function and Human Disease. *PLoS Genetics*, **2016**, 12, e1006220	6	24

18	Nephrocystin-1 forms a complex with polycystin-1 via a polyproline motif/SH3 domain interaction and regulates the apoptotic response in mammals. *PLoS ONE*, **2010**, 5, e12719	3.7	22
17	Loss-of-function mutations in KIF14 cause severe microcephaly and kidney development defects in humans and zebrafish. *Human Molecular Genetics*, **2019**, 28, 778-795	5.6	22
16	Mutations in MAPKBP1 Cause Juvenile or Late-Onset Cilia-Independent Nephronophthisis. *American Journal of Human Genetics*, **2017**, 100, 323-333	11	19
15	Functional characterization of tektin-1 in motile cilia and evidence for TEKT1 as a new candidate gene for motile ciliopathies. *Human Molecular Genetics*, **2018**, 27, 266-282	5.6	14
14	Familial juvenile nephronophthisis. *Journal of Molecular Medicine*, **1998**, 76, 310-6	5.5	14
13	A 11 Mb YAC-based contig spanning the familial juvenile nephronophthisis region (NPH1) located on chromosome 2q. *Genomics*, **1995**, 30, 514-20	4.3	14
12	Mouse genetics reveals Barttin as a genetic modifier of Joubert syndrome. *Proceedings of the National Academy of Sciences of the United States of America*, **2020**, 117, 1113-1118	11.5	13
11	Whole-genome sequencing in patients with ciliopathies uncovers a novel recurrent tandem duplication in IFT140. *Human Mutation*, **2018**, 39, 983-992	4.7	10
10	Human IFT52 mutations uncover a novel role for the protein in microtubule dynamics and centrosome cohesion. *Human Molecular Genetics*, **2019**, 28, 2720-2737	5.6	7
9	Bi-allelic pathogenic variations in DNAJB11 cause Ivemark II syndrome, a renal-hepatic-pancreatic dysplasia. *Kidney International*, **2021**, 99, 405-409	9.9	7
8	Solution NMR structure of the SH3 domain of human nephrocystin and analysis of a mutation-causing juvenile nephronophthisis. *Proteins: Structure, Function and Bioinformatics*, **2005**, 59, 347-55	4.2	6
7	Renal Ciliopathies: Sorting Out Therapeutic Approaches for Nephronophthisis. *Frontiers in Cell and Developmental Biology*, **2021**, 9, 653138	5.7	6
6	QMPSF is sensitive and specific in the detection of NPHP1 heterozygous deletions. *Clinical Chemistry and Laboratory Medicine*, **2017**, 55, 809-816	5.9	5
5	Novel nephronophthisis-associated variants reveal functional importance of MAPKBP1 dimerization for centriolar recruitment. *Kidney International*, **2020**, 98, 958-969	9.9	4
4	Casein kinase 1 and 1 as novel players in polycystic kidney disease and mechanistic targets for (R)-roscovitine and (S)-CR8. *American Journal of Physiology - Renal Physiology*, **2018**, 315, F57-F73	4.3	3
3	Genetic bases and pathogenic mechanisms of nephronophthisis. *Drug Discovery Today Disease Mechanisms*, **2013**, 10, e143-e151		2
2	Cystic kidney diseases associated with mutations in phosphomannomutase 2 promotor: a large spectrum of phenotypes. *Pediatric Nephrology*, **2021**, 36, 2361-2369	3.2	2
1	Agonists of prostaglandin E receptors as potential first in class treatment for nephronophthisis and related ciliopathies.. *Proceedings of the National Academy of Sciences of the United States of America* , **2022**, 119, e2115960119	11.5	1