Gavin Chapman

List of Publications by Year in Descending Order

41 papers	1,848 citations	24 h-index	42 g-index
43 ext. papers	2,182 ext. citations	8.2 avg, IF	4.11 L-index

#	Paper	IF	Citations
41	An image analysis protocol using CellProfiler for automated quantification of post-ischemic cardiac parameters.. *STAR Protocols*, **2022**, 3, 101097	1.4	0
40	Quantitative 3D analysis and visualization of cardiac fibrosis by microcomputed tomography.. *STAR Protocols*, **2022**, 3, 101055	1.4	0
39	New cases that expand the genotypic and phenotypic spectrum of Congenital NAD Deficiency Disorder. *Human Mutation*, **2021**, 42, 862-876	4.7	2
38	KIAA1217: A novel candidate gene associated with isolated and syndromic vertebral malformations. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1664-1672	2.5	3
37	Functional characterization of a novel PBX1 de novo missense variant identified in a patient with syndromic congenital heart disease. *Human Molecular Genetics*, **2020**, 29, 1068-1082	5.6	13
36	Bi-allelic Mutations in NADSYN1 Cause Multiple Organ Defects and Expand the Genotypic Spectrum of Congenital NAD Deficiency Disorders. *American Journal of Human Genetics*, **2020**, 106, 129-136	11	12
35	Functional genomics and gene-environment interaction highlight the complexity of congenital heart disease caused by Notch pathway variants. *Human Molecular Genetics*, **2020**, 29, 566-579	5.6	16
34	Heterozygous loss of WBP11 function causes multiple congenital defects in humans and mice. *Human Molecular Genetics*, **2020**, 29, 3662-3678	5.6	3
33	Gene-environment interaction impacts on heart development and embryo survival. *Development (Cambridge)*, **2019**, 146,	6.6	29
32	VPOT: A Customizable Variant Prioritization Ordering Tool for Annotated Variants. *Genomics, Proteomics and Bioinformatics*, **2019**, 17, 540-545	6.5	2
31	Identification of clinically actionable variants from genome sequencing of families with congenital heart disease. *Genetics in Medicine*, **2019**, 21, 1111-1120	8.1	25
30	A Screening Approach to Identify Clinically Actionable Variants Causing Congenital Heart Disease in Exome Data. *Circulation Genomic and Precision Medicine*, **2018**, 11, e001978	5.2	37
29	The promises and challenges of exome sequencing in familial, non-syndromic congenital heart disease. *International Journal of Cardiology*, **2017**, 230, 155-163	3.2	8
28	De novo, deleterious sequence variants that alter the transcriptional activity of the homeoprotein PBX1 are associated with intellectual disability and pleiotropic developmental defects. *Human Molecular Genetics*, **2017**, 26, 4849-4860	5.6	26
27	NAD Deficiency, Congenital Malformations, and Niacin Supplementation. *New England Journal of Medicine*, **2017**, 377, 544-552	59.2	114
26	Gestational stress induces the unfolded protein response, resulting in heart defects. *Development (Cambridge)*, **2016**, 143, 2561-72	6.6	31
25	Notch1 endocytosis is induced by ligand and is required for signal transduction. *Biochimica Et Biophysica Acta - Molecular Cell Research*, **2016**, 1863, 166-77	4.9	12

24	NKX2-5 mutations causative for congenital heart disease retain functionality and are directed to hundreds of targets. *ELife*, **2015**, 4,	8.9	34
23	Cited2 is required in trophoblasts for correct placental capillary patterning. *Developmental Biology*, **2014**, 392, 62-79	3.1	31
22	Differential, dominant activation and inhibition of Notch signalling and APP cleavage by truncations of PSEN1 in human disease. *Human Molecular Genetics*, **2014**, 23, 602-17	5.6	40
21	Notch4 reveals a novel mechanism regulating Notch signal transduction. *Biochimica Et Biophysica Acta - Molecular Cell Research*, **2014**, 1843, 1272-84	4.9	31
20	Gene-environment interaction demonstrates the vulnerability of the embryonic heart. *Developmental Biology*, **2014**, 391, 99-110	3.1	12
19	A mechanism for gene-environment interaction in the etiology of congenital scoliosis. *Cell*, **2012**, 149, 295-306	56.2	145
18	Cooperation between somatic Ikaros and Notch1 mutations at the inception of T-ALL. *Leukemia Research*, **2011**, 35, 1512-9	2.7	2
17	The mouse notches up another success: understanding the causes of human vertebral malformation. *Mammalian Genome*, **2011**, 22, 362-76	3.2	25
16	Notch inhibition by the ligand DELTA-LIKE 3 defines the mechanism of abnormal vertebral segmentation in spondylocostal dysostosis. *Human Molecular Genetics*, **2011**, 20, 905-16	5.6	119
15	Loss of Cited2 causes congenital heart disease by perturbing left-right patterning of the body axis. *Human Molecular Genetics*, **2011**, 20, 1097-110	5.6	42
14	A cell autonomous role for the Notch ligand Delta-like 3 in T-cell development. *Immunology and Cell Biology*, **2011**, 89, 696-705	5	22
13	Role of Delta-like-3 in mammalian somitogenesis and vertebral column formation. *Advances in Experimental Medicine and Biology*, **2008**, 638, 95-112	3.6	3
12	Disruption of the somitic molecular clock causes abnormal vertebral segmentation. *Birth Defects Research Part C: Embryo Today Reviews*, **2007**, 81, 93-110		21
11	Divergent functions and distinct localization of the Notch ligands DLL1 and DLL3 in vivo. *Journal of Cell Biology*, **2007**, 178, 465-76	7.3	114
10	Divergent functions and distinct localization of the Notch ligands DLL1 and DLL3 in vivo. *Journal of Experimental Medicine*, **2007**, 204, i20-i20	16.6	
9	High levels of Notch signaling down-regulate Numb and Numblike. *Journal of Cell Biology*, **2006**, 175, 535-40	7.3	71
8	Mutation of the LUNATIC FRINGE gene in humans causes spondylocostal dysostosis with a severe vertebral phenotype. *American Journal of Human Genetics*, **2006**, 78, 28-37	11	191
7	Recording Notch signaling in real time. *Developmental Neuroscience*, **2006**, 28, 118-27	2.2	36

6	Notch signaling in development and disease. *Seminars in Cancer Biology*, **2004**, 14, 320-8	12.7	182
5	Functional Notch signaling is required for BMP4-induced inhibition of myogenic differentiation. *Development (Cambridge)*, **2003**, 130, 6089-99	6.6	204
4	A CADASIL-mutated Notch 3 receptor exhibits impaired intracellular trafficking and maturation but normal ligand-induced signaling. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 17119-24	11.5	90
3	CRTR-1, a developmentally regulated transcriptional repressor related to the CP2 family of transcription factors. *Journal of Biological Chemistry*, **2001**, 276, 3324-32	5.4	33
2	The mouse homeobox gene, Gbx2: genomic organization and expression in pluripotent cells in vitro and in vivo. *Genomics*, **1997**, 46, 223-33	4.3	41
1	Sequence and evolutionary conservation of the murine Gbx-2 homeobox gene. *FEBS Letters*, **1995**, 364, 289-92	3.8	26