Heather C Etchevers

List of Publications by Citations

68 papers	4,241 citations	30 h-index	65 g-index
83 ext. papers	4,856 ext. citations	7.5 avg, IF	4.82 L-index

#	Paper	IF	Citations
68	Polyalanine expansion and frameshift mutations of the paired-like homeobox gene PHOX2B in congenital central hypoventilation syndrome. *Nature Genetics*, **2003**, 33, 459-61	36.3	665
67	The cephalic neural crest provides pericytes and smooth muscle cells to all blood vessels of the face and forebrain. *Development (Cambridge)*, **2001**, 128, 1059-1068	6.6	432
66	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. *Nature Genetics*, **2009**, 41, 359-64	36.3	304
65	Loss-of-function mutation in the dioxygenase-encoding FTO gene causes severe growth retardation and multiple malformations. *American Journal of Human Genetics*, **2009**, 85, 106-11	11	275
64	Human neural tube defects: developmental biology, epidemiology, and genetics. *Neurotoxicology and Teratology*, **2005**, 27, 515-24	3.9	266
63	Heterogeneity of neuroblastoma cell identity defined by transcriptional circuitries. *Nature Genetics*, **2017**, 49, 1408-1413	36.3	174
62	Phenotypic spectrum of CHARGE syndrome in fetuses with CHD7 truncating mutations correlates with expression during human development. *Journal of Medical Genetics*, **2006**, 43, 211-217	5.8	171
61	PAX8, TITF1, and FOXE1 gene expression patterns during human development: new insights into human thyroid development and thyroid dysgenesis-associated malformations. *Journal of Clinical Endocrinology and Metabolism*, **2005**, 90, 455-62	5.6	166
60	Matthew-Wood syndrome is caused by truncating mutations in the retinol-binding protein receptor gene STRA6. *American Journal of Human Genetics*, **2007**, 80, 1179-87	11	155
59	Homozygous silencing of T-box transcription factor EOMES leads to microcephaly with polymicrogyria and corpus callosum agenesis. *Nature Genetics*, **2007**, 39, 454-6	36.3	152
58	Mutational, functional, and expression studies of the TCF4 gene in Pitt-Hopkins syndrome. *Human Mutation*, **2009**, 30, 669-76	4.7	112
57	Antenatal presentation of Bardet-Biedl syndrome may mimic Meckel syndrome. *American Journal of Human Genetics*, **2005**, 76, 493-504	11	101
56	Human neural crest cells display molecular and phenotypic hallmarks of stem cells. *Human Molecular Genetics*, **2008**, 17, 3411-25	5.6	79
55	Anterior cephalic neural crest is required for forebrain viability. *Development (Cambridge)*, **1999**, 126, 3533-3543	6.6	78
54	Phenotypic spectrum of STRA6 mutations: from Matthew-Wood syndrome to non-lethal anophthalmia. *Human Mutation*, **2009**, 30, E673-81	4.7	76
53	Molecular bases of human neurocristopathies. *Advances in Experimental Medicine and Biology*, **2006**, 589, 213-34	3.6	66
52	OTX2 mutations contribute to the otocephaly-dysgnathia complex. *Journal of Medical Genetics*, **2012**, 49, 373-9	5.8	49

51	The diverse neural crest: from embryology to human pathology. *Development (Cambridge)*, **2019**, 146,	6.6	48
50	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, 13921-6	11.5	47
49	Embryonic expression of the human MID1 gene and its mutations in Opitz syndrome. *Journal of Medical Genetics*, **2004**, 41, 381-6	5.8	47
48	CLMP is required for intestinal development, and loss-of-function mutations cause congenital short-bowel syndrome. *Gastroenterology*, **2012**, 142, 453-462.e3	13.3	43
47	Analysis of mouse models carrying the I26T and R160C substitutions in the transcriptional repressor HESX1 as models for septo-optic dysplasia and hypopituitarism. *DMM Disease Models and Mechanisms*, **2008**, 1, 241-54	4.1	43
46	Primary culture of chick, mouse or human neural crest cells. *Nature Protocols*, **2011**, 6, 1568-77	18.8	37
45	Expression of Frzb-1 during chick development. *Mechanisms of Development*, **1999**, 89, 179-83	1.7	36
44	ISL1 directly regulates FGF10 transcription during human cardiac outflow formation. *PLoS ONE*, **2012**, 7, e30677	3.7	35
43	Confirmation of RAX gene involvement in human anophthalmia. *Clinical Genetics*, **2008**, 74, 392-5	4	33
42	Comparative transcriptome and network biology analyses demonstrate antiproliferative and hyperapoptotic phenotypes in human keratoconus corneas **2011**, 52, 6181-91		32
41	Morphogenesis of the branchial vascular sector. *Trends in Cardiovascular Medicine*, **2002**, 12, 299-304	6.9	32
40	Germline gain-of-function mutations of ALK disrupt central nervous system development. *Human Mutation*, **2011**, 32, 272-6	4.7	31
39	Gene expression in pharyngeal arch 1 during human embryonic development. *Human Molecular Genetics*, **2005**, 14, 903-12	5.6	30
38	Macrophage-Derived IL1 and TNF Regulate Arginine Metabolism in Neuroblastoma. *Cancer Research*, **2019**, 79, 611-624	10.1	29
37	SNPs in the neural cell adhesion molecule 1 gene (NCAM1) may be associated with human neural tube defects. *Human Genetics*, **2005**, 117, 133-42	6.3	28
36	Targeted resequencing identifies PTCH1 as a major contributor to ocular developmental anomalies and extends the SOX2 regulatory network. *Genome Research*, **2016**, 26, 474-85	9.7	27
35	The hedgehog pathway and ocular developmental anomalies. *Human Genetics*, **2019**, 138, 917-936	6.3	25
34	Practical application of the new classification scheme for congenital melanocytic nevi. *Pediatric Dermatology*, **2015**, 32, 23-7	1.9	22

33	Dissection of the MYCN locus in Feingold syndrome and isolated oesophageal atresia. *European Journal of Human Genetics*, **2011**, 19, 602-6	5.3	21
32	Cardiac outflow morphogenesis depends on effects of retinoic acid signaling on multiple cell lineages. *Developmental Dynamics*, **2016**, 245, 388-401	2.9	21
31	Hiding in plain sight: molecular genetics applied to giant congenital melanocytic nevi. *Journal of Investigative Dermatology*, **2014**, 134, 879-882	4.3	20
30	High-throughput sequencing of a 4.1 Mb linkage interval reveals FLVCR2 deletions and mutations in lethal cerebral vasculopathy. *Human Mutation*, **2010**, 31, 1134-41	4.7	18
29	Methylation-associated PHOX2B gene silencing is a rare event in human neuroblastoma. *European Journal of Cancer*, **2007**, 43, 2366-72	7.5	18
28	A roadmap for the Human Developmental Cell Atlas. *Nature*, **2021**, 597, 196-205	50.4	18
27	Giant congenital melanocytic nevus with vascular malformation and epidermal cysts associated with a somatic activating mutation in BRAF. *Pigment Cell and Melanoma Research*, **2018**, 31, 437-441	4.5	16
26	A subpopulation of smooth muscle cells, derived from melanocyte-competent precursors, prevents patent ductus arteriosus. *PLoS ONE*, **2013**, 8, e53183	3.7	15
25	Refining the clinicopathological pattern of cerebral proliferative glomeruloid vasculopathy (Fowler syndrome): report of 16 fetal cases. *European Journal of Medical Genetics*, **2009**, 52, 386-92	2.6	15
24	Genome-wide DNA methylation analysis identifies MEGF10 as a novel epigenetically repressed candidate tumor suppressor gene in neuroblastoma. *Molecular Carcinogenesis*, **2017**, 56, 1290-1301	5	12
23	Matthew-Wood syndrome: report of two new cases supporting autosomal recessive inheritance and exclusion of FGF10 and FGFR2. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 219-28	2.5	12
22	Ectopic expression of Hoxb1 induces cardiac and craniofacial malformations. *Genesis*, **2018**, 56, e23221	1.9	12
21	Transcriptome profiling of genes involved in neural tube closure during human embryonic development using long serial analysis of gene expression (long-SAGE). *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2012**, 94, 683-92		11
20	The cap 'n' collar family member NF-E2-related factor 3 (Nrf3) is expressed in mesodermal derivatives of the avian embryo. *International Journal of Developmental Biology*, **2005**, 49, 363-7	1.9	11
19	Early expression of hypoxia-inducible factor 1alpha in the chicken embryo. *Gene Expression Patterns* , **2003**, 3, 49-52	1.5	10
18	Epigenetic deregulation of GATA3 in neuroblastoma is associated with increased GATA3 protein expression and with poor outcomes. *Scientific Reports*, **2019**, 9, 18934	4.9	10
17	Neural Crest and Pituitary Development **2001**, 4, 13-29		5
16	Widespread dynamic and pleiotropic expression of the melanocortin-1-receptor (MC1R) system is conserved across chick, mouse and human embryonic development. *Birth Defects Research*, **2018**, 110, 443-455	2.9	4

List of Publications

15	Cytogenetic and histological features of a human embryo with homogeneous chromosome 8 trisomy. *Prenatal Diagnosis*, **2006**, 26, 1201-5	3.2	4
14	Cutaneous Melanocytic Tumors With Concomitant NRASQ61R and IDH1R132C Mutations: A Report of 6 Cases. *American Journal of Surgical Pathology*, **2020**, 44, 1398-1405	6.7	4
13	Development of an international core domain set for medium, large and giant congenital melanocytic naevi as a first step towards a core outcome set for clinical practice and research. *British Journal of Dermatology*, **2021**, 185, 371-379	4	4
12	A severe clinical phenotype of Noonan syndrome with neonatal hypertrophic cardiomyopathy in the second case worldwide with RAF1 S259Y neomutation. *Genetical Research*, **2019**, 101, e6	1.1	3
11	Identification of the IRXB gene cluster as candidate genes in severe dysgenesis of the ocular anterior segment **2010**, 51, 4380-6		2
10	Etiology of Congenital Melanocytic Nevi and Related Conditions **2012**, 73-97		2
9	DNA sequencing and quick clean-up. *Protocol Exchange*,		2
8	Outflow Tract Formation-Embryonic Origins of Conotruncal Congenital Heart Disease. *Journal of Cardiovascular Development and Disease*, **2021**, 8,	4.2	2
7	Pericyte Ontogeny: The Use of Chimeras to Track a Cell Lineage of Diverse Germ Line Origins. *Methods in Molecular Biology*, **2021**, 2235, 61-87	1.4	2
6	Domains and outcomes of the core outcome set of congenital melanocytic naevi for clinical practice and research (the OCOMEN project): part 2. *British Journal of Dermatology*, **2021**, 185, 970-977	4	2
5	Reduced H3K27me3 Expression is Common in Nodular Melanomas of Childhood Associated With Congenital Melanocytic Nevi But Not in Proliferative Nodules. *American Journal of Surgical Pathology*, **2018**, 42, 701-704	6.7	1
4	Sustained experimental activation of FGF8/ERK in the developing chicken spinal cord models early events in ERK-mediated tumorigenesis.. *Neoplasia*, **2021**, 24, 120-132	6.4	1
3	Sustained experimental activation of FGF8/ERK in the developing chicken spinal cord reproducibly models early events in ERK-mediated tumorigenesis		1
2	Melanocortin-1 receptor (MC1R) genotypes do not correlate with size in two cohorts of medium-to-giant congenital melanocytic nevi. *Pigment Cell and Melanoma Research*, **2020**, 33, 685-694	4.5	0
1	Cutaneous Melanomas Arising during Childhood: An Overview of the Main Entities. *Dermatopathology (Basel, Switzerland)*, **2021**, 8, 301-314	1.9	0