

Heather C Etchevers

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Sustained experimental activation of FGF8/ERK in the developing chicken spinal cord models early events in ERK-mediated tumorigenesis.. <i>Neoplasia</i> , 2021 , 24, 120-132	6.4	1
67	Outflow Tract Formation-Embryonic Origins of Conotruncal Congenital Heart Disease. <i>Journal of Cardiovascular Development and Disease</i> , 2021 , 8,	4.2	2
66	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. <i>British Journal of Dermatology</i> , 2021 , 185, 371-379	4	4
65	Pericyte Ontogeny: The Use of Chimeras to Track a Cell Lineage of Diverse Germ Line Origins. <i>Methods in Molecular Biology</i> , 2021 , 2235, 61-87	1.4	2
64	Domains and outcomes of the core outcome set of congenital melanocytic naevi for clinical practice and research (the OCOMEN project): part 2. <i>British Journal of Dermatology</i> , 2021 , 185, 970-977	4	2
63	Cutaneous Melanomas Arising during Childhood: An Overview of the Main Entities. <i>Dermatopathology (Basel, Switzerland)</i> , 2021 , 8, 301-314	1.9	0
62	A roadmap for the Human Developmental Cell Atlas. <i>Nature</i> , 2021 , 597, 196-205	50.4	18
61	Melanocortin-1 receptor (MC1R) genotypes do not correlate with size in two cohorts of medium-to-giant congenital melanocytic nevi. <i>Pigment Cell and Melanoma Research</i> , 2020 , 33, 685-694	4.5	0
60	Cutaneous Melanocytic Tumors With Concomitant NRASQ61R and IDH1R132C Mutations: A Report of 6 Cases. <i>American Journal of Surgical Pathology</i> , 2020 , 44, 1398-1405	6.7	4
59	A severe clinical phenotype of Noonan syndrome with neonatal hypertrophic cardiomyopathy in the second case worldwide with RAF1 S259Y neomutation. <i>Genetical Research</i> , 2019 , 101, e6	1.1	3
58	The diverse neural crest: from embryology to human pathology. <i>Development (Cambridge)</i> , 2019 , 146,	6.6	48
57	The hedgehog pathway and ocular developmental anomalies. <i>Human Genetics</i> , 2019 , 138, 917-936	6.3	25
56	Epigenetic deregulation of GATA3 in neuroblastoma is associated with increased GATA3 protein expression and with poor outcomes. <i>Scientific Reports</i> , 2019 , 9, 18934	4.9	10
55	Macrophage-Derived IL1 β and TNF α Regulate Arginine Metabolism in Neuroblastoma. <i>Cancer Research</i> , 2019 , 79, 611-624	10.1	29
54	Reduced H3K27me3 Expression is Common in Nodular Melanomas of Childhood Associated With Congenital Melanocytic Nevi But Not in Proliferative Nodules. <i>American Journal of Surgical Pathology</i> , 2018 , 42, 701-704	6.7	1
53	Widespread dynamic and pleiotropic expression of the melanocortin-1-receptor (MC1R) system is conserved across chick, mouse and human embryonic development. <i>Birth Defects Research</i> , 2018 , 110, 443-455	2.9	4
52	Giant congenital melanocytic nevus with vascular malformation and epidermal cysts associated with a somatic activating mutation in BRAF. <i>Pigment Cell and Melanoma Research</i> , 2018 , 31, 437-441	4.5	16

51	Ectopic expression of Hoxb1 induces cardiac and craniofacial malformations. <i>Genesis</i> , 2018 , 56, e23221	1.9	12
50	Heterogeneity of neuroblastoma cell identity defined by transcriptional circuitries. <i>Nature Genetics</i> , 2017 , 49, 1408-1413	36.3	174
49	Genome-wide DNA methylation analysis identifies MEGF10 as a novel epigenetically repressed candidate tumor suppressor gene in neuroblastoma. <i>Molecular Carcinogenesis</i> , 2017 , 56, 1290-1301	5	12
48	Targeted resequencing identifies PTCH1 as a major contributor to ocular developmental anomalies and extends the SOX2 regulatory network. <i>Genome Research</i> , 2016 , 26, 474-85	9.7	27
47	Cardiac outflow morphogenesis depends on effects of retinoic acid signaling on multiple cell lineages. <i>Developmental Dynamics</i> , 2016 , 245, 388-401	2.9	21
46	Practical application of the new classification scheme for congenital melanocytic nevi. <i>Pediatric Dermatology</i> , 2015 , 32, 23-7	1.9	22
45	Hiding in plain sight: molecular genetics applied to giant congenital melanocytic nevi. <i>Journal of Investigative Dermatology</i> , 2014 , 134, 879-882	4.3	20
44	A subpopulation of smooth muscle cells, derived from melanocyte-competent precursors, prevents patent ductus arteriosus. <i>PLoS ONE</i> , 2013 , 8, e53183	3.7	15
43	CLMP is required for intestinal development, and loss-of-function mutations cause congenital short-bowel syndrome. <i>Gastroenterology</i> , 2012 , 142, 453-462.e3	13.3	43
42	ISL1 directly regulates FGF10 transcription during human cardiac outflow formation. <i>PLoS ONE</i> , 2012 , 7, e30677	3.7	35
41	Transcriptome profiling of genes involved in neural tube closure during human embryonic development using long serial analysis of gene expression (long-SAGE). <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2012 , 94, 683-92		11
40	OTX2 mutations contribute to the otocephaly-dysgnathia complex. <i>Journal of Medical Genetics</i> , 2012 , 49, 373-9	5.8	49
39	Etiology of Congenital Melanocytic Nevi and Related Conditions 2012 , 73-97		2
38	Primary culture of chick, mouse or human neural crest cells. <i>Nature Protocols</i> , 2011 , 6, 1568-77	18.8	37
37	Comparative transcriptome and network biology analyses demonstrate antiproliferative and hyperapoptotic phenotypes in human keratoconus corneas 2011 , 52, 6181-91		32
36	Dissection of the MYCN locus in Feingold syndrome and isolated oesophageal atresia. <i>European Journal of Human Genetics</i> , 2011 , 19, 602-6	5.3	21
35	Germline gain-of-function mutations of ALK disrupt central nervous system development. <i>Human Mutation</i> , 2011 , 32, 272-6	4.7	31
34	Identification of the IRXB gene cluster as candidate genes in severe dysgenesis of the ocular anterior segment 2010 , 51, 4380-6		2

33	High-throughput sequencing of a 4.1 Mb linkage interval reveals FLVCR2 deletions and mutations in lethal cerebral vasculopathy. <i>Human Mutation</i> , 2010 , 31, 1134-41	4.7	18
32	Epistasis between RET and BBS 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, 13921-6	11.5	47
31	Mutational, functional, and expression studies of the TCF4 gene in Pitt-Hopkins syndrome. <i>Human Mutation</i> , 2009 , 30, 669-76	4.7	112
30	Phenotypic spectrum of STRA6 mutations: from Matthew-Wood syndrome to non-lethal anophthalmia. <i>Human Mutation</i> , 2009 , 30, E673-81	4.7	76
29	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. <i>Nature Genetics</i> , 2009 , 41, 359-64	36.3	304
28	Loss-of-function mutation in the dioxygenase-encoding FTO gene causes severe growth retardation and multiple malformations. <i>American Journal of Human Genetics</i> , 2009 , 85, 106-11	11	275
27	Refining the clinicopathological pattern of cerebral proliferative glomeruloid vasculopathy (Fowler syndrome): report of 16 fetal cases. <i>European Journal of Medical Genetics</i> , 2009 , 52, 386-92	2.6	15
26	Confirmation of RAX gene involvement in human anophthalmia. <i>Clinical Genetics</i> , 2008 , 74, 392-5	4	33
25	Human neural crest cells display molecular and phenotypic hallmarks of stem cells. <i>Human Molecular Genetics</i> , 2008 , 17, 3411-25	5.6	79
24	Analysis of mouse models carrying the I26T and R160C substitutions in the transcriptional repressor HESX1 as models for septo-optic dysplasia and hypopituitarism. <i>DMM Disease Models and Mechanisms</i> , 2008 , 1, 241-54	4.1	43
23	Matthew-Wood syndrome: report of two new cases supporting autosomal recessive inheritance and exclusion of FGF10 and FGFR2. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 219-28	2.5	12
22	Homozygous silencing of T-box transcription factor EOMES leads to microcephaly with polymicrogyria and corpus callosum agenesis. <i>Nature Genetics</i> , 2007 , 39, 454-6	36.3	152
21	Methylation-associated PHOX2B gene silencing is a rare event in human neuroblastoma. <i>European Journal of Cancer</i> , 2007 , 43, 2366-72	7.5	18
20	Matthew-Wood syndrome is caused by truncating mutations in the retinol-binding protein receptor gene STRA6. <i>American Journal of Human Genetics</i> , 2007 , 80, 1179-87	11	155
19	Molecular bases of human neurocristopathies. <i>Advances in Experimental Medicine and Biology</i> , 2006 , 589, 213-34	3.6	66
18	Phenotypic spectrum of CHARGE syndrome in fetuses with CHD7 truncating mutations correlates with expression during human development. <i>Journal of Medical Genetics</i> , 2006 , 43, 211-217	5.8	171
17	Cytogenetic and histological features of a human embryo with homogeneous chromosome 8 trisomy. <i>Prenatal Diagnosis</i> , 2006 , 26, 1201-5	3.2	4
16	Antenatal presentation of Bardet-Biedl syndrome may mimic Meckel syndrome. <i>American Journal of Human Genetics</i> , 2005 , 76, 493-504	11	101

15	Human neural tube defects: developmental biology, epidemiology, and genetics. <i>Neurotoxicology and Teratology</i> , 2005 , 27, 515-24	3.9	266
14	SNPs in the neural cell adhesion molecule 1 gene (NCAM1) may be associated with human neural tube defects. <i>Human Genetics</i> , 2005 , 117, 133-42	6.3	28
13	PAX8, TITF1, and FOXE1 gene expression patterns during human development: new insights into human thyroid development and thyroid dysgenesis-associated malformations. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2005 , 90, 455-62	5.6	166
12	Gene expression in pharyngeal arch 1 during human embryonic development. <i>Human Molecular Genetics</i> , 2005 , 14, 903-12	5.6	30
11	The cap Wcollar family member NF-E2-related factor 3 (Nrf3) is expressed in mesodermal derivatives of the avian embryo. <i>International Journal of Developmental Biology</i> , 2005 , 49, 363-7	1.9	11
10	Embryonic expression of the human MID1 gene and its mutations in Opitz syndrome. <i>Journal of Medical Genetics</i> , 2004 , 41, 381-6	5.8	47
9	Early expression of hypoxia-inducible factor 1alpha in the chicken embryo. <i>Gene Expression Patterns</i> , 2003 , 3, 49-52	1.5	10
8	Polyalanine expansion and frameshift mutations of the paired-like homeobox gene PHOX2B in congenital central hypoventilation syndrome. <i>Nature Genetics</i> , 2003 , 33, 459-61	36.3	665
7	Morphogenesis of the branchial vascular sector. <i>Trends in Cardiovascular Medicine</i> , 2002 , 12, 299-304	6.9	32
6	Neural Crest and Pituitary Development 2001 , 4, 13-29		5
5	The cephalic neural crest provides pericytes and smooth muscle cells to all blood vessels of the face and forebrain. <i>Development (Cambridge)</i> , 2001 , 128, 1059-1068	6.6	432
4	Expression of Frzb-1 during chick development. <i>Mechanisms of Development</i> , 1999 , 89, 179-83	1.7	36
3	Anterior cephalic neural crest is required for forebrain viability. <i>Development (Cambridge)</i> , 1999 , 126, 3533-3543	6.6	78
2	DNA sequencing and quick clean-up. <i>Protocol Exchange</i> ,		2
1	Sustained experimental activation of FGF8/ERK in the developing chicken spinal cord reproducibly models early events in ERK-mediated tumorigenesis		1