Heather C Etchevers

List of Publications by Year
in descending order

69 papers	5,413 citations	136940 32 h-index	106340 65 g-index
84 all docs	84 docs citations	84 times ranked	8599 citing authors

#	Article	IF	Citations
1	Polyalanine expansion and frameshift mutations of the paired-like homeobox gene PHOX2B in congenital central hypoventilation syndrome. Nature Genetics, 2003, 33, 459-461.	21.4	771
2	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.	2.5	546
3	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. Nature Genetics, 2009, 41, 359-364.	21.4	364
4	Loss-of-Function Mutation in the Dioxygenase-Encoding FTO Gene Causes Severe Growth Retardation and Multiple Malformations. American Journal of Human Genetics, 2009, 85, 106-111.	6.2	340
5	Heterogeneity of neuroblastoma cell identity defined by transcriptional circuitries. Nature Genetics, 2017, 49, 1408-1413.	21.4	331
6	Human neural tube defects: Developmental biology, epidemiology, and genetics. Neurotoxicology and Teratology, 2005, 27, 515-524.	2.4	317
7	Phenotypic spectrum of CHARGE syndrome in fetuses with CHD7 truncating mutations correlates with expression during human development. Journal of Medical Genetics, 2005, 43, 211-317.	3.2	199
8	PAX8,TITF1, andFOXE1Gene Expression Patterns during Human Development: New Insights into Human Thyroid Development and Thyroid Dysgenesis-Associated Malformations. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 455-462.	3.6	195
9	Homozygous silencing of T-box transcription factor EOMES leads to microcephaly with polymicrogyria and corpus callosum agenesis. Nature Genetics, 2007, 39, 454-456.	21.4	181
10	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
11	Mutational, functional, and expression studies of theTCF4gene in Pitt-Hopkins syndrome. Human Mutation, 2009, 30, 669-676.	2.5	126
12	Antenatal Presentation of Bardet-Biedl Syndrome May Mimic Meckel Syndrome. American Journal of Human Genetics, 2005, 76, 493-504.	6.2	120
13	A roadmap for the Human Developmental Cell Atlas. Nature, 2021, 597, 196-205.	27.8	114
14	Anterior cephalic neural crest is required for forebrain viability. Development (Cambridge), 1999, 126, 3533-3543.	2.5	107
15	Phenotypic spectrum ofSTRA6mutations: from Matthew-Wood syndrome to non-lethal anophthalmia. Human Mutation, 2009, 30, E673-E681.	2.5	89
16	Human neural crest cells display molecular and phenotypic hallmarks of stem cells. Human Molecular Genetics, 2008, 17, 3411-3425.	2.9	87
17	The diverse neural crest: from embryology to human pathology. Development (Cambridge), 2019, 146, .	2.5	82
18	Molecular Bases of Human Neurocristopathies. , 2006, 589, 213-234.		79

#	Article	IF	Citations
19	Embryonic expression of the human MID1 gene and its mutations in Opitz syndrome. Journal of Medical Genetics, 2004, 41, 381-386.	3.2	58
20	OTX2mutations contribute to the otocephaly-dysgnathia complex. Journal of Medical Genetics, 2012, 49, 373-379.	3.2	58
21	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, 13921-13926.	7.1	51
22	Macrophage-Derived IL1Î² and TNFÎ± Regulate Arginine Metabolism in Neuroblastoma. Cancer Research, 2019, 79, 611-624.	0.9	50
23	CLMP Is Required for Intestinal Development, and Loss-of-Function Mutations Cause Congenital Short-Bowel Syndrome. Gastroenterology, 2012, 142, 453-462.e3.	1.3	49
24	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, 241-254.	2.4	46
25	ISL1 Directly Regulates FGF10 Transcription during Human Cardiac Outflow Formation. PLoS ONE, 2012, 7, e30677.	2.5	46
26	Primary culture of chick, mouse or human neural crest cells. Nature Protocols, 2011, 6, 1568-1577.	12.0	41
27	Morphogenesis of the Branchial Vascular Sector. Trends in Cardiovascular Medicine, 2002, 12, 299-304.	4.9	40
28	Comparative Transcriptome and Network Biology Analyses Demonstrate Antiproliferative and Hyperapoptotic Phenotypes in Human Keratoconus Corneas. , 2011, 52, 6181.		40
29	The hedgehog pathway and ocular developmental anomalies. Human Genetics, 2019, 138, 917-936.	3.8	40
30	Germline gain-of-function mutations of ALK disrupt central nervous system development. Human Mutation, 2011, 32, 272-276.	2.5	38
31	Expression of Frzb-1 during chick development. Mechanisms of Development, 1999, 89, 179-183.	1.7	37
32	Confirmation of RAX gene involvement in human anophthalmia. Clinical Genetics, 2008, 74, 392-395.	2.0	37
33	Targeted resequencing identifies PTCH1 as a major contributor to ocular developmental anomalies and extends the SOX2 regulatory network. Genome Research, 2016, 26, 474-485.	5.5	37
34	SNPs in the neural cell adhesion molecule 1 gene (NCAM1) may be associated with human neural tube defects. Human Genetics, 2005, 117, 133-142.	3.8	35
35	Gene expression in pharyngeal arch 1 during human embryonic development. Human Molecular Genetics, 2005, 14, 903-912.	2.9	35
36	Cardiac outflow morphogenesis depends on effects of retinoic acid signaling on multiple cell lineages. Developmental Dynamics, 2016, 245, 388-401.	1.8	30

#	Article	IF	Citations
37	High-throughput sequencing of a 4.1â€‰Mb linkage interval reveals FLVCR2 deletions and mutations in lethal cerebral vasculopathy. Human Mutation, 2010, 31, 1134-1141.	2.5	27
38	Hiding in Plain Sight: Molecular Genetics Applied to Giant Congenital Melanocytic Nevi. Journal of Investigative Dermatology, 2014, 134, 879-882.	0.7	27
39	Practical Application of the New Classification Scheme for Congenital Melanocytic Nevi. Pediatric Dermatology, 2015, 32, 23-27.	0.9	25
40	Dissection of the MYCN locus in Feingold syndrome and isolated oesophageal atresia. European Journal of Human Genetics, 2011, 19, 602-606.	2.8	24
41	A Subpopulation of Smooth Muscle Cells, Derived from Melanocyte-Competent Precursors, Prevents Patent Ductus Arteriosus. PLoS ONE, 2013, 8, e53183.	2.5	24
42	Genomeâ€wide DNA methylation analysis identifies MEGF10 as a novel epigenetically repressed candidate tumor suppressor gene in neuroblastoma. Molecular Carcinogenesis, 2017, 56, 1290-1301.	2.7	23
43	Giant congenital melanocytic nevus with vascular malformation and epidermal cysts associated with a somatic activating mutation in <scp>BRAF</scp>. Pigment Cell and Melanoma Research, 2018, 31, 437-441.	3.3	22
44	Methylation-associated PHOX2B gene silencing is a rare event in human neuroblastoma. European Journal of Cancer, 2007, 43, 2366-2372.	2.8	20
45	Refining the clinicopathological pattern of cerebral proliferative glomeruloid vasculopathy (Fowler) Tj ETQq1 1 0.784314 rgBT /Overlo	1.3	18
46	Transcriptome profiling of genes involved in neural tube closure during human embryonic development using long serial analysis of gene expression (longâ€SAGE). Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 683-692.	1.6	18
47	Ectopic expression of Hoxb1 induces cardiac and craniofacial malformations. Genesis, 2018, 56, e23221.	1.6	18
48	Outflow Tract Formationâ€”Embryonic Origins of Conotruncal Congenital Heart Disease. Journal of Cardiovascular Development and Disease, 2021, 8, 42.	1.6	18
49	Epigenetic deregulation of GATA3 in neuroblastoma is associated with increased GATA3 protein expression and with poor outcomes. Scientific Reports, 2019, 9, 18934.	3.3	17
50	Matthew-Wood syndrome: Report of two new cases supporting autosomal recessive inheritance and exclusion ofFGF10 andFGFR2. American Journal of Medical Genetics, Part A, 2007, 143A, 219-228.	1.2	12
51	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, 363-367.	0.6	12
52	Early expression of hypoxia-inducible factor 1Î± in the chicken embryo. Gene Expression Patterns, 2003, 3, 49-52.	0.8	11
53	Widespread dynamic and pleiotropic expression of the melanocortinâ€1â€receptor (MC1R) system is conserved across chick, mouse and human embryonic development. Birth Defects Research, 2018, 110, 443-455.	1.5	11
54	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.	1.5	9

#	Article	IF	Citations
55	Neural Crest and Pituitary Development. , 2001, 4, 13-29.		8
56	Cutaneous Melanocytic Tumors With Concomitant NRAS Q61R and IDH1 R132C Mutations. American Journal of Surgical Pathology, 2020, 44, 1398-1405.	3.7	7
57	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.	1.5	7
58	Somatotroph Tumors and the Epigenetic Status of the GNAS Locus. International Journal of Molecular Sciences, 2021, 22, 7570.	4.1	6
59	Cytogenetic and histological features of a human embryo with homogeneous chromosome 8 trisomy. Prenatal Diagnosis, 2006, 26, 1201-1205.	2.3	5
60	Melanocortinâ€1 receptor (MC1R) genotypes do not correlate with size in two cohorts of mediumâ€toâ€giant congenital melanocytic nevi. Pigment Cell and Melanoma Research, 2020, 33, 685-694.	3.3	5
61	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.	3.7	4
62	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.	0.9	4
63	Cutaneous Melanomas Arising during Childhood: An Overview of the Main Entities. Dermatopathology (Basel, Switzerland), 2021, 8, 301-314.	1.5	3
64	Etiology of Congenital Melanocytic Nevi and Related Conditions. , 2012, , 73-97.		3
65	Sustained experimental activation of FGF8/ERK in the developing chicken spinal cord models early events in ERK-mediated tumorigenesis. Neoplasia, 2022, 24, 120-132.	5.3	3
66	Identification of theIRXBGene Cluster as Candidate Genes in Severe Dysgenesis of the Ocular Anterior Segment. , 2010, 51, 4380.		2
67	Pericyte Ontogeny: The Use of Chimeras to Track a Cell Lineage of Diverse Germ Line Origins. Methods in Molecular Biology, 2021, 2235, 61-87.	0.9	2
68	DNA sequencing and quick clean-up. Protocol Exchange, 0, , .	0.3	2
69	Identification of the Gene Underlying Congenital Short Bowel Syndrome, Pointing to Its Major Role in Intestinal Development. Gastroenterology, 2011, 140, S-89.	1.3	0