Peter J Scambler

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3107591/publications.pdf

Version: 2024-02-01

207 papers	26,864 citations	4960 84 h-index	6300 158 g-index
311 all docs	311 docs citations	311 times ranked	23045 citing authors

#	Article	IF	Citations
1	Localization of the gene for familial adenomatous polyposis on chromosome 5. Nature, 1987, 328, 614-616.	27.8	1,362
2	Distinct Factors Control Histone Variant H3.3 Localization at Specific Genomic Regions. Cell, 2010, 140, 678-691.	28.9	1,069
3	Spectrum of clinical features associated with interstitial chromosome 22q11 deletions: a European collaborative study.. Journal of Medical Genetics, 1997, 34, 798-804.	3.2	1,032
4	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	27.8	1,014
5	22q11.2 deletion syndrome. Nature Reviews Disease Primers, 2015, 1, 15071.	30.5	954
6	Tbx1 haploinsufficiency in the DiGeorge syndrome region causes aortic arch defects in mice. Nature, 2001, 410, 97-101.	27.8	940
7	Hyperdynamic Plasticity of Chromatin Proteins in Pluripotent Embryonic Stem Cells. Developmental Cell, 2006, 10, 105-116.	7.0	915
8	TBX1 Is Responsible for Cardiovascular Defects in Velo-Cardio-Facial/DiGeorge Syndrome. Cell, 2001, 104, 619-629.	28.9	884
9	The gene for familial polyposis coli maps to the long arm of chromosome 5. Science, 1987, 238, 1411-1413.	12.6	666
10	Triallelic Inheritance in Bardet-Biedl Syndrome, a Mendelian Recessive Disorder. Science, 2001, 293, 2256-2259.	12.6	599
11	Localization of cystic fibrosis locus to human chromosome 7cenâ€“q22. Nature, 1985, 318, 384-385.	27.8	494
12	DiGeorge syndrome: part of CATCH 22.. Journal of Medical Genetics, 1993, 30, 852-856.	3.2	452
13	The 22q11 deletion syndromes. Human Molecular Genetics, 2000, 9, 2421-2426.	2.9	447
14	Velo-cardio-facial syndrome: A review of 120 patients. American Journal of Medical Genetics Part A, 1993, 45, 313-319.	2.4	403
15	Velo-cardio-facial syndrome associated with chromosome 22 deletions encompassing the DiGeorge locus. Lancet, The, 1992, 339, 1138-1139.	13.7	399
16	A candidate for the cystic fibrosis locus isolated by selection for methylation-free islands. Nature, 1987, 326, 840-845.	27.8	364
17	Molecular Definition of 22q11 Deletions in 151 Velo-Cardio-Facial Syndrome Patients. American Journal of Human Genetics, 1997, 61, 620-629.	6.2	336
18	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	21.4	326

#	Article	IF	Citations
19	Mutation and deletion of the pseudoautosomal gene SHOX cause Leri-Weill dyschondrosteosis. Nature Genetics, 1998, 19, 70-73.	21.4	316
20	IFT80, which encodes a conserved intraflagellar transport protein, is mutated in Jeune asphyxiating thoracic dystrophy. Nature Genetics, 2007, 39, 727-729.	21.4	310
21	Frasier syndrome is caused by defective alternative splicing of WT1 leading to an altered ratio of WT1 +/-KTS splice isoforms. Human Molecular Genetics, 1998, 7, 709-714.	2.9	303
22	Tbx1 haploinsufficiency is linked to behavioral disorders in mice and humans: Implications for 22q11 deletion syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 7729-7734.	7.1	289
23	VEGF: A modifier of the del22q11 (DiGeorge) syndrome?. Nature Medicine, 2003, 9, 173-182.	30.7	288
24	A homeobox gene, HLXB9, is the major locus for dominantly inherited sacral agenesis. Nature Genetics, 1998, 20, 358-361.	21.4	287
25	Neonatal epileptic encephalopathy caused by mutations in the PNPO gene encoding pyridox(am)ine 5â€²-phosphate oxidase. Human Molecular Genetics, 2005, 14, 1077-1086.	2.9	281
26	Conotruncal anomaly face syndrome is associated with a deletion within chromosome 22q11.. Journal of Medical Genetics, 1993, 30, 822-824.	3.2	250
27	Genotype/phenotype correlations of NPHS1 and NPHS2 mutations in nephrotic syndrome advocate a functional inter-relationship in glomerular filtration. Human Molecular Genetics, 2002, 11, 379-388.	2.9	247
28	Genetic Interaction of BBS1 Mutations with Alleles at Other BBS Loci Can Result in Non-Mendelian Bardet-Biedl Syndrome. American Journal of Human Genetics, 2003, 72, 1187-1199.	6.2	246
29	Microdeletions within 22q11 associated with sporadic and familial DiGeorge syndrome. Genomics, 1991, 10, 201-206.	2.9	241
30	Fraser syndrome and mouse blebbed phenotype caused by mutations in FRAS1/Fras1 encoding a putative extracellular matrix protein. Nature Genetics, 2003, 34, 203-208.	21.4	235
31	Novel HOXA13 Mutations and the Phenotypic Spectrum of Hand-Foot-Genital Syndrome. American Journal of Human Genetics, 2000, 67, 197-202.	6.2	232
32	A common region of 10p deleted in DiGeorge and velocardiofacial syndromes. Nature Genetics, 1996, 13, 458-460.	21.4	214
33	Synpolydactyly phenotypes correlate with size of expansions in HOXD13 polyalanine tract. Proceedings of the National Academy of Sciences of the United States of America, 1997, 94, 7458-7463.	7.1	205
34	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	6.2	196
35	Human haploinsufficiency â€" one for sorrow, two for joy. Nature Genetics, 1994, 7, 5-7.	21.4	193
36	RhoE Regulates Actin Cytoskeleton Organization and Cell Migration. Molecular and Cellular Biology, 1998, 18, 4761-4771.	2.3	191

#	Article	IF	Citations
37	Confirmation that the velo-cardio-facial syndrome is associated with haplo-insufficiency of genes at chromosome 22q11. American Journal of Medical Genetics Part A, 1993, 45, 308-312.	2.4	184
38	Splice-Site Mutations in the Axonemal Outer Dynein Arm Docking Complex Gene CCDC114 Cause Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 92, 88-98.	6.2	176
39	Mutations inCCDC39andCCDC40are the Major Cause of Primary Ciliary Dyskinesia with Axonemal Disorganization and Absent Inner Dynein Arms. Human Mutation, 2013, 34, 462-472.	2.5	176
40	Patterns of polymorphism and linkage disequilibrium for cystic fibrosis. Genomics, 1987, 1, 257-263.	2.9	166
41	Deletions within chromosome 22q11 in familial congenital heart disease. Lancet, The, 1992, 340, 573-575.	13.7	166
42	Evolving Concepts in Human Renal Dysplasia. Journal of the American Society of Nephrology: JASN, 2004, 15, 998-1007.	6.1	159
43	Identification of mutations in CUL7 in 3-M syndrome. Nature Genetics, 2005, 37, 1119-1124.	21.4	158
44	LRP4 Mutations Alter Wnt/Î²-Catenin Signaling and Cause Limb and Kidney Malformations in Cenani-Lenz Syndrome. American Journal of Human Genetics, 2010, 86, 696-706.	6.2	151
45	Identification of a new gene mutated in Fraser syndrome and mouse myelencephalic blebs. Nature Genetics, 2005, 37, 520-525.	21.4	148
46	No evidence for allelic association between schizophrenia and a polymorphism determining high or low catechol O-methyltransferase activity. American Journal of Psychiatry, 1996, 153, 268-270.	7.2	143
47	Isolation of a putative transcriptional regulator from the region of 22q11 deleted in DiGeorge syndrome, Shprintzen syndrome and familial congenital heart disease. Human Molecular Genetics, 1993, 2, 2099-2107.	2.9	140
48	HIRA, a mammalian homologue of Saccharomyces cerevisiae transcriptional co-repressors, interacts with Pax3. Nature Genetics, 1998, 20, 74-77.	21.4	139
49	Mutation Analysis and Embryonic Expression of the HLXB9 Currarino Syndrome Gene. American Journal of Human Genetics, 2000, 66, 1504-1515.	6.2	133
50	Discriminating Power of Localized Three-Dimensional Facial Morphology. American Journal of Human Genetics, 2005, 77, 999-1010.	6.2	133
51	A direct functional link between the multi-PDZ domain protein GRIP1 and the Fraser syndrome protein Fras1. Nature Genetics, 2004, 36, 172-177.	21.4	131
52	Exome sequencing identifiesDYNC2H1mutations as a common cause of asphyxiating thoracic dystrophy (Jeune syndrome) without major polydactyly, renal or retinal involvement. Journal of Medical Genetics, 2013, 50, 309-323.	3.2	127
53	Targeted Mutagenesis of the Hira Gene Results in Gastrulation Defects and Patterning Abnormalities of Mesoendodermal Derivatives Prior to Early Embryonic Lethality. Molecular and Cellular Biology, 2002, 22, 2318-2328.	2.3	126
54	COMT Val108/158Met Modifies Mismatch Negativity and Cognitive Function in 22q11 Deletion Syndrome. Biological Psychiatry, 2005, 58, 23-31.	1.3	126

#	Article	IF	Citations
55	Submicroscopic deletions at 22q11.2: Variability of the clinical picture and delineation of a commonly deleted region. American Journal of Medical Genetics Part A, 1995, 56, 191-197.	2.4	125
56	Tbx1 controls cardiac neural crest cell migration during arch artery development by regulatingGbx2expression in the pharyngeal ectoderm. Development (Cambridge), 2009, 136, 3173-3183.	2.5	124
57	Combined <scp>NGS</scp> Approaches Identify Mutations in the Intraflagellar Transport Gene IFT140 in Skeletal Ciliopathies with Early Progressive Kidney Disease. Human Mutation, 2013, 34, 714-724.	2.5	120
58	Great vessel development requires biallelic expression of Chd7 and Tbx1 in pharyngeal ectoderm in mice. Journal of Clinical Investigation, 2009, 119, 3301-10.	8.2	119
59	Frontorhiny, a Distinctive Presentation of Frontonasal Dysplasia Caused by Recessive Mutations in the ALX3 Homeobox Gene. American Journal of Human Genetics, 2009, 84, 698-705.	6.2	118
60	Short-Rib Polydactyly and Jeune Syndromes Are Caused by Mutations in WDR60. American Journal of Human Genetics, 2013, 93, 515-523.	6.2	116
61	Monozygotic twins with chromosome 22q11 deletion and discordant phenotype.. Journal of Medical Genetics, 1995, 32, 746-748.	3.2	114
62	Molecular and clinical study of 183 patients with conotruncal anomaly face syndrome. Human Genetics, 1998, 103, 70-80.	3.8	114
63	22q11 Deletion Syndrome: A Role for TBX1 in Pharyngeal and Cardiovascular Development. Pediatric Cardiology, 2010, 31, 378-390.	1.3	114
64	BBS4 Is a Minor Contributor to Bardet-Biedl Syndrome and May Also Participate in Triallelic Inheritance. American Journal of Human Genetics, 2002, 71, 22-29.	6.2	110
65	Fras1 deficiency results in cryptophthalmos, renal agenesis and blebbed phenotype in mice. Nature Genetics, 2003, 34, 209-214.	21.4	108
66	Mutations in the Gene Encoding IFT Dynein Complex Component WDR34 Cause Jeune Asphyxiating Thoracic Dystrophy. American Journal of Human Genetics, 2013, 93, 932-944.	6.2	108
67	The CXCL12/CXCR4 Axis Plays a Critical Role in Coronary Artery Development. Developmental Cell, 2015, 33, 455-468.	7.0	108
68	FIRST-TRIMESTER PRENATAL DIAGNOSIS OF CYSTIC FIBROSIS WITH LINKED DNA PROBES. Lancet, The, 1986, 327, 1402-1405.	13.7	106
69	Identification of a New Human Catenin Gene Family Member (ARVCF) from the Region Deleted in Veloâ€“Cardioâ€“Facial Syndrome. Genomics, 1997, 41, 75-83.	2.9	103
70	HumanHOXgene mutations. Clinical Genetics, 2001, 59, 1-11.	2.0	103
71	Cloning the mouse homolog of the human cystic fibrosis transmembrane conductance regulator gene. Genomics, 1991, 10, 301-307.	2.9	100
72	Mutations in SRD5B1 (AKR1D1), the gene encoding Â4-3-oxosteroid 5Â-reductase, in hepatitis and liver failure in infancy. Gut, 2003, 52, 1494-1499.	12.1	100

#	Article	IF	Citations
73	Retinoic acid down-regulatesTbx1 expression in vivo and in vitro. Developmental Dynamics, 2005, 232, 928-938.	1.8	99
74	Cyp26 genes a1, b1 and c1 are down-regulated in Tbx1 null mice and inhibition of Cyp26 enzyme function produces a phenocopy of DiGeorge Syndrome in the chick. Human Molecular Genetics, 2006, 15, 3394-3410.	2.9	98
75	Deletion of 150 kb in the Minimal Digeorge/Velocardiofacial Syndrome Critical Region in Mouse. Human Molecular Genetics, 1999, 8, 2229-2237.	2.9	96
76	Molecular genetics of 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2070-2081.	1.2	96
77	Toward an animal model of cystic fibrosis: targeted interruption of exon 10 of the cystic fibrosis transmembrane regulator gene in embryonic stem cells.. Proceedings of the National Academy of Sciences of the United States of America, 1991, 88, 10730-10734.	7.1	95
78	Deletions in HOXD13 Segregate with an Identical, Novel Foot Malformation in Two Unrelated Families. American Journal of Human Genetics, 1998, 63, 992-1000.	6.2	94
79	Fraser syndrome: A clinical study of 59 cases and evaluation of diagnostic criteria. American Journal of Medical Genetics, Part A, 2007, 143A, 3194-3203.	1.2	93
80	The Primordial Growth Disorder 3-M Syndrome Connects Ubiquitination to the Cytoskeletal Adaptor OBSL1. American Journal of Human Genetics, 2009, 84, 801-806.	6.2	93
81	Molecular Cytogenetic Characterization of the DiGeorge Syndrome Region Using Fluorescence in Situ Hybridization. Genomics, 1993, 17, 403-407.	2.9	91
82	HIRA, a DiGeorge Syndrome Candidate Gene, Is Required for Cardiac Outflow Tract Septation. Circulation Research, 1999, 84, 127-135.	4.5	91
83	A 117-kb Microdeletion Removing HOXD9â€“HOXD13 and EVX2 Causes Synpolydactyly. American Journal of Human Genetics, 2002, 70, 547-555.	6.2	91
84	Mitral valve dynamics in structural and fluidâ€“structure interaction models. Medical Engineering and Physics, 2010, 32, 1057-1064.	1.7	90
85	Isolation of a further anonymous informative DNA sequence from chromosome seven closely linked to cystic fibrosis. Nucleic Acids Research, 1986, 14, 1951-1956.	14.5	88
86	Low-copy-number repeat sequences flank the DiGeorge/velo-cardio-facial syndrome loci at 22q11. Human Molecular Genetics, 1993, 2, 191-196.	2.9	88
87	Combined exome and whole-genome sequencing identifies mutations inARMC4as a cause of primary ciliary dyskinesia with defects in the outer dynein arm. Journal of Medical Genetics, 2014, 51, 61-67.	3.2	88
88	Microarray analysis detects differentially expressed genes in the pharyngeal region of mice lacking Tbx1. Developmental Biology, 2005, 285, 554-569.	2.0	86
89	An Ift80 mouse model of short rib polydactyly syndromes shows defects in hedgehog signalling without loss or malformation of cilia. Human Molecular Genetics, 2011, 20, 1306-1314.	2.9	85
90	MOZ Regulates the Tbx1 Locus, and Moz Mutation Partially Phenocopies DiGeorge Syndrome. Developmental Cell, 2012, 23, 652-663.	7.0	84

#	Article	IF	Citations
91	Tbx1 Regulates the BMP-Smad1 Pathway in a Transcription Independent Manner. PLoS ONE, 2009, 4, e6049.	2.5	80
92	Mutations inGRIP1cause Fraser syndrome. Journal of Medical Genetics, 2012, 49, 303-306.	3.2	79
93	Identification of the complete coding sequence and genomic organization of the Treacher Collins syndrome gene.. Genome Research, 1997, 7, 223-234.	5.5	77
94	CHD7 Maintains Neural Stem Cell Quiescence and Prevents Premature Stem Cell Depletion in the Adult Hippocampus. Stem Cells, 2015, 33, 196-210.	3.2	74
95	The genetics of Fraser syndrome and the blebs mouse mutants. Human Molecular Genetics, 2005, 14, R269-R274.	2.9	72
96	Molecular study of 33 families with Fraser syndrome new data and mutation review. American Journal of Medical Genetics, Part A, 2008, 146A, 2252-2257.	1.2	72
97	Replication and extension studies of inflammatory bowel disease susceptibility regions confirm linkage to chromosome 6p (IBD3). European Journal of Human Genetics, 2001, 9, 627-633.	2.8	70
98	Fras1, a basement membrane-associated protein mutated in Fraser syndrome, mediates both the initiation of the mammalian kidney and the integrity of renal glomeruli. Human Molecular Genetics, 2008, 17, 3953-3964.	2.9	70
99	Tbx1 regulation of myogenic differentiation in the limb and cranial mesoderm. Developmental Dynamics, 2007, 236, 353-363.	1.8	68
100	A locus for asphyxiating thoracic dystrophy, ATD, maps to chromosome 15q13. Journal of Medical Genetics, 2003, 40, 431-435.	3.2	67
101	Common arterial trunk associated with a homeodomain mutation of NKX2.6. Human Molecular Genetics, 2005, 14, 585-593.	2.9	66
102	The 22q11 deletion: DiGeorge and velocardiofacial syndromes and the role of TBX1. Wiley Interdisciplinary Reviews: Developmental Biology, 2013, 2, 393-403.	5.9	64
103	Possible role for COMT in psychosis associated with velo-cardio-facial syndrome. Lancet, The, 1992, 340, 1361-1362.	13.7	63
104	Neural crestâ€“derived SEMA3C activates endothelial NRP1 for cardiac outflow tract septation. Journal of Clinical Investigation, 2015, 125, 2661-2676.	8.2	63
105	AHOXA13 allele with a missense mutation in the homeobox and a dinucleotide deletion in the promoter underlies Guttmacher syndrome. Human Mutation, 2002, 19, 573-574.	2.5	62
106	An I47L substitution in the HOXD13 homeodomain causes a novel human limb malformation by producing a selective loss of function. Development (Cambridge), 2003, 130, 1701-1712.	2.5	61
107	Systematic survey of variants in TBX1 in non-syndromic tetralogy of Fallot identifies a novel 57 base pair deletion that reduces transcriptional activity but finds no evidence for association with common variants. Heart, 2010, 96, 1651-1655.	2.9	61
108	Chromosome 22q11.2 interstitial deletions among childhood-onset schizophrenics and ?multidimensionally impaired?. , 1998, 81, 41-43.		58

#	Article	IF	Citations
109	Isolation of a gene encoding an integral membrane protein from the vicinity of a balanced translocation breakpoint associated with DiGeorge syndrome. Human Molecular Genetics, 1995, 4, 1027-1033.	2.9	57
110	Hes1 expression is reduced in Tbx1 null cells and is required for the development of structures affected in 22q11 deletion syndrome. Developmental Biology, 2010, 340, 369-380.	2.0	57
111	Magnetic resonance virtual histology for embryos: 3D atlases for automated high-throughput phenotyping. NeuroImage, 2011, 54, 769-778.	4.2	57
112	Deregulated FGF and homeotic gene expression underlies cerebellar vermis hypoplasia in CHARGE syndrome. ELife, 2013, 2, e01305.	6.0	55
113	Transposition of the great arteries associated with deletion of chromosome 22q11. American Journal of Cardiology, 1995, 75, 95-98.	1.6	54
114	Nephrin Deficiency Activates NF-Î¹ºB and Promotes Glomerular Injury. Journal of the American Society of Nephrology: JASN, 2009, 20, 1733-1743.	6.1	54
115	Detection of an Atypical 22q11 Deletion That Has No Overlap with the DiGeorge Syndrome Critical Region. American Journal of Human Genetics, 1997, 60, 1544-1547.	6.2	52
116	Detection of an Atypical 22q11 Deletion That Has No Overlap with the DiGeorge Syndrome Critical Region. American Journal of Human Genetics, 1997, 60, 1544-1548.	6.2	51
117	TCTEX1D2 mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. Nature Communications, 2015, 6, 7074.	12.8	51
118	XTbx1 is a transcriptional activator involved in head and pharyngeal arch development inXenopus laevis. Developmental Dynamics, 2005, 232, 979-991.	1.8	49
119	Tbx1 Genetically Interacts With the Transforming Growth Factor-Î²/Bone Morphogenetic Protein Inhibitor Smad7 During Great Vessel Remodeling. Circulation Research, 2013, 112, 90-102.	4.5	46
120	Spatiotemporal dynamics and heterogeneity of renal lymphatics in mammalian development and cystic kidney disease. ELife, 2019, 8, .	6.0	46
121	A Transcription Map in the CATCH22 Critical Region: Identification, Mapping, and Ordering of Four Novel Transcripts Expressed in Heart. Genomics, 1996, 32, 104-112.	2.9	45
122	DNAAF1 links heart laterality with the AAA+ ATPase RUVBL1 and ciliary intraflagellar transport. Human Molecular Genetics, 2018, 27, 529-545.	2.9	45
123	Cloning of the human heparan sulfate-N-deacetylase/N-sulfotransferase gene from the treacher Collins syndrome candidate region at 5q32â€“q33.1. Genomics, 1995, 26, 239-244.	2.9	44
124	Loss of heterozygosity for the short arm of chromosome 7 in sporadic Wilms tumour. Oncogene, 1998, 17, 395-400.	5.9	43
125	SHOX interacts with the chondrogenic transcription factors SOX5 and SOX6 to activate the aggrecan enhancer. Human Molecular Genetics, 2011, 20, 1547-1559.	2.9	43
126	A coming of age: advanced imaging technologies for characterising the developing mouse. Trends in Genetics, 2013, 29, 700-711.	6.7	42

#	Article	IF	Citations
127	Mutations of UFD1L Are Not Responsible for the Majority of Cases of DiGeorge Syndrome/Velocardiofacial Syndrome without Deletions within Chromosome 22q11. American Journal of Human Genetics, 1999, 65, 247-249.	6.2	36
128	Characterization of a familial t(16;22) balanced translocation associated with congenital cataract leads to identification of a novel gene,TMEM114, expressed in the lens and disrupted by the translocation. Human Mutation, 2007, 28, 968-977.	2.5	36
129	The representation of heart development in the gene ontology. Developmental Biology, 2011, 354, 9-17.	2.0	35
130	Partial DiGeorge syndrome in two patients with a 10p rearrangement. Clinical Genetics, 1999, 55, 269-276.	2.0	34
131	Pseudodominant inheritance of Langer mesomelic dysplasia caused by aSHOX homeobox missense mutation. American Journal of Medical Genetics Part A, 2002, 110, 153-157.	2.4	34
132	Dual-probe fluorescence in situ hybridization assay for detecting deletions associated with VCFS/DiGeorge syndrome I and DiGeorge syndrome II loci. American Journal of Medical Genetics Part A, 2000, 91, 313-317.	2.4	33
133	Differential gene expression in the hippocampus of the Df1/+ mice: A model for 22q11.2 deletion syndrome and schizophrenia. Brain Research, 2007, 1139, 48-59.	2.2	33
134	Cardiac phenotyping in ex vivo murine embryos using ÂμMRI. NMR in Biomedicine, 2009, 22, 857-866.	2.8	33
135	Deletions of human chromosome 22 and associated birth defects. Current Opinion in Genetics and Development, 1993, 3, 432-437.	3.3	32
136	Cloning and Comparative Mapping of the DiGeorge Syndrome Critical Region in the Mouse. Genomics, 1998, 52, 37-43.	2.9	32
137	Duplications on Human Chromosome 22 Reveal a Novel Ret Finger Protein-Like Gene Family with Sense and Endogenous Antisense Transcripts. Genome Research, 1999, 9, 803-814.	5.5	32
138	Activation of podocyte Notch mediates early Wt1 glomerulopathy. Kidney International, 2018, 93, 903-920.	5.2	30
139	Mechanisms and cell lineages in lymphatic vascular development. Angiogenesis, 2021, 24, 271-288.	7.2	29
140	Cortical dysgenesis in 2 patients with chromosome 22q11 deletion. Clinical Genetics, 2000, 58, 64-68.	2.0	28
141	A novel 5q11.2 deletion detected by microarray comparative genomic hybridisation in a child referred as a case of suspected 22q11 deletion syndrome. Human Genetics, 2005, 116, 83-90.	3.8	28
142	Cloning and mapping of murine Nfe2l1. Genomics, 1995, 25, 716-719.	2.9	27
143	Microarray analysis of the Df1 mouse model of the 22q11 deletion syndrome. Human Genetics, 2005, 116, 486-496.	3.8	27
144	A critical role for the chromatin remodeller CHD7 in anterior mesoderm during cardiovascular development. Developmental Biology, 2015, 405, 82-95.	2.0	27

#	Article	IF	Citations
145	HIRA Is Required for Heart Development and Directly Regulates Tnni2 and Tnnt3. PLoS ONE, 2016, 11, e0161096.	2.5	27
146	HIC2 Is a Novel Dosage-Dependent Regulator of Cardiac Development Located Within the Distal 22q11 Deletion Syndrome Region. Circulation Research, 2014, 115, 23-31.	4.5	26
147	Sprouty1 Haploinsufficiency Prevents Renal Agenesis in a Model of Fraser Syndrome. Journal of the American Society of Nephrology: JASN, 2012, 23, 1790-1796.	6.1	24
148	Histone Chaperone HIRA in Regulation of Transcription Factor RUNX1. Journal of Biological Chemistry, 2015, 290, 13053-13063.	3.4	24
149	The Genetic Basis of Conotruncal Cardiac Defects. , 1999, , 463-478.		24
150	The Impact of Focused Gene Ontology Curation of Specific Mammalian Systems. PLoS ONE, 2011, 6, e27541.	2.5	23
151	Hearing Loss in a Mouse Model of 22q11.2 Deletion Syndrome. PLoS ONE, 2013, 8, e80104.	2.5	23
152	A rapid and sensitive assay for quantification of siRNA efficiency and specificity. Biological Procedures Online, 2005, 7, 1-7.	2.9	22
153	Evaluation of multiplex capillary heteroduplex analysis: A rapid and sensitive mutation screening technique. Human Mutation, 2003, 22, 151-157.	2.5	21
154	Cloning and mapping of murine Dgcr2 and its homology to the Sez-12 seizure-related protein. Mammalian Genome, 1997, 8, 371-375.	2.2	20
155	Haplotype analysis to determine the position of a mutation among closely linked DNA markers. Human Molecular Genetics, 1993, 2, 1007-1014.	2.9	19
156	Interstitial deletions in DiGeorge syndrome detected with microclones from 22q11. Mammalian Genome, 1992, 3, 101-105.	2.2	18
157	Endogenous Retinoic Acid Activity in Principal Cells and Intercalated Cells of Mouse Collecting Duct System. PLoS ONE, 2011, 6, e16770.	2.5	17
158	Familial gigantism caused by an NSD1 mutation. American Journal of Medical Genetics, Part A, 2005, 139A, 40-44.	1.2	16
159	The cognitive spectrum in velo-cardio-facial syndrome. , 2005, , 147-164.		16
160	Clinical and molecular effects of CHD7 in the heart. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 487-495.	1.6	16
161	Genetic homogeneity of cystic fibrosis. Nucleic Acids Research, 1986, 14, 8681-8686.	14.5	15
162	Direct Selection of Conserved cDNAs from the DiGeorge Critical Region: Isolation of a Novel CDC45-Like Gene. Genome Research, 1998, 8, 834-841.	5.5	15

#	Article	IF	Citations
163	Cardiovascular GO Annotation Initiative Year 1 Report: Why Cardiovascular GO?. Proteomics, 2008, 8, 1950-1953.	2.2	15
164	Absence of the vagus nerve in the stomach of Tbx1â^'/â^' mutant mice. Neurogastroenterology and Motility, 2011, 23, 125-130.	3.0	14
165	Segmentation propagation using a 3D embryo atlas for highâ€throughput MRI phenotyping: Comparison and validation with manual segmentation. Magnetic Resonance in Medicine, 2013, 69, 877-883.	3.0	14
166	HIC2 regulates isoform switching during maturation of the cardiovascular system. Journal of Molecular and Cellular Cardiology, 2018, 114, 29-37.	1.9	14
167	Isolation and genomic characterization of the TUPLE1/HIRA gene of the pufferfish Fugu rubripes. Gene, 1998, 208, 279-283.	2.2	13
168	The Renal Gene Ontology Annotation Initiative. Organogenesis, 2010, 6, 71-75.	1.2	13
169	Tissue Clearing and Deep Imaging of the Kidney Using Confocal and Two-Photon Microscopy. Methods in Molecular Biology, 2020, 2067, 103-126.	0.9	13
170	Cloning of HOX D1 from unfertilised human oocytes and expression analyses during murine oogenesis and embryogenesis. Mechanisms of Development, 2001, 109, 377-381.	1.7	12
171	Loss of CXCL12/CXCR4 signalling impacts several aspects of cardiovascular development but does not exacerbate Tbx1 haploinsufficiency. PLoS ONE, 2018, 13, e0207251.	2.5	11
172	An FDA-Approved Drug Screen for Compounds Influencing Craniofacial Skeletal Development and Craniosynostosis. Molecular Syndromology, 2019, 10, 98-114.	0.8	11
173	Enhanced tissue differentiation in the developing mouse brain using magnetic resonance microâ€histology. Magnetic Resonance in Medicine, 2013, 70, 1380-1388.	3.0	10
174	Mapping of the Tuple1 Gene to Mouse Chromosome 16A-B1. Genomics, 1994, 23, 717-718.	2.9	9
175	Cloning, Chromosome Mapping and Expression Analysis of theHIRAgene fromDrosophila melanogaster. Biochemical and Biophysical Research Communications, 1998, 249, 486-491.	2.1	9
176	Novel exomphalos genetic mouse model: The importance of accurate phenotypic classification. Journal of Pediatric Surgery, 2013, 48, 2036-2042.	1.6	8
177	Increased nuchal translucency origins from abnormal lymphatic development and is independent of the presence of a cardiac defect. Prenatal Diagnosis, 2015, 35, 1278-1286.	2.3	8
178	Dual role for CXCL12 signaling in semilunar valve development. Cell Reports, 2021, 36, 109610.	6.4	8
179	Palatal anomalies and velopharyngeal dysfunction associated with velo-cardio-facial syndrome. , 2005, , 83-104.		7
180	Congenital cardiovascular disease and velo-cardio-facial syndrome. , 2005, , 47-82.		6

#	Article	IF	Citations
181	Generation of mice with a conditional null fraser syndrome 1 (Fras1) allele. Genesis, 2012, 50, 892-898.	1.6	6
182	Analysis of Coronary Vessels in Cleared Embryonic Hearts. Journal of Visualized Experiments, 2016, , .	0.3	6
183	Setd5 is required in cardiopharyngeal mesoderm for heart development and its haploinsufficiency is associated with outflow tract defects in mouse. Genesis, 2021, 59, e23421.	1.6	6
184	Genetic inactivation of Semaphorin 3C protects mice from acute kidney injury. Kidney International, 2022, 101, 720-732.	5.2	6
185	Recombinant DNA techniques in diagnostic and preventive medicine. BioEssays, 1984, 1, 12-15.	2.5	5
186	A novel C2H2 zinc-finger protein gene (ZNF160) maps to human chromosome 19q13.3â€“q13.4. Genomics, 1995, 25, 322-323.	2.9	5
187	Engineering a broken heart. Nature, 1999, 401, 335-337.	27.8	5
188	Neuroimaging in velo-cardio-facial syndrome. , 2005, , 165-180.		5
189	Defective Vagal Innervation in Murine Tbx1 Mutant Hearts. Journal of Cardiovascular Development and Disease, 2018, 5, 49.	1.6	5
190	HIRA directly targets the enhancers of selected cardiac transcription factors during in vitro differentiation of mouse embryonic stem cells. Molecular Biology Reports, 2018, 45, 1001-1011.	2.3	5
191	Immunodeficiency in velo-cardio-facial syndrome. , 2005, , 123-134.		4
192	Bcar1/p130Cas is essential for ventricular development and neural crest cell remodelling of the cardiac outflow tract. Cardiovascular Research, 2022, 118, 1993-2005.	3.8	4
193	Behavioral and psychiatric disorder in velo-cardio-facial syndrome. , 2005, , 135-146.		3
194	Micro-MRI phenotyping of a novel double-knockout mouse model of congenital heart disease. Journal of Cardiovascular Magnetic Resonance, 2010, 12, .	3.3	3
195	Expression of Fraser syndrome genes in normal and polycystic murine kidneys. Pediatric Nephrology, 2012, 27, 991-998.	1.7	3
196	Diffusion microscopic MRI of the mouse embryo: Protocol and practical implementation in the splotch mouse model. Magnetic Resonance in Medicine, 2015, 73, 731-739.	3.0	3
197	Cardiac defects, nuchal edema and abnormal lymphatic development are not associated with morphological changes in the ductus venosus. Early Human Development, 2016, 101, 39-48.	1.8	3
198	The Cystic Fibrosis Locus. Enzyme, 1987, 38, 8-13.	0.7	2

#	Article	IF	Citations
199	Molecular genetics of velo-cardio-facial syndrome. , 2005, , 19-46.		2
200	Probe pJ1 [D7S402] detects a MspI RFLP on chromosome 7q31-32. Nucleic Acids Research, 1989, 17, 1793-1793.	14.5	1
201	Cystic fibrosis. Nature, 1989, 338, 24-24.	27.8	1
202	06-P038 Great vessel development requires dizygous expression of Chd7 and Tbx1 in pharyngeal ectoderm. Mechanisms of Development, 2009, 126, S131.	1.7	1
203	13-P005 Tbx1 controls cardiac neural crest cell migration during arch artery development by regulating Gbx2 expression in the pharyngeal ectoderm. Mechanisms of Development, 2009, 126, S195-S196.	1.7	1
204	Cardiovascular Gene Ontology Annotation Initiative. Nature Precedings, 2009, , .	0.1	0
205	06-P002 Genetic interaction of tbx1 with the Notch pathway effector her6 is required for pharyngeal development in the zebrafish. Mechanisms of Development, 2009, 126, S120.	1.7	0
206	In Amnio MRI of Mouse Embryos. PLoS ONE, 2014, 9, e109143.	2.5	0
207	HOXD13 and Synpolydactyly. , 2016, , 687-691.		0