Christopher Gordon

List of Publications by Year
in descending order

71 papers	2,689 citations	186209 28 h-index	206029 48 g-index
74 all docs	74 docs citations	74 times ranked	4928 citing authors

#	Article	IF	Citations
1	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. Nature Genetics, 2009, 41, 359-364.	9.4	364
2	Disruption of a long distance regulatory region upstream of SOX9 in isolated disorders of sex development. Journal of Medical Genetics, 2011, 48, 825-830.	1.5	162
3	Long-range regulation at the SOX9 locus in development and disease. Journal of Medical Genetics, 2009, 46, 649-656.	1.5	148
4	EFTUD2haploinsufficiency leads to syndromic oesophageal atresia. Journal of Medical Genetics, 2012, 49, 737-746.	1.5	89
5	De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. Nature Genetics, 2017, 49, 249-255.	9.4	88
6	A review of craniofacial disorders caused by spliceosomal defects. Clinical Genetics, 2015, 88, 405-415.	1.0	85
7	De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. American Journal of Human Genetics, 2016, 99, 711-719.	2.6	81
8	A Human Homeotic Transformation Resulting from Mutations in PLCB4 and GNAI3 Causes Auriculocondylar Syndrome. American Journal of Human Genetics, 2012, 90, 907-914.	2.6	75
9	Identification of Novel Craniofacial Regulatory Domains Located far Upstream ofSOX9and Disrupted in Pierre Robin Sequence. Human Mutation, 2014, 35, 1011-1020.	1.1	69
10	Mutations in KCTD1 Cause Scalp-Ear-Nipple Syndrome. American Journal of Human Genetics, 2013, 92, 621-626.	2.6	65
11	MMP21 is mutated in human heterotaxy and is required for normal left-right asymmetry in vertebrates. Nature Genetics, 2015, 47, 1260-1263.	9.4	65
12	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	2.6	61
13	Phenotypic variability of distal 22q11.2 copy number abnormalities. American Journal of Medical Genetics, Part A, 2011, 155, 1623-1633.	0.7	59
14	Mutations in Endothelin 1 Cause Recessive Auriculocondylar Syndrome and Dominant Isolated Question-Mark Ears. American Journal of Human Genetics, 2013, 93, 1118-1125.	2.6	59
15	Delineation ofEFTUD2Haploinsufficiency-Related Phenotypes Through a Series of 36 Patients. Human Mutation, 2014, 35, 478-485.	1.1	50
16	Understanding the basis of auriculocondylar syndrome: Insights from human, mouse and zebrafish genetic studies. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 306-317.	0.7	48
17	Mutations in the Endothelin Receptor Type A Cause Mandibulofacial Dysostosis with Alopecia. American Journal of Human Genetics, 2015, 96, 519-531.	2.6	47
18	Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome. Journal of Medical Genetics, 2013, 50, 174-186.	1.5	44

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19	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. Nature Communications, 2021, 12, 833.	5.8	41
20	Biallelic PPA2 Mutations Cause Sudden Unexpected Cardiac Arrest in Infancy. American Journal of Human Genetics, 2016, 99, 666-673.	2.6	39
21	SLC10A7 mutations cause a skeletal dysplasia with amelogenesis imperfecta mediated by GAG biosynthesis defects. Nature Communications, 2018, 9, 3087.	5.8	39
22	SMCHD1 is involved inde novomethylation of theDUX4-encoding D4Z4 macrosatellite. Nucleic Acids Research, 2019, 47, 2822-2839.	6.5	39
23	De Novo Mutations Affecting the Catalytic CÎ± Subunit of PP2A, PPP2CA, Cause Syndromic Intellectual Disability Resembling Other PP2A-Related Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 104, 139-156.	2.6	39
24	Germline gain-of-function mutations of ALK disrupt central nervous system development. Human Mutation, 2011, 32, 272-276.	1.1	38
25	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	3.7	38
26	Congenital Heart Defects in Patients with Deletions Upstream ofSOX9. Human Mutation, 2013, 34, 1628-1631.	1.1	33
27	Ermap, a gene coding for a novel erythroid specific adhesion/receptor membrane protein. Gene, 2000, 242, 337-345.	1.0	31
28	Functional Assessment of Disease-Associated Regulatory Variants In Vivo Using a Versatile Dual Colour Transgenesis Strategy in Zebrafish. PLoS Genetics, 2015, 11, e1005193.	1.5	31
29	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30
30	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	2.6	30
31	Role of Dlx genes in craniofacial morphogenesis: Dlx2 influences skeletal patterning by inducing ectomesenchymal aggregation in ovo. Evolution & Development, 2010, 12, 459-473.	1.1	29
32	Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La RÃ©union Island, in patients with Fryns syndrome. European Journal of Human Genetics, 2018, 26, 340-349.	1.4	27
33	Bi-allelic Variations of SMO in Humans Cause a Broad Spectrum of Developmental Anomalies Due to Abnormal Hedgehog Signaling. American Journal of Human Genetics, 2020, 106, 779-792.	2.6	25
34	Human ERMAP: An Erythroid Adhesion/Receptor Transmembrane Protein. Blood Cells, Molecules, and Diseases, 2001, 27, 938-949.	0.6	24
35	Enhancer mutations and phenotype modularity. Nature Genetics, 2014, 46, 3-4.	9.4	24
36	Dysregulation of the NRG1/ERBB pathway causes a developmental disorder with gastrointestinal dysmotility in humans. Journal of Clinical Investigation, 2021, 131, .	3.9	24

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37	The RCAS retroviral expression system in the study of skeletal development. Developmental Dynamics, 2009, 238, 797-811.	0.8	23
38	A Recurrent De Novo Nonsense Variant in ZSWIM6 Results in Severe Intellectual Disability without Frontonasal or Limb Malformations. American Journal of Human Genetics, 2017, 101, 995-1005.	2.6	23
39	Further characterization of atypical features in auriculocondylar syndrome caused by recessive PLCB4 mutations. American Journal of Medical Genetics, Part A, 2013, 161, 2339-2346.	0.7	22
40	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	1.1	22
41	Novel variants in GNAI3 associated with auriculocondylar syndrome strengthen a common dominant negative effect. European Journal of Human Genetics, 2015, 23, 481-485.	1.4	21
42	Mandibulofacial dysostosis Guion-Almeida type caused by novel EFTUD2 splice site variants in two Asian children. Clinical Dysmorphology, 2018, 27, 31-35.	0.1	21
43	A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX. European Journal of Medical Genetics, 2014, 57, 587-595.	0.7	18
44	A hypomorphic BMPR1B mutation causes du Pan acromesomelic dysplasia. Orphanet Journal of Rare Diseases, 2015, 10, 84.	1.2	18
45	Efficiency of prenatal diagnosis in Pierre Robin sequence. Prenatal Diagnosis, 2017, 37, 1169-1175.	1.1	18
46	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	2.6	17
47	CXCL14 expression during chick embryonic development. International Journal of Developmental Biology, 2011, 55, 335-340.	0.3	16
48	Novel de novo ZBTB20 mutations in three cases with Primrose syndrome and constant corpus callosum anomalies. American Journal of Medical Genetics, Part A, 2018, 176, 1091-1098.	0.7	16
49	Discovery of a genetic module essential for assigning leftâ€“right asymmetry in humans and ancestral vertebrates. Nature Genetics, 2022, 54, 62-72.	9.4	16
50	C/EBPÎ´Âand C/EBPÎ³ bind the CCAAT-box in the human Î²-globin promoter and modulate the activity of the CACC-box binding protein, EKLF. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2005, 1729, 74-80.	2.4	15
51	Disruption of longâ€distance highly conserved noncoding elements in neurocristopathies. Annals of the New York Academy of Sciences, 2010, 1214, 34-46.	1.8	15
52	Genome-Wide ENU Mutagenesis in Combination with High Density SNP Analysis and Exome Sequencing Provides Rapid Identification of Novel Mouse Models of Developmental Disease. PLoS ONE, 2013, 8, e55429.	1.1	15
53	Heterozygous Mutations in TBX1 as a Cause of Isolated Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 4023-4032.	1.8	15
54	EFTUD2 missense variants disrupt protein function and splicing in mandibulofacial dysostosis Guionâ€Almeida type. Human Mutation, 2020, 41, 1372-1382.	1.1	15

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55	Heterozygous loss of WBP11 function causes multiple congenital defects in humans and mice. Human Molecular Genetics, 2021, 29, 3662-3678.	1.4	14
56	Respiratory and gastrointestinal dysfunctions associated with auriculoâ€condylar syndrome and a homozygous PLCB4 lossâ€ofâ€function mutation. American Journal of Medical Genetics, Part A, 2016, 170, 1471-1478.	0.7	12
57	Developmental perspectives on copy number abnormalities of the 22q11.2 region. Clinical Genetics, 2010, 78, 201-218.	1.0	11
58	Targeted molecular investigation in patients within the clinical spectrum of Auriculocondylar syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 938-945.	0.7	11
59	YPEL1 overexpression in early avian craniofacial mesenchyme causes mandibular dysmorphogenesis by upâ€regulating apoptosis. Developmental Dynamics, 2015, 244, 1022-1030.	0.8	10
60	Altered SOX9 genital tubercle enhancer region in hypospadias. Journal of Steroid Biochemistry and Molecular Biology, 2017, 170, 28-38.	1.2	10
61	MED13L lossâ€ofâ€function variants in two patients with syndromic Pierre Robin sequence. American Journal of Medical Genetics, Part A, 2018, 176, 181-186.	0.7	9
62	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. Genetics in Medicine, 2021, 23, 2415-2425.	1.1	8
63	bfb, a Novel ENU-Induced blebs Mutant Resulting from a Missense Mutation in Fras1. PLoS ONE, 2013, 8, e76342.	1.1	7
64	Clinical evidence for a mandibular to maxillary transformation in Auriculocondylar syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1850-1853.	0.7	6
65	Further delineation of auriculocondylar syndrome based on 14 novel cases and reassessment of 25 published cases. Human Mutation, 2022, 43, 582-594.	1.1	6
66	The association of severe encephalopathy and question mark ear is highly suggestive of loss of <scp>MEF2C</scp> function. Clinical Genetics, 2018, 93, 356-359.	1.0	5
67	A novel de novo PDGFRB variant in a child with severe cerebral malformations, intracerebral calcifications, and infantile myofibromatosis. American Journal of Medical Genetics, Part A, 2019, 179, 1304-1309.	0.7	5
68	Reply: MN1 gene loss-of-function mutation causes cleft palate in a pedigree. Brain, 2021, 144, e19-e19.	3.7	3
69	Biallelic alterations in PLXND1 cause common arterial trunk and other cardiac malformations in humans. Human Molecular Genetics, 2023, 32, 353-356.	1.4	3
70	Cleft palate lateral synechia syndrome in two patients and literature review. International Journal of Oral and Maxillofacial Surgery, 2021, , .	0.7	0
71	Cis-Regulatory Disruption at the SOX9 Locus as a Cause of Pierre Robin Sequence. , 2012, , 123-136.		0