

Christopher Gordon

List of Publications by Year in descending order

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Version: 2024-02-01

71
papers

2,689
citations

186209

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206029

48
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74
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docs citations

74
times ranked

4928
citing authors

#	ARTICLE	IF	CITATIONS
1	Biallelic alterations in <i>PLXND1</i> cause common arterial trunk and other cardiac malformations in humans. <i>Human Molecular Genetics</i> , 2023, 32, 353-356.	1.4	3
2	Further delineation of auriculocondylar syndrome based on 14 novel cases and reassessment of 25 published cases. <i>Human Mutation</i> , 2022, 43, 582-594.	1.1	6
3	Discovery of a genetic module essential for assigning left-right asymmetry in humans and ancestral vertebrates. <i>Nature Genetics</i> , 2022, 54, 62-72.	9.4	16
4	Heterozygous loss of <i>WBP11</i> function causes multiple congenital defects in humans and mice. <i>Human Molecular Genetics</i> , 2021, 29, 3662-3678.	1.4	14
5	Reply: <i>MN1</i> gene loss-of-function mutation causes cleft palate in a pedigree. <i>Brain</i> , 2021, 144, e19-e19.	3.7	3
6	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. <i>Nature Communications</i> , 2021, 12, 833.	5.8	41
7	Dysregulation of the <i>NRG1/ERBB</i> pathway causes a developmental disorder with gastrointestinal dysmotility in humans. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	24
8	Heterozygous <i>ANKRD17</i> loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. <i>American Journal of Human Genetics</i> , 2021, 108, 1138-1150.	2.6	17
9	Cleft palate lateral synchia syndrome in two patients and literature review. <i>International Journal of Oral and Maxillofacial Surgery</i> , 2021, , .	0.7	0
10	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. <i>Genetics in Medicine</i> , 2021, 23, 2415-2425.	1.1	8
11	<i>MN1</i> C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. <i>Brain</i> , 2020, 143, 55-68.	3.7	38
12	Phenotypic spectrum and transcriptomic profile associated with germline variants in <i>TRAF7</i> . <i>Genetics in Medicine</i> , 2020, 22, 1215-1226.	1.1	22
13	<i>EFTUD2</i> missense variants disrupt protein function and splicing in mandibulofacial dysostosis Guion-Almeida type. <i>Human Mutation</i> , 2020, 41, 1372-1382.	1.1	15
14	Bi-allelic Variations of <i>SMO</i> in Humans Cause a Broad Spectrum of Developmental Anomalies Due to Abnormal Hedgehog Signaling. <i>American Journal of Human Genetics</i> , 2020, 106, 779-792.	2.6	25
15	<i>SMCHD1</i> is involved in <i>de novo</i> methylation of the <i>DUX4</i> -encoding D4Z4 macrosatellite. <i>Nucleic Acids Research</i> , 2019, 47, 2822-2839.	6.5	39
16	A novel <i>de novo</i> <i>PDGFRB</i> variant in a child with severe cerebral malformations, intracerebral calcifications, and infantile myofibromatosis. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1304-1309.	0.7	5
17	Missense Variants in the Histone Acetyltransferase Complex Component Gene <i>TRRAP</i> Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	2.6	30
18	De Novo Mutations Affecting the Catalytic C \pm Subunit of PP2A, PPP2CA, Cause Syndromic Intellectual Disability Resembling Other PP2A-Related Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2019, 104, 139-156.	2.6	39

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19	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	2.6	30
20	Novel de novo <i>ZBTB20</i> 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
21	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
22	Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La R�union Island, in patients with Fryns syndrome. European Journal of Human Genetics, 2018, 26, 340-349.	1.4	27
23	The association of severe encephalopathy and question mark ear is highly suggestive of loss of <i>MEF2C</i> function. Clinical Genetics, 2018, 93, 356-359.	1.0	5
24	<i>MED13L</i> loss-of-function variants in two patients with syndromic Pierre Robin sequence. American Journal of Medical Genetics, Part A, 2018, 176, 181-186.	0.7	9
25	Heterozygous Mutations in TBX1 as a Cause of Isolated Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 4023-4032.	1.8	15
26	SLC10A7 mutations cause a skeletal dysplasia with amelogenesis imperfecta mediated by GAG biosynthesis defects. Nature Communications, 2018, 9, 3087.	5.8	39
27	De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. Nature Genetics, 2017, 49, 249-255.	9.4	88
28	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
29	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
30	Efficiency of prenatal diagnosis in Pierre Robin sequence. Prenatal Diagnosis, 2017, 37, 1169-1175.	1.1	18
31	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
32	Altered SOX9 genital tubercle enhancer region in hypospadias. Journal of Steroid Biochemistry and Molecular Biology, 2017, 170, 28-38.	1.2	10
33	Respiratory and gastrointestinal dysfunctions associated with auriculocondylar syndrome and a homozygous PLCB4 loss-of-function mutation. American Journal of Medical Genetics, Part A, 2016, 170, 1471-1478.	0.7	12
34	Biallelic PPA2 Mutations Cause Sudden Unexpected Cardiac Arrest in Infancy. American Journal of Human Genetics, 2016, 99, 666-673.	2.6	39
35	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
36	A hypomorphic BMPR1B mutation causes du Pan acromesomelic dysplasia. Orphanet Journal of Rare Diseases, 2015, 10, 84.	1.2	18

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37	<i>YPEL1</i> overexpression in early avian craniofacial mesenchyme causes mandibular dysmorphogenesis by up-regulating apoptosis. <i>Developmental Dynamics</i> , 2015, 244, 1022-1030.	0.8	10
38	Functional Assessment of Disease-Associated Regulatory Variants In Vivo Using a Versatile Dual Colour Transgenesis Strategy in Zebrafish. <i>PLoS Genetics</i> , 2015, 11, e1005193.	1.5	31
39	Novel variants in <i>GNAI3</i> associated with auriculocondylar syndrome strengthen a common dominant negative effect. <i>European Journal of Human Genetics</i> , 2015, 23, 481-485.	1.4	21
40	A review of craniofacial disorders caused by spliceosomal defects. <i>Clinical Genetics</i> , 2015, 88, 405-415.	1.0	85
41	Mutations in the Endothelin Receptor Type A Cause Mandibulofacial Dysostosis with Alopecia. <i>American Journal of Human Genetics</i> , 2015, 96, 519-531.	2.6	47
42	<i>MMP21</i> is mutated in human heterotaxy and is required for normal left-right asymmetry in vertebrates. <i>Nature Genetics</i> , 2015, 47, 1260-1263.	9.4	65
43	Delineation of <i>EFTUD2</i> Haploinsufficiency-Related Phenotypes Through a Series of 36 Patients. <i>Human Mutation</i> , 2014, 35, 478-485.	1.1	50
44	Clinical evidence for a mandibular to maxillary transformation in Auriculocondylar syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1850-1853.	0.7	6
45	Enhancer mutations and phenotype modularity. <i>Nature Genetics</i> , 2014, 46, 3-4.	9.4	24
46	A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing <i>FBN2</i> and <i>PHAX</i> . <i>European Journal of Medical Genetics</i> , 2014, 57, 587-595.	0.7	18
47	Identification of Novel Craniofacial Regulatory Domains Located far Upstream of <i>SOX9</i> and Disrupted in Pierre Robin Sequence. <i>Human Mutation</i> , 2014, 35, 1011-1020.	1.1	69
48	Mutations in <i>KCTD1</i> Cause Scalp-Ear-Nipple Syndrome. <i>American Journal of Human Genetics</i> , 2013, 92, 621-626.	2.6	65
49	Further characterization of atypical features in auriculocondylar syndrome caused by recessive <i>PLCB4</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2339-2346.	0.7	22
50	Mutations in Endothelin 1 Cause Recessive Auriculocondylar Syndrome and Dominant Isolated Question-Mark Ears. <i>American Journal of Human Genetics</i> , 2013, 93, 1118-1125.	2.6	59
51	Understanding the basis of auriculocondylar syndrome: Insights from human, mouse and zebrafish genetic studies. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2013, 163, 306-317.	0.7	48
52	Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome. <i>Journal of Medical Genetics</i> , 2013, 50, 174-186.	1.5	44
53	Congenital Heart Defects in Patients with Deletions Upstream of <i>SOX9</i> . <i>Human Mutation</i> , 2013, 34, 1628-1631.	1.1	33
54	Genome-Wide ENU Mutagenesis in Combination with High Density SNP Analysis and Exome Sequencing Provides Rapid Identification of Novel Mouse Models of Developmental Disease. <i>PLoS ONE</i> , 2013, 8, e55429.	1.1	15

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55	bfb, a Novel ENU-Induced blebs Mutant Resulting from a Missense Mutation in Fras1. PLoS ONE, 2013, 8, e76342.	1.1	7
56	<i>EFTUD2</i> haploinsufficiency leads to syndromic oesophageal atresia. Journal of Medical Genetics, 2012, 49, 737-746.	1.5	89
57	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
58	Cis-Regulatory Disruption at the SOX9 Locus as a Cause of Pierre Robin Sequence. , 2012, , 123-136.		0
59	CXCL14 expression during chick embryonic development. International Journal of Developmental Biology, 2011, 55, 335-340.	0.3	16
60	Phenotypic variability of distal 22q11.2 copy number abnormalities. American Journal of Medical Genetics, Part A, 2011, 155, 1623-1633.	0.7	59
61	Germline gain-of-function mutations of ALK disrupt central nervous system development. Human Mutation, 2011, 32, 272-276.	1.1	38
62	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
63	Role of <i>Dlx</i> genes in craniofacial morphogenesis: <i>Dlx2</i> influences skeletal patterning by inducing ectomesenchymal aggregation in ovo. Evolution & Development, 2010, 12, 459-473.	1.1	29
64	Disruption of long distance highly conserved noncoding elements in neurocristopathies. Annals of the New York Academy of Sciences, 2010, 1214, 34-46.	1.8	15
65	Developmental perspectives on copy number abnormalities of the 22q11.2 region. Clinical Genetics, 2010, 78, 201-218.	1.0	11
66	Long-range regulation at the SOX9 locus in development and disease. Journal of Medical Genetics, 2009, 46, 649-656.	1.5	148
67	The RCAS retroviral expression system in the study of skeletal development. Developmental Dynamics, 2009, 238, 797-811.	0.8	23
68	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. Nature Genetics, 2009, 41, 359-364.	9.4	364
69	C/EBP β and C/EBP δ bind the CCAAT-box in the human β -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
70	Human ERMAP: An Erythroid Adhesion/Receptor Transmembrane Protein. Blood Cells, Molecules, and Diseases, 2001, 27, 938-949.	0.6	24
71	Ermmap, a gene coding for a novel erythroid specific adhesion/receptor membrane protein. Gene, 2000, 242, 337-345.	1.0	31