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List of Publications by Year
in descending order

Version: 2024-02-01

74 papers	2,847 citations	186265 28 h-index	182427 51 g-index
75 all docs	75 docs citations	75 times ranked	4996 citing authors

#	Article	IF	Citations
1	NOTCH2 Mutations Cause Alagille Syndrome, a Heterogeneous Disorder of the Notch Signaling Pathway. American Journal of Human Genetics, 2006, 79, 169-173.	6.2	663
2	Rhombencephalosynapsis: a hindbrain malformation associated with incomplete separation of midbrain and forebrain, hydrocephalus and a broad spectrum of severity. Brain, 2012, 135, 1370-1386.	7.6	131
3	A genome-wide association study identifies susceptibility loci for nonsyndromic sagittal craniosynostosis near BMP2 and within BBS9. Nature Genetics, 2012, 44, 1360-1364.	21.4	120
4	Modulation of noncanonical TGF-Î² signaling prevents cleft palate in Tgfbr2 mutant mice. Journal of Clinical Investigation, 2012, 122, 873-885.	8.2	104
5	Neuroimaging findings in macrocephalyâ€“capillary malformation: A longitudinal study of 17 patients. American Journal of Medical Genetics, Part A, 2007, 143A, 2981-3008.	1.2	103
6	Clinical Characterization of Patients With Autosomal Dominant Short Stature due to Aggrecan Mutations. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 460-469.	3.6	95
7	Investigation of NRXN1 deletions: Clinical and molecular characterization. American Journal of Medical Genetics, Part A, 2013, 161, 717-731.	1.2	94
8	Fetal constraint as a potential risk factor for craniosynostosis. American Journal of Medical Genetics, Part A, 2010, 152A, 394-400.	1.2	79
9	Smad4-Irf6 genetic interaction and TGFÎ²-mediated IRF6 signaling cascade are crucial for palatal fusion in mice. Development (Cambridge), 2013, 140, 1220-1230.	2.5	74
10	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
11	The Coffinâ€“Siris syndrome: A proposed diagnostic approach and assessment of 15 overlapping cases. American Journal of Medical Genetics, Part A, 2012, 158A, 1865-1876.	1.2	69
12	The morphogenesis of wormian bones: A study of craniosynostosis and purposeful cranial deformation. American Journal of Medical Genetics, Part A, 2007, 143A, 3243-3251.	1.2	68
13	Transforming Growth Factor-Î² Regulates Basal Transcriptional Regulatory Machinery to Control Cell Proliferation and Differentiation in Cranial Neural Crest-derived Osteoprogenitor Cells. Journal of Biological Chemistry, 2010, 285, 4975-4982.	3.4	64
14	Genomic analyses in African populations identify novel risk loci for cleft palate. Human Molecular Genetics, 2019, 28, 1038-1051.	2.9	61
15	Neonatal and Infant Mandibular Distraction as an Alternative to Tracheostomy in Severe Obstructive Sleep Apnea. Cleft Palate-Craniofacial Journal, 2012, 49, 32-38.	0.9	53
16	Fibroblast Growth Factor 9 (FGF9)-Pituitary Homeobox 2 (PITX2) Pathway Mediates Transforming Growth Factor Î² (TGFÎ²) Signaling to Regulate Cell Proliferation in Palatal Mesenchyme during Mouse Palatogenesis. Journal of Biological Chemistry, 2012, 287, 2353-2363.	3.4	52
17	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
18	Hypertrophic scarring in cleft lip repair: a comparison of incidence among ethnic groups. Clinical Epidemiology, 2012, 4, 187.	3.0	44

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19	Microdeletion 20p12.3 involving BMP2 contributes to syndromic forms of cleft palate. American Journal of Medical Genetics, Part A, 2011, 155, 1646-1653.	1.2	43
20	Disruption of the ERK/MAPK pathway in neural crest cells as a potential cause of Pierre Robin sequence. Development (Cambridge), 2015, 142, 3734-45.	2.5	42
21	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
22	TGFÎ² regulates epithelial-mesenchymal interactions through WNT signaling activity to control muscle development in the soft palate. Development (Cambridge), 2014, 141, 909-917.	2.5	41
23	Intraflagellar transport 88 (IFT88) is crucial for craniofacial development in mice and is a candidate gene for human cleft lip and palate. Human Molecular Genetics, 2017, 26, ddx002.	2.9	41
24	Beyond GÃ³mezâ€ŁÃ³pezâ€HernÃ¡ndez syndrome: Recurring phenotypic themes in rhombencephalosynapsis. American Journal of Medical Genetics, Part A, 2012, 158A, 2393-2406.	1.2	40
25	Microdeletion del(22)(q12.2) encompassing the facial development-associated gene, MN1 (meningioma 1) in a child with Pierre-Robin sequence (including cleft palate) and neurofibromatosis 2 (NF2): a case report and review of the literature. BMC Medical Genetics, 2012, 13, 19.	2.1	37
26	Integration of comprehensive 3D microCT and signaling analysis reveals differential regulatory mechanisms of craniofacial bone development. Developmental Biology, 2015, 400, 180-190.	2.0	37
27	Parental risk factors for oral clefts among Central Africans, Southeast Asians, and Central Americans. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 863-879.	1.6	36
28	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
29	Extrinsic Factors Influencing Fetal Deformations and Intrauterine Growth Restriction. Journal of Pregnancy, 2012, 2012, 1-11.	2.4	28
30	ALX4 gain-of-function mutations in nonsyndromic craniosynostosis. Human Mutation, 2012, 33, 1626-1629.	2.5	28
31	Genetic risk factors for orofacial clefts in Central Africans and Southeast Asians. American Journal of Medical Genetics, Part A, 2014, 164, 2572-2580.	1.2	28
32	A Rapid and Sensitive Next-Generation Sequencing Method to Detect RB1 Mutations Improves Care for Retinoblastoma Patients and Their Families. Journal of Molecular Diagnostics, 2016, 18, 480-493.	2.8	26
33	Complex management of a patient with a contiguous Xp11.4 gene deletion involving ornithine transcarbamylase: A role for detailed molecular analysis in complex presentations of classical diseases. Molecular Genetics and Metabolism, 2008, 94, 498-502.	1.1	25
34	Requirement for Jagged1-Notch2 signaling in patterning the bones of the mouse and human middle ear. Scientific Reports, 2017, 7, 2497.	3.3	25
35	An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery. Npj Genomic Medicine, 2018, 3, 21.	3.8	24
36	Phenotype delineation of ZNF462 related syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2075-2082.	1.2	23

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37	Modulation of lipid metabolic defects rescues cleft palate in Tgfbr2 mutant mice. Human Molecular Genetics, 2014, 23, 182-193.	2.9	21
38	Concurrent triplication and uniparental isodisomy: evidence for microhomology-mediated break-induced replication model for genomic rearrangements. European Journal of Human Genetics, 2015, 23, 61-66.	2.8	21
39	A novel nonsense substitution identified in the <scp>AMIGO</scp>2 gene in an Occuloâ€Auriculoâ€Vertebral spectrum patient. Orthodontics and Craniofacial Research, 2019, 22, 163-167.	2.8	20
40	Fatal infantile lactic acidosis and a novel homozygous mutation in the SUCLG1 gene: A mitochondrial DNA depletion disorder. Molecular Genetics and Metabolism, 2011, 102, 149-152.	1.1	17
41	A study of 534 fetal pathology cases from prenatal diagnosis referrals analyzed from 1989 through 2000. American Journal of Medical Genetics, Part A, 2007, 143A, 3107-3120.	1.2	16
42	Bilateral maxillary duplication: case report and literature review. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2012, 113, e29-e32.	0.4	16
43	Proximal variants in <scp>CCND2</scp> associated with microcephaly, short stature, and developmental delay: A case series and review of inverse brain growth phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 2719-2738.	1.2	14
44	Impact of Stem Cells in Craniofacial Regenerative Medicine. Frontiers in Physiology, 2012, 3, 188.	2.8	12
45	Generation and characterization of tamoxifenâ€inducible Pax9â€CreER knockâ€in mice using CrispR/Cas9. Genesis, 2016, 54, 490-496.	1.6	12
46	An Unusual Accessory Mandible and a Submucosal Cleft Palateâ€"A Case Report and Review of the Literature. Cleft Palate-Craniofacial Journal, 2013, 50, 369-375.	0.9	9
47	Congenital Heart Disease in Patients With Cleft Lip/Palate and Its Impact on Cleft Management. Cleft Palate-Craniofacial Journal, 2020, 57, 957-966.	0.9	9
48	Paternal Risk Factors for Oral Clefts in Northern Africans, Southeast Asians, and Central Americans. International Journal of Environmental Research and Public Health, 2017, 14, 657.	2.6	8
49	Risk Factors for Preoperative Developmental Delay in Patients with Nonsyndromic Sagittal Craniosynostosis. Plastic and Reconstructive Surgery, 2019, 143, 133e-139e.	1.4	7
50	A Synonymous Exonic Splice Silencer Variant in IRF6 as a Novel and Cryptic Cause of Non-Syndromic Cleft Lip and Palate. Genes, 2020, 11, 903.	2.4	6
51	Mosaic de novo SNRPN gene variant associated with Prader-Willi syndrome. Journal of Medical Genetics, 2022, 59, 719-722.	3.2	6
52	Clinical and Genomic Approaches for the Diagnosis of Craniofacial Disorders. Current Topics in Developmental Biology, 2015, 115, 543-559.	2.2	5
53	Non-Cystic Fibrosisâ€'Related Meconium Ileus: GUCY2C-Associated Disease Discovered through Rapid Neonatal Whole-Exome Sequencing. Journal of Pediatrics, 2019, 211, 207-210.	1.8	5
54	Further delineation of van den Endeâ€Gupta syndrome: Genetic heterogeneity and overlap with congenital heart defects and skeletal malformations syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2136-2149.	1.2	5

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55	Whole genome sequencing identifies a cryptic <scp>SOX9</scp> regulatory element duplication underlying a case of 46,<scp>XX</scp> ovotesticular difference of sexual development. American Journal of Medical Genetics, Part A, 2021, 185, 2782-2788.	1.2	5
56	Postoperative helmet therapy following frontoâ€orbital advancement and cranial vault remodeling in patients with unilateral coronal synostosis. American Journal of Medical Genetics, Part A, 2021, 185, 2670-2675.	1.2	5
57	X-linked Hypophosphatemic Rickets, del(2)(q37.1;q37.3) Deletion Syndrome and Mosaic Turner Syndrome, mos 45,X/46,X, del(2)(q37.1;q37.3) in a 3-year-old Female. Journal of Bone Metabolism, 2017, 24, 257.	1.3	4
58	Expanding the phenotypic spectrum of RPL13 â€related skeletal dysplasia. American Journal of Medical Genetics, Part A, 2020, 185, 2776-2781.	1.2	4
59	A model for interprofessional health care: lessons learned from craniofacial teams. Journal of the California Dental Association, 2014, 42, 637-44.	0.1	4
60	Situs inversus totalis and prenatal diagnosis of a primary ciliary dyskinesia. Journal of Clinical Ultrasound, 2021, 49, 71-73.	0.8	3
61	Thinking outside â€œThe Boxâ€ Caseâ€based didactics for medical education and the instructional legacy of Dr John M. Graham, Jr. American Journal of Medical Genetics, Part A, 2021, 185, 2636-2645.	1.2	3
62	Hypoxia: A teratogen underlying a range of congenital disruptions, dysplasias, and malformations. American Journal of Medical Genetics, Part A, 2021, 185, 2801-2808.	1.2	3
63	Impact of Genetic and Genomic Testing on the Clinical Management of Patients with Autism Spectrum Disorder. Genes, 2022, 13, 585.	2.4	3
64	High prevalence of deleterious mutations in concomitant nonsyndromic cleft and outflow tract heart defects. American Journal of Medical Genetics, Part A, 2022, , .	1.2	3
65	Role of Pediatric Geneticists in Craniofacial Teams: The Identification of Craniofacial Conditions with Cancer Predisposition. Journal of Pediatrics, 2016, 175, 216-223.e1.	1.8	2
66	Identification of an Identical de Novo SCAMP5 Missense Variant in Four Unrelated Patients With Seizures and Severe Neurodevelopmental Delay. Frontiers in Pharmacology, 2020, 11, 599191.	3.5	2
67	Characterization of sleep habits of children with Sotos syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2815-2820.	1.2	2
68	Congenital Anomalies of the Skull. , 2012, , 247-262.		1
69	Congenital hyperinsulinism in a newborn presenting with poor feeding. SAGE Open Medical Case Reports, 2022, 10, 2050313X2210831.	0.3	1
70	Reintroduction of Diazoxide after Diagnosis of Pulmonary Hypertension in a Patient with Transient Hyperinsulinism. Journal of Child Science, 2021, 11, e80-e82.	0.2	0
71	The International Family Study of Nonsyndromic Orofacial Clefts: Design and Methods. Cleft Palate-Craniofacial Journal, 2022, 59, S37-S47.	0.9	0
72	Pediatric Cushing syndrome: An early sign of an underling cancer predisposition syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2824-2828.	1.2	0

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73	A celebration in honor of John M. Graham, Jr, <scp>MD</scp>, <scp>ScD</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 2617-2619.	1.2	0
74	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0