Maximilian Muenke

List of Publications by Year
in descending order

264 papers	17,561 citations	13865 67 h-index	16650 123 g-index
279 all docs	279 docs citations	279 times ranked	13164 citing authors

#	Article	IF	Citations
1	Mutations in the human Sonic Hedgehog gene cause holoprosencephaly. Nature Genetics, 1996, 14, 357-360.	21.4	1,075
2	A common mutation in the fibroblast growth factor receptor 1 gene in Pfeiffer syndrome. Nature Genetics, 1994, 8, 269-274.	21.4	615
3	Holoprosencephaly due to mutations in ZIC2, a homologue of Drosophila odd-paired. Nature Genetics, 1998, 20, 180-183.	21.4	448
4	Mutations in the homeodomain of the human SIX3 gene cause holoprosencephaly. Nature Genetics, 1999, 22, 196-198.	21.4	398
5	Mutations in TGIF cause holoprosencephaly and link NODAL signalling to human neural axis determination. Nature Genetics, 2000, 25, 205-208.	21.4	368
6	Overexpression of an Osteogenic Morphogen in Fibrodysplasia Ossificans Progressiva. New England Journal of Medicine, 1996, 335, 555-561.	27.0	364
7	Familial dementia caused by polymerization of mutant neuroserpin. Nature, 1999, 401, 376-379.	27.8	342
8	Opitz G/BBB syndrome, a defect of midline development, is due to mutations in a new RING finger gene on Xp22. Nature Genetics, 1997, 17, 285-291.	21.4	331
9	Loss-of-function mutations in the EGF-CFC gene CFC1 are associated with human left-right laterality defects. Nature Genetics, 2000, 26, 365-369.	21.4	319
10	Loss-of-function mutations in the human GLI2 gene are associated with pituitary anomalies and holoprosencephaly-like features. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 13424-13429.	7.1	313
11	Identical mutations in three different fibroblast growth factor receptor genes in autosomal dominant craniosynostosis syndromes. Nature Genetics, 1996, 14, 174-176.	21.4	306
12	Multiple Hits during Early Embryonic Development: Digenic Diseases and Holoprosencephaly. American Journal of Human Genetics, 2002, 71, 1017-1032.	6.2	293
13	Fibroblast-growth-factor receptor mutations in human skeletal disorders. Trends in Genetics, 1995, 11, 308-313.	6.7	292
14	Genetics of ventral forebrain development and holoprosencephaly. Current Opinion in Genetics and Development, 2000, 10, 262-269.	3.3	245
15	Genomic Screening of Fibroblast Growth-Factor Receptor 2 Reveals a Wide Spectrum of Mutations in Patients with Syndromic Craniosynostosis. American Journal of Human Genetics, 2002, 70, 472-486.	6.2	238
16	Mutations in FGFR1 and FGFR2 cause familial and sporadic Pfeiffer syndrome. Human Molecular Genetics, 1995, 4, 323-328.	2.9	221
17	Mutations in PATCHED-1, the receptor for SONIC HEDGEHOG, are associated with holoprosencephaly. Human Genetics, 2002, 110, 297-301.	3.8	220
18	The molecular genetics of holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 52-61.	1.6	220

#	Article	IF	Citations
19	Central Nervous System and Limb Anomalies in Case Reports of First-Trimester Statin Exposure. New England Journal of Medicine, 2004, 350, 1579-1582.	27.0	210
20	CbfÎ² interacts with Runx2 and has a critical role in bone development. Nature Genetics, 2002, 32, 639-644.	21.4	207
21	Young adult outcomes in the followâ€up of the multimodal treatment study of attentionâ€deficit/hyperactivity disorder: symptom persistence, source discrepancy, and height suppression. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2017, 58, 663-678.	5.2	207
22	A functional screen for sonic hedgehog regulatory elements across a 1 Mb interval identifies long-range ventral forebrain enhancers. Development (Cambridge), 2006, 133, 761-772.	2.5	198
23	Mutations in holoprosencephaly. Human Mutation, 2000, 16, 99-108.	2.5	194
24	Holoprosencephaly in RSH/Smith-Lemli-Opitz syndrome: Does abnormal cholesterol metabolism affect the function ofsonic hedgehog?. American Journal of Medical Genetics Part A, 1996, 66, 478-484.	2.4	192
25	Attention-Deficit/Hyperactivity Disorder in a Population Isolate: Linkage to Loci at 4q13.2, 5q33.3, 11q22, and 17p11. American Journal of Human Genetics, 2004, 75, 998-1014.	6.2	192
26	Adverse Birth Outcome Among Mothers With Low Serum Cholesterol. Pediatrics, 2007, 120, 723-733.	2.1	188
27	CFC1 Mutations in Patients with Transposition of the Great Arteries and Double-Outlet Right Ventricle. American Journal of Human Genetics, 2002, 70, 776-780.	6.2	182
28	Regulation of a remote Shh forebrain enhancer by the Six3 homeoprotein. Nature Genetics, 2008, 40, 1348-1353.	21.4	182
29	Mechanistic and epidemiologic considerations in the evaluation of adverse birth outcomes following gestational exposure to statins. American Journal of Medical Genetics Part A, 2004, 131A, 287-298.	2.4	171
30	Metaâ€analysis of genomeâ€wide linkage scans of attention deficit hyperactivity disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1392-1398.	1.7	160
31	SHH mutation is associated with solitary median maxillary central incisor: A study of 13 patients and review of the literature. American Journal of Medical Genetics Part A, 2001, 102, 1-10.	2.4	157
32	A previously unidentified amino-terminal domain regulates transcriptional activity of wild-type and disease-associated human GLI2. Human Molecular Genetics, 2005, 14, 2181-2188.	2.9	156
33	Reduced NODAL Signaling Strength via Mutation of Several Pathway Members Including FOXH1 Is Linked to Human Heart Defects and Holoprosencephaly. American Journal of Human Genetics, 2008, 83, 18-29.	6.2	153
34	Human developmental disorders and the Sonic hedgehog pathway. Trends in Molecular Medicine, 1998, 4, 343-349.	2.6	152
35	Localization of DNA sequences required for human centromere function through an analysis of rearranged Y chromosomes. Nature Genetics, 1993, 5, 368-375.	21.4	149
36	Agenesis and dysgenesis of the corpus callosum: Clinical, genetic and neuroimaging findings in a series of 41 patients. American Journal of Medical Genetics, Part A, 2008, 146A, 2501-2511.	1.2	148

#	Article	IF	Citations
37	Association between conformational mutations in neuroserpin and onset and severity of dementia. Lancet, The, 2002, 359, 2242-2247.	13.7	145
38	Analysis of genotypeâ€"phenotype correlations in human holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 133-141.	1.6	139
39	Novel mutation in sonic hedgehog in nonâ€syndromic colobomatous microphthalmia. American Journal of Medical Genetics Part A, 2003, 116A, 215-221.	2.4	135
40	Holoprosencephaly: from Homer to Hedgehog. Clinical Genetics, 1998, 53, 155-163.	2.0	130
41	Craniosynostosis Syndromes: From Genes to Premature Fusion of Skull Bones. Molecular Genetics and Metabolism, 1999, 68, 139-151.	1.1	123
42	Molecular Mechanisms of Holoprosencephaly. Molecular Genetics and Metabolism, 1999, 68, 126-138.	1.1	121
43	The human osmoregulatory Na+/myo-inositol cotransporter gene (SLC5A3): molecular cloning and localization to chromosome 21. Genomics, 1995, 25, 507-513.	2.9	119
44	Mutations in CDON, Encoding a Hedgehog Receptor, Result in Holoprosencephaly and Defective Interactions with Other Hedgehog Receptors. American Journal of Human Genetics, 2011, 89, 231-240.	6.2	116
45	Title is missing!. Nature, 1999, 401, 376-379.	27.8	113
46	De Novo Mutations in CHD4 , an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. American Journal of Human Genetics, 2016, 99, 934-941.	6.2	111
47	Increased Prevalence of ADHD in Turner Syndrome with No Evidence of Imprinting Effects. Journal of Pediatric Psychology, 2006, 31, 945-955.	2.1	110
48	Symptomatology of autism spectrum disorder in a population with neurofibromatosis type 1. Developmental Medicine and Child Neurology, 2013, 55, 131-138.	2.1	109
49	Opitz syndrome is genetically heterogeneous, with one locus on Xp22, and a second locus on 22q11.2. Nature Genetics, 1995, 11, 459-461.	21.4	103
50	22q11.2 deletion syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 879-888.	1.2	103
51	Muenke syndrome (FGFR3â€related craniosynostosis): Expansion of the phenotype and review of the literature. American Journal of Medical Genetics, Part A, 2007, 143A, 3204-3215.	1.2	99
52	Physical mapping of the holoprosencephaly critical region on chromosome 7q36. Nature Genetics, 1993, 3, 247-251.	21.4	95
53	Identification of a genetic cause for isolated unilateral coronal synostosis: A unique mutation in the fibroblast growth factor receptor 3. Journal of Pediatrics, 1998, 132, 714-716.	1.8	95
54	A loss-of-function mutation in the CFC domain of TDGF1 is associated with human forebrain defects. Human Genetics, 2002, 110, 422-428.	3.8	93

#	Article	IF	Citations
55	Sex differences in neutrophil biology modulate response to type I interferons and immunometabolism. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 16481-16491.	7.1	91
56	Copy-Number Variations Involving the IHH Locus Are Associated with Syndactyly and Craniosynostosis. American Journal of Human Genetics, 2011, 88, 70-75.	6.2	89
57	Fibrodysplasia Ossificans Progressiva, a Heritable Disorder of Severe Heterotopic Ossification, Maps to Human Chromosome 4q27-31*. American Journal of Human Genetics, 2000, 66, 128-135.	6.2	88
58	Opitz G/BBB syndrome: Clinical comparisons of families linked to Xp22 and 22Q and a review of the literature. , 1996, 62, 305-317.		81
59	Compound heterozygosity for mutations in PAX6 in a patient with complex brain anomaly, neonatal diabetes mellitus, and microophthalmia. American Journal of Medical Genetics, Part A, 2009, 149A, 2543-2546.	1.2	80
60	The mutational spectrum of holoprosencephaly-associated changes within theSHHgene in humans predicts loss-of-function through either key structural alterations of the ligand or its altered synthesis. Human Mutation, 2009, 30, E921-E935.	2.5	77
61	The decision to continue: The experiences and needs of parents who receive a prenatal diagnosis of holoprosencephaly. American Journal of Medical Genetics Part A, 2002, 112, 369-378.	2.4	75
62	Mutations in ZIC2 in human holoprosencephaly: description of a Novel ZIC2 specific phenotype and comprehensive analysis of 157 individuals. Journal of Medical Genetics, 2010, 47, 513-524.	3.2	75
63	Down syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 42-53.	1.2	75
64	Human Enteric Defensin Genes: Chromosomal Map Position and a Model for Possible Evolutionary Relationships. Genomics, 1996, 31, 95-106.	2.9	74
65	Latent Class Subtyping of Attention-Deficit/Hyperactivity Disorder and Comorbid Conditions. Journal of the American Academy of Child and Adolescent Psychiatry, 2008, 47, 797-807.	0.5	73
66	Phenotype of the fibroblast growth factor receptor 2 Ser351Cys mutation: Pfeiffer syndrome type III. American Journal of Medical Genetics Part A, 1998, 78, 356-360.	2.4	72
67	Midline and laterality defects: Left and right meet in the middleâ€. BioEssays, 2001, 23, 888-900.	2.5	70
68	Impact of genetics on the diagnosis and clinical management of syndromic craniosynostoses. Child's Nervous System, 2012, 28, 1447-1463.	1.1	70
69	The 2019 US medical genetics workforce: a focus on clinical genetics. Genetics in Medicine, 2021, 23, 1458-1464.	2.4	70
70	Genetic approaches to understanding brain development: Holoprosencephaly as a model. , 2000, 6, 15-21.		69
71	Attention-Deficit/Hyperactivity Disorder and Comorbid Disruptive Behavior Disorders: Evidence of Pleiotropy and New Susceptibility Loci. Biological Psychiatry, 2007, 61, 1329-1339.	1.3	69
72	Review: Genetics of Attention Deficit/Hyperactivity Disorder. Journal of Pediatric Psychology, 2008, 33, 1085-1099.	2.1	69

#	Article	IF	Citations
73	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	1.2	68
74	The interplay of genetic and environmental factors in craniofacial morphogenesis: holoprosencephaly and the role of cholesterol. Congenital Anomalies (discontinued), 2003, 43, 1-21.	0.6	67
75	Cumulative ligand activity of NODAL mutations and modifiers are linked to human heart defects and holoprosencephaly. Molecular Genetics and Metabolism, 2009, 98, 225-234.	1.1	67
76	Genotypic and phenotypic analysis of 396 individuals with mutations inSonic Hedgehog. Journal of Medical Genetics, 2012, 49, 473-479.	3.2	67
77	Attention deficit/hyperactivity disorder (ADHD): Complex phenotype, simple genotype?. Genetics in Medicine, 2004, 6, 1-15.	2.4	65
78	How a Hedgehog might see holoprosencephaly. Human Molecular Genetics, 2003, 12, 15R-25.	2.9	64
79	Opitz G/BBB Syndrome in Xp22: Mutations in the MID1 Gene Cluster in the Carboxy-Terminal Domain. American Journal of Human Genetics, 1998, 63, 703-710.	6.2	63
80	Functional analysis of mutations in TGIF associated with holoprosencephaly. Molecular Genetics and Metabolism, 2007, 90, 97-111.	1.1	63
81	Current recommendations for the molecular evaluation of newly diagnosed holoprosencephaly patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 93-101.	1.6	62
82	Truncating loss-of-function mutations of DISP1 contribute to holoprosencephaly-like microform features in humans. Human Genetics, 2009, 125, 393-400.	3.8	61
83	Molecular Characterization of Breakpoints in Patients with Holoprosencephaly and Definition of the HPE2 Critical Region 2p21. Human Molecular Genetics, 1996, 5, 223-229.	2.9	60
84	A common genetic network underlies substance use disorders and disruptive or externalizing disorders. Human Genetics, 2012, 131, 917-929.	3.8	60
85	REPORTS. Human Molecular Genetics, 1994, 3, 2153-2158.	2.9	59
86	Holoprosencephaly as a genetic model for normal craniofacial development. Seminars in Developmental Biology, 1994, 5, 293-301.	1.3	58
87	Mutations in the human SIX3 gene in holoprosencephaly are loss of function. Human Molecular Genetics, 2008, 17, 3919-3928.	2.9	56
88	The full spectrum of holoprosencephaly-associated mutations within theZIC2gene in humans predicts loss-of-function as the predominant disease mechanism. Human Mutation, 2009, 30, E541-E554.	2.5	56
89	Holoprosencephaly due to numeric chromosome abnormalities. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 146-148.	1.6	56
90	Williamsâ€"Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	1.2	55

#	Article	IF	Citations
91	Pathogenic mutations inGLI2cause a specific phenotype that is distinct from holoprosencephaly. Journal of Medical Genetics, 2014, 51, 413-418.	3.2	53
92	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotypeâ€"phenotype correlations, and molecular basis. Genetics in Medicine, 2020, 22, 389-397.	2.4	53
93	The Topographic Organization of Repetitive DNA in the Human Nucleolus. Genomics, 1993, 15, 123-132.	2.9	52
94	Attention-Deficit/Hyperactivity Disorder and Comorbidities in 18 Paisa Colombian Multigenerational Families. Journal of the American Academy of Child and Adolescent Psychiatry, 2004, 43, 1506-1515.	0.5	52
95	Missense substitutions in the GAS1 protein present in holoprosencephaly patients reduce the affinity for its ligand, SHH. Human Genetics, 2012, 131, 301-310.	3.8	52
96	Heterozygous mutations in SIX3 and SHH are associated with schizencephaly and further expand the clinical spectrum of holoprosencephaly. Human Genetics, 2010, 127, 555-561.	3.8	51
97	Holoprosencephaly: A guide to diagnosis and clinical management. Indian Pediatrics, 2011, 48, 457-466.	0.4	51
98	Screening of human LPHN3 for variants with a potential impact on ADHD susceptibility. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 11-18.	1.7	49
99	Utilizing prospective sequence analysis of SHH, ZIC2, SIX3 and TGIF in holoprosencephaly probands to describe the parameters limiting the observed frequency of mutant geneÃ—gene interactions. Molecular Genetics and Metabolism, 2012, 105, 658-664.	1.1	48
100	An Ultraconserved Brain-Specific Enhancer Within ADGRL3 (LPHN3) Underpins Attention-Deficit/Hyperactivity Disorder Susceptibility. Biological Psychiatry, 2016, 80, 943-954.	1.3	48
101	Noonan syndrome. American Family Physician, 2014, 89, 37-43.	0.1	48
102	On lumping and splitting: A fetus with clinical findings of the oral-facial-digital syndrome type VI, the hydrolethalus syndrome, and the Pallister-Hall syndrome. American Journal of Medical Genetics Part A, 1991, 41, 548-556.	2.4	47
103	Identification of novel mutations in SHH and ZIC2 in a South American (ECLAMC) population with holoprosencephaly. Human Genetics, 2001, 109, 1-6.	3.8	46
104	Dominant-negative kinase domain mutations inFGFR1can explain the clinical severity of Hartsfield syndrome. Human Molecular Genetics, 2016, 25, 1912-1922.	2.9	46
105	Syndromes associated with holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 229-237.	1.6	45
106	Holoprosencephaly: recommendations for diagnosis and management. Current Opinion in Pediatrics, 2010, 22, 687-695.	2.0	44
107	An electronic atlas of human malformation syndromes in diverse populations. Genetics in Medicine, 2016, 18, 1085-1087.	2.4	44
108	Holoprosencephaly: Molecular study of a California Population. , 2000, 90, 315-319.		43

#	Article	IF	Citations
109	Cohesin complex-associated holoprosencephaly. Brain, 2019, 142, 2631-2643.	7.6	43
110	ADHD latent class clusters: DSM-IV subtypes and comorbidity. Psychiatry Research, 2009, 170, 192-198.	3.3	42
111	Cyclopia (synophthalmia) in Smithâ€"Lemliâ€"Opitz syndrome: First reported case and consideration of mechanism. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 142-145.	1.6	40
112	Mutations inSPECC1L, encoding sperm antigen with calponin homology and coiled-coil domains 1-like, are found in some cases of autosomal dominant Opitz G/BBB syndrome. Journal of Medical Genetics, 2015, 52, 104-110.	3.2	40
113	Holoprosencephaly in the genomics era. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 165-174.	1.6	40
114	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
115	SONIC HEDGEHOG mutations causing human holoprosencephaly impair neural patterning activity. Human Genetics, 2003, 113, 170-177.	3.8	38
116	Craniosynostosis and Noonan syndrome with KRAS mutations: Expanding the phenotype with a case report and review of the literature. American Journal of Medical Genetics, Part A, 2015, 167, 2657-2663.	1.2	38
117	Muenke syndrome: An international multicenter natural history study. American Journal of Medical Genetics, Part A, 2016, 170, 918-929.	1.2	37
118	FISH diagnosis of the common 57-kb deletion in CTNS causing cystinosis. Human Genetics, 2004, 115, 510-514.	3.8	36
119	Toward a better understanding of ADHD: LPHN3 gene variants and the susceptibility to develop ADHD. ADHD Attention Deficit and Hyperactivity Disorders, 2010, 2, 139-147.	1.7	36
120	Holoprosencephaly and agnathia spectrum: Presentation of two new patients and review of the literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 158-169.	1.6	36
121	A novel SIX3 mutation segregates with holoprosencephaly in a large family. American Journal of Medical Genetics, Part A, 2009, 149A, 919-925.	1.2	35
122	Abnormal sterol metabolism in holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 102-108.	1.6	35
123	A broad range of ophthalmologic anomalies is part of the holoprosencephaly spectrum. American Journal of Medical Genetics, Part A, 2011, 155, 2713-2720.	1.2	35
124	Influence of a Latrophilin 3 (LPHN3) risk haplotype on event-related potential measures of cognitive response control in attention-deficit hyperactivity disorder (ADHD). European Neuropsychopharmacology, 2013, 23, 458-468.	0.7	35
125	<scp>ADGRL</scp> 3 (<scp>LPHN</scp> 3) variants are associated with a refined phenotype of <scp>ADHD</scp> in the <scp>MTA</scp> study. Molecular Genetics & Genomic Medicine, 2016, 4, 540-547.	1.2	35
126	Additional EFNB1 mutations in craniofrontonasal syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 2008-2012.	1.2	34

#	Article	IF	Citations
127	Structure of the Human Gene Encoding the Associated Microfibrillar Protein (MFAP1) and Localization to Chromosome 15q15-q21. Genomics, 1994, 23, 443-449.	2.9	33
128	Haploinsufficiency of ZNF462 is associated with craniofacial anomalies, corpus callosum dysgenesis, ptosis, and developmental delay. European Journal of Human Genetics, 2017, 25, 946-951.	2.8	33
129	Reciprocal Mouse and Human Limb Phenotypes Caused by Gain- and Loss-of-Function Mutations Affecting Lmbr1. Genetics, 2001, 159, 715-726.	2.9	32
130	A call for global action for rare diseases in Africa. Nature Genetics, 2020, 52, 21-26.	21.4	31
131	The genetics of addiction. Human Genetics, 2012, 131, 773-777.	3.8	30
132	A CCR4-NOT Transcription Complex, Subunit 1, CNOT1, Variant Associated with Holoprosencephaly. American Journal of Human Genetics, 2019, 104, 990-993.	6.2	30
133	Loss-of-Function Variants in PPP1R12A: From Isolated Sex Reversal to Holoprosencephaly Spectrum and Urogenital Malformations. American Journal of Human Genetics, 2020, 106, 121-128.	6.2	30
134	ARP3Î², the gene encoding a new human actin-related protein, is alternatively spliced and predominantly expressed in brain neuronal cells. FEBS Journal, 2000, 267, 2921-2928.	0.2	29
135	Clinical Characteristics of Patients with Unicoronal Synostosis and Mutations of Fibroblast Growth Factor Receptor 3: A Preliminary Report. Plastic and Reconstructive Surgery, 2001, 108, 1849-1854.	1.4	29
136	From the black widow spider to human behavior: Latrophilins, a relatively unknown class of G proteinâ€coupled receptors, are implicated in psychiatric disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 1-10.	1.7	29
137	ADGRL3 (LPHN3) variants predict substance use disorder. Translational Psychiatry, 2019, 9, 42.	4.8	29
138	Fibrodysplasia ossificans progressiva in two half-sisters: Evidence for maternal mosaicism. , 1996, 61, 320-324.		28
139	Mutational analysis of the Sonic Hedgehog gene in 220 newborns with oral clefts in a South American (ECLAMC) populationâ€. American Journal of Medical Genetics Part A, 2002, 108, 12-15.	2.4	28
140	VACTERL Association With High Prenatal Lead Exposure: Similarities to Animal Models of Lead Teratogenicity. Pediatrics, 1991, 87, 390-392.	2.1	28
141	Towards a more representative morphology: clinical and ethical considerations for including diverse populations in diagnostic genetic atlases. Genetics in Medicine, 2016, 18, 1069-1074.	2.4	27
142	Loss of function inROBO1is associated with tetralogy of Fallot and septal defects. Journal of Medical Genetics, 2017, 54, 825-829.	3.2	27
143	BOC is a modifier gene in holoprosencephaly. Human Mutation, 2017, 38, 1464-1470.	2.5	27
144	Hearing loss in syndromic craniosynostoses: Otologic manifestations and clinical findings. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 2037-2047.	1.0	26

#	Article	IF	Citations
145	Whole-Exome Sequencing for Diagnosis of Turner Syndrome: Toward Next-Generation Sequencing and Newborn Screening. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1529-1537.	3.6	26
146	SIX3 mutations with holoprosencephaly. American Journal of Medical Genetics, Part A, 2006, 140A, 2577-2583.	1.2	25
147	Palatal and Oral Manifestations of Muenke Syndrome (FGFR3-Related Craniosynostosis). Journal of Craniofacial Surgery, 2012, 23, 664-668.	0.7	25
148	Common genetic causes of holoprosencephaly are limited to a small set of evolutionarily conserved driver genes of midline development coordinated by TGF-Î², hedgehog, and FGF signaling. Human Mutation, 2018, 39, 1416-1427.	2.5	25
149	Human germline hedgehog pathway mutations predispose to fatty liver. Journal of Hepatology, 2017, 67, 809-817.	3.7	24
150	Holoprosencephaly flashcards: A summary for the clinician. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 3-7.	1.6	23
151	Phenotype delineation of ZNF462 related syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2075-2082.	1.2	23
152	Molecular testing in holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 187-193.	1.6	22
153	When to suspect a genetic syndrome. American Family Physician, 2012, 86, 826-33.	0.1	21
154	Craniosynostosis, Philadelphia type: A new autosomal dominant syndrome with sagittal craniosynostosis and syndactyly of the fingers and toes. , 1996, 62, 184-191.		20
155	Phenotype profile of a genetic mouse model for Muenke syndrome. Child's Nervous System, 2012, 28, 1483-1493.	1.1	20
156	Epilepsy in Muenke Syndrome: FGFR3-Related Craniosynostosis. Pediatric Neurology, 2012, 47, 355-361.	2.1	20
157	Analysis of brain metabolism by proton magnetic resonance spectroscopy (1H-MRS) in attention-deficit/hyperactivity disorder suggests a generalized differential ontogenic pattern from controls. ADHD Attention Deficit and Hyperactivity Disorders, 2012, 4, 205-212.	1.7	20
158	Prenatal exposure to pesticides and risk for holoprosencephaly: a case-control study. Environmental Health, 2020, 19, 65.	4.0	20
159	Polymorphisms in the neural nicotinic acetylcholine receptor Î±4 subunit (CHRNA4) are associated with ADHD in a genetic isolate. ADHD Attention Deficit and Hyperactivity Disorders, 2009, 1, 19-24.	1.7	19
160	Genomics and Epigenomics of Congenital Heart Defects: Expert Review and Lessons Learned in Africa. OMICS A Journal of Integrative Biology, 2018, 22, 301-321.	2.0	18
161	The pit, the cleft and the web. Nature Genetics, 2002, 32, 219-220.	21.4	17
162	Potential cognitive endophenotypes in multigenerational families: segregating ADHD from a genetic isolate. ADHD Attention Deficit and Hyperactivity Disorders, 2011, 3, 291-299.	1.7	17

#	Article	IF	Citations
163	Limb body wall complex, amniotic band sequence, or new syndrome caused by mutation in IQ Motif containing K (IQCK)?. Molecular Genetics & Genomic Medicine, 2015, 3, 424-432.	1.2	17
164	Diversity and dysmorphology. Current Opinion in Pediatrics, 2019, 31, 702-707.	2.0	17
165	Genomic structure, sequence, and mapping of humanFGF8 with no evidence for its role in craniosynostosis/limb defect syndromes. , 1997, 72, 354-362.		16
166	Central Nervous System Embryogenesis and Its Failures. Pediatric and Developmental Pathology, 2002, 5, 425-447.	1.0	16
167	Holoprosencephaly and craniosynostosis: A report of two siblings and review of the literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 176-182.	1.6	16
168	Rubinsteinâ€“Taybi syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 2939-2950.	1.2	16
169	Exome Sequencing and Congenital Heart Disease in Sub-Saharan Africa. Circulation Genomic and Precision Medicine, 2021, 14, e003108.	3.6	16
170	Holoprosencephaly and ectrodactyly: Report of three new patients and review of the literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 170-175.	1.6	15
171	Clinical utility gene card for: Holoprosencephaly. European Journal of Human Genetics, 2011, 19, 3-3.	2.8	15
172	Holoprosencephaly in a family segregating novel variants in ZIC2 and GLI2. , 2011, 155, 860-864.		15
173	Talocalcaneal coalition in Muenke syndrome: Report of a patient, review of the literature in FGFRâ€related craniosynostoses, and consideration of mechanism. American Journal of Medical Genetics, Part A, 2013, 161, 453-460.	1.2	15
174	Expanding the Phenotypic Expression of Sonic Hedgehog Mutations Beyond Holoprosencephaly. Journal of Craniofacial Surgery, 2015, 26, 3-5.	0.7	15
175	In-depth investigations of adolescents and adults with holoprosencephaly identify unique characteristics. Genetics in Medicine, 2018, 20, 14-23.	2.4	15
176	Clinical epidemiology of congenital heart disease in Nigerian children, 2012â€“2017. Birth Defects Research, 2018, 110, 1233-1240.	1.5	15
177	Turner syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 303-313.	1.2	15
178	Sub-band deletion of 7q36.3 in a patient with ring chromosome 7: Association with holoprosencephaly. , 1996, 65, 113-116.		14
179	Novel SNP at the common primer site of exon IIIa ofFGFR2 gene causes error in molecular diagnosis of craniosynostosis syndrome. American Journal of Medical Genetics Part A, 2001, 102, 282-285.	2.4	14
180	Rearrangement in the PITX2 and MIPOL1 genes in a patient with a t(4;14) chromosome. European Journal of Human Genetics, 2003, 11, 315-324.	2.8	14

#	Article	IF	Citations
181	Loss-of-function mutations in FGF8 can be independent risk factors for holoprosencephaly. Human Molecular Genetics, 2018, 27, 1989-1998.	2.9	14
182	Identifying environmental risk factors and <scp>geneâ€"environment</scp> interactions in holoprosencephaly. Birth Defects Research, 2021, 113, 63-76.	1.5	14
183	Clinical and Demographic Evaluation of a Holoprosencephaly Cohort From the Kyoto Collection of Human Embryos. Anatomical Record, 2018, 301, 973-986.	1.4	13
184	Novel heterozygous variants in KMT2D associated with holoprosencephaly. Clinical Genetics, 2019, 96, 266-270.	2.0	13
185	PRDM15 loss of function links NOTCH and WNT/PCP signaling to patterning defects in holoprosencephaly. Science Advances, 2020, 6, eaax9852.	10.3	13
186	Loss of chromosome 8p sequences in human breast carcinoma cell lines. Cancer Genetics and Cytogenetics, 1994, 76, 23-28.	1.0	12
187	Maternally inherited heterozygous sequence change in the sonic hedgehog gene in a male patient with bilateral closedâ€lip schizencephaly and partial absence of the corpus callosum. American Journal of Medical Genetics, Part A, 2009, 149A, 1592-1594.	1.2	12
188	Hearing Loss in Syndromic Craniosynostoses: Introduction and Consideration of Mechanisms. American Journal of Audiology, 2014, 23, 135-141.	1.2	12
189	SIX3 deletions and incomplete penetrance in families affected by holoprosencephaly. Congenital Anomalies (discontinued), 2018, 58, 29-32.	0.6	12
190	Biallelic variants in KYNU cause a multisystemic syndrome with hand hyperphalangism. Bone, 2020, 133, 115219.	2.9	12
191	Anomalies of the forebrain with radial limb defects: Garcia-Lurie-Steinfeld syndrome?. Birth Defects Research Part A: Clinical and Molecular Teratology, 2004, 70, 537-544.	1.6	11
192	Holoprosencephalyâ€"Polydactyly syndrome: In search of an etiology. European Journal of Medical Genetics, 2008, 51, 106-112.	1.3	11
193	Partial deletions of a sequence family (â€œDXS278â€) and its physical linkage to steroid sulfatase as detected by pulsed-field gel electrophoresis. Genomics, 1990, 8, 255-262.	2.9	10
194	Finding genes involved in human developmental disorders. Current Opinion in Genetics and Development, 1995, 5, 354-361.	3.3	10
195	Analysis of Patients with Craniosynostosis Syndromes for a Pro246Arg Mutation in FGFR4. Molecular Genetics and Metabolism, 1998, 64, 76-79.	1.1	10
196	Lobar holoprosencephaly in an infant born to a mother with classic phenylketonuria. American Journal of Medical Genetics Part A, 2000, 95, 187-188.	2.4	10
197	Semilobar holoprosencephaly in a 46,XY female fetus. Prenatal Diagnosis, 2001, 21, 839-841.	2.3	10
198	Minimal evidence for a direct involvement of twisted gastrulation homolog 1 (TWSG1) gene in human holoprosencephalyâ˜†. Molecular Genetics and Metabolism, 2011, 102, 470-480.	1.1	10

#	Article	IF	Citations
199	Medical genetics and genomic medicine in the United States. Part 2: Reproductive genetics, newborn screening, genetic counseling, training, and registries. Molecular Genetics & Genomic Medicine, 2017, 5, 621-630.	1.2	10
200	Regional Assignment of the Human Homeobox-Containing Gene EN1 to Chromosome 2q13-q21. Genomics, 1993, 15, 233-235.	2.9	9
201	Holoprosencephalyâ€“polydactyly/pseudotrisomy 13. Clinical Dysmorphology, 2012, 21, 183-190.	0.3	9
202	Executive Function and Adaptive Behavior in Muenke Syndrome. Journal of Pediatrics, 2015, 167, 428-434.	1.8	9
203	Low-level parental mosaicism affects the recurrence risk of holoprosencephaly. Genetics in Medicine, 2019, 21, 1015-1020.	2.4	9
204	Unique Alterations of an Ultraconserved Non-Coding Element in the 3â€²UTR of ZIC2 in Holoprosencephaly. PLoS ONE, 2012, 7, e39026.	2.5	8
205	Infantile Cirrhosis, Growth Impairment, and Neurodevelopmental Anomalies Associated with Deficiency of PPP1R15B. Journal of Pediatrics, 2016, 179, 144-149.e2.	1.8	8
206	Identification of a novel PCNT founder pathogenic variant in the Israeli Druze population. European Journal of Medical Genetics, 2020, 63, 103643.	1.3	8
207	New Syndrome of Congenital Circumferential Skin Folds Associated with Multiple Congenital Anomalies. Pediatric Dermatology, 2012, 29, 89-95.	0.9	7
208	Holoprosencephaly flashcards: An updated summary for the clinician. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 117-121.	1.6	7
209	Mutations in sphingolipid metabolism genes are associated with ADHD. Translational Psychiatry, 2020, 10, 231.	4.8	7
210	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. Genetics in Medicine, 2021, 23, 1952-1960.	2.4	7
211	Locus homogeneity between syndactyly type 1A and craniosynostosis Philadelphia type?. American Journal of Medical Genetics, Part A, 2008, 146A, 2308-2311.	1.2	6
212	Evidence for SHH as a candidate gene for encephalocele. Clinical Dysmorphology, 2012, 21, 148-151.	0.3	6
213	Comparison of mutation findings in ZIC2 between microform and classical holoprosencephaly in a Brazilian cohort. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 912-917.	1.6	6
214	Individualized genomics and the future of translational medicine. Molecular Genetics & Genomic Medicine, 2013, 1, 1-3.	1.2	6
215	Medical genetics and genomic medicine in the United States of America. Part 1: history, demographics, legislation, and burden of disease. Molecular Genetics & Genomic Medicine, 2017, 5, 307-316.	1.2	6
216	Comorbidity of congenital heart defects and holoprosencephaly is likely genetically driven and geneâ€specific. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 154-158.	1.6	6

#	Article	IF	Citations
217	Introduction to the American Journal of Medical Genetics Part C on holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 1-2.	1.6	5
218	Molecular analysis of the Noggin (NOG) gene in holoprosencephaly patients. Molecular Genetics and Metabolism, 2012, 106, 241-243.	1.1	5
219	Mentors without Borders. Molecular Genetics & Genomic Medicine, 2016, 4, 489-493.	1.2	5
220	Standing on the shoulders of giants. American Journal of Medical Genetics, Part A, 2017, 173, 13-15.	1.2	5
221	Cytogenetics and holoprosencephaly: A chromosomal microarray study of 222 individuals with holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 175-186.	1.6	5
222	Holoprosencephaly: ZIC2 mutation in a case with panhypopituitarism. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 777-81.	0.9	4
223	Retrospective assessment of childhood ADHD symptoms for diagnosis in adults: validity of a short 8-item version of the Wender-Utah Rating Scale. ADHD Attention Deficit and Hyperactivity Disorders, 2016, 8, 215-223.	1.7	4
224	Introducing in AJMG Part A: Case reports in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1547-1548.	1.2	4
225	Challenging issues arising in counseling families experiencing holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 238-245.	1.6	4
226	Tuberous sclerosis in a patient from Nigeria. American Journal of Medical Genetics, Part A, 2019, 179, 1423-1425.	1.2	4
227	Functional analysis of Sonic Hedgehog variants associated with holoprosencephaly in humans using a CRISPR/Cas9 zebrafish model. Human Mutation, 2020, 41, 2155-2166.	2.5	4
228	Fibroblast Growth Factor Receptor-Related Skeletal Disorders. , 1998, , 1029-1038.		4
229	The structure and function of genes causing human holoprosencephaly. Gene Function & Disease, 2000, 1, 7-20.	0.3	3
230	Genetics and genomic medicine around the world. Molecular Genetics & Genomic Medicine, 2014, 2, 1-2.	1.2	3
231	Extracephalic manifestations of nonchromosomal, nonsyndromic holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 246-257.	1.6	3
232	Introducing in AJMG Part A : Genetic Syndromes in Adults. American Journal of Medical Genetics, Part A, 2019, 179, 1413-1414.	1.2	3
233	Rare hypomorphic human variation in the heptahelical domain ofSMOcontributes to holoprosencephaly phenotypes. Human Mutation, 2020, 41, 2105-2118.	2.5	3
234	Quantitative Craniofacial Analysis and Generation of Human Induced Pluripotent Stem Cells for Muenke Syndrome: A Case Report. Journal of Developmental Biology, 2021, 9, 39.	1.7	3

#	Article	IF	Citations
235	Analysis of Smoothened as a candidate gene for human holoprosencephaly. Gene Function & Disease, 2002, 3, 93-97.	0.3	2
236	Deletion of 8q24 in an adult with mild dysmorphic features, developmental delay, and ketotic hypoglycemia. American Journal of Medical Genetics, Part A, 2010, 152A, 1545-1549.	1.2	2
237	The Healing Energy of Breath in Traditional Chinese Medicine and Other Eastern Traditions. , 2012, , 301-323.		2
238	Circle of Willis anomalies in Turner syndrome: Absent A1 segment of the anterior cerebral artery. Birth Defects Research, 2019, 111, 1584-1588.	1.5	2
239	Generation of human induced pluripotent stem cell line (NIDCRi001-A) from a Muenke syndrome patient with an FGFR3 p.Pro250Arg mutation. Stem Cell Research, 2020, 46, 101823.	0.7	2
240	High prenatal lead levels and congenital anomalies. American Journal of Medical Genetics Part A, 1991, 41, 388-388.	2.4	1
241	Determination of the chromosomal location and genomic structure of the Hedgehog-Interacting Protein gene, and analysis of its role in Holoprosencephaly. Gene Function & Disease, 2000, 1, 119-127.	0.3	1
242	Molecular genetics of holoprosencephaly. Frontiers in Bioscience - Landmark, 2000, 5, d334-342.	3.0	1
243	Skeletal Dysplasias: Craniosynostosis Syndromes and Skeletal Dysplasias Caused by Mutations in Fibroblast Growth Factor Receptor Genes. , 0, , 961-991.		1
244	The interplay of genetic and environmental factors in craniofacial morphogenesis: holoprosencephaly and the role of cholesterol. Clinical Genetics, 2003, 43, 1-21.	2.0	1
245	Reply to â€œStatin Drugs and Congenital Anomaliesâ€•by Gibb and Scialli. American Journal of Medical Genetics, Part A, 2005, 135A, 232-234.	1.2	1
246	Holoprosencephaly in an 8.5-week triploidy gestation. Clinical Dysmorphology, 2009, 18, 166-167.	0.3	1
247	2011 William Allan Award Introduction: John M. Opitz 1. American Journal of Human Genetics, 2012, 90, 390-391.	6.2	1
248	The Genetic Workup for Congenital Structural Heart Disease: From Clinical to Genetic Evaluation. , 2015, , 238-256.		1
249	Muenke syndrome. Middle East Journal of Medical Genetics, 2015, 4, 1-6.	0.0	1
250	Neural Plasticity in Obesity and Psychiatric Disorders. Neural Plasticity, 2016, 2016, 1-3.	2.2	1
251	Looking back and looking forward. Molecular Genetics & Genomic Medicine, 2018, 6, 3-8.	1.2	1
252	Early inspirations from times gone by. American Journal of Medical Genetics, Part A, 2018, 176, 1797-1798.	1.2	1

#	Article	IF	Citations
253	Onward and upward. American Journal of Medical Genetics, Part A, 2019, 179, 1119-1121.	1.2	1
254	Love in the time of<scp>COVID</scp>â€19. American Journal of Medical Genetics, Part A, 2020, 182, 1299-1301.	1.2	1
255	Become an ambassador to recruit the next generation in genomic medicine. Genetics in Medicine, 2021, , .	2.4	1
256	Reply to Professor FrÃ©zal. American Journal of Medical Genetics Part A, 1990, 37, 440-440.	2.4	0
257	Molecular Genetics of Holoprosencephaly. Fetal and Pediatric Pathology, 2000, 19, 1-19.	0.3	0
258	Adverse Birth Outcome Among Mothers With Low Serum Cholesterol. Obstetrical and Gynecological Survey, 2008, 63, 81-82.	0.4	0
259	Geneticâ€"environmental interaction in a unique case of Muenke syndrome with intracranial hypertension. Child's Nervous System, 2011, 27, 2183-2186.	1.1	0
260	Patients within the broad holoprosencephaly spectrum have distinct and subtle ophthalmologic anomalies: Response to Khan. American Journal of Medical Genetics, Part A, 2012, 158A, 1244-1245.	1.2	0
261	Cover Image, Volume 173A, Number 4, April 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
262	Cover Image, Volume 173A, Number 9, September 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
263	Introduction. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 113-116.	1.6	0
264	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0