Maximilian Muenke

List of Publications by Year
in descending order

264 papers	17,561 citations	14644 66 h-index	16636 123 g-index
279 all docs	279 docs citations	279 times ranked	13164 citing authors

#	Article	IF	Citations
1	Identifying environmental risk factors and <scp>geneâ€“environment</scp> interactions in holoprosencephaly. Birth Defects Research, 2021, 113, 63-76.	0.8	14
2	Exome Sequencing and Congenital Heart Disease in Sub-Saharan Africa. Circulation Genomic and Precision Medicine, 2021, 14, e003108.	1.6	16
3	The 2019 US medical genetics workforce: a focus on clinical genetics. Genetics in Medicine, 2021, 23, 1458-1464.	1.1	70
4	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. Genetics in Medicine, 2021, 23, 1952-1960.	1.1	7
5	Quantitative Craniofacial Analysis and Generation of Human Induced Pluripotent Stem Cells for Muenke Syndrome: A Case Report. Journal of Developmental Biology, 2021, 9, 39.	0.9	3
6	Become an ambassador to recruit the next generation in genomic medicine. Genetics in Medicine, 2021, , .	1.1	1
7	Identification of a novel PCNT founder pathogenic variant in the Israeli Druze population. European Journal of Medical Genetics, 2020, 63, 103643.	0.7	8
8	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotypeâ€“phenotype correlations, and molecular basis. Genetics in Medicine, 2020, 22, 389-397.	1.1	53
9	Biallelic variants in KYNU cause a multisystemic syndrome with hand hyperphalangism. Bone, 2020, 133, 115219.	1.4	12
10	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.	2.6	30
11	A call for global action for rare diseases in Africa. Nature Genetics, 2020, 52, 21-26.	9.4	31
12	Turner syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 303-313.	0.7	15
13	Rubinsteinâ€“Taybi syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 2939-2950.	0.7	16
14	Mutations in sphingolipid metabolism genes are associated with ADHD. Translational Psychiatry, 2020, 10, 231.	2.4	7
15	Rare hypomorphic human variation in the heptahelical domain ofSMOcontributes to holoprosencephaly phenotypes. Human Mutation, 2020, 41, 2105-2118.	1.1	3
16	Functional analysis of Sonic Hedgehog variants associated with holoprosencephaly in humans using a CRISPR/Cas9 zebrafish model. Human Mutation, 2020, 41, 2155-2166.	1.1	4
17	Love in the time of<scp>COVID</scp>â€19. American Journal of Medical Genetics, Part A, 2020, 182, 1299-1301.	0.7	1
18	Prenatal exposure to pesticides and risk for holoprosencephaly: a case-control study. Environmental Health, 2020, 19, 65.	1.7	20

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19	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.	3.3	91
20	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.	0.3	2
21	PRDM15 loss of function links NOTCH and WNT/PCP signaling to patterning defects in holoprosencephaly. Science Advances, 2020, 6, eaax9852.	4.7	13
22	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.	0.7	6
23	Novel heterozygous variants in KMT2D associated with holoprosencephaly. Clinical Genetics, 2019, 96, 266-270.	1.0	13
24	Cohesin complex-associated holoprosencephaly. Brain, 2019, 142, 2631-2643.	3.7	43
25	Phenotype delineation of ZNF462 related syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2075-2082.	0.7	23
26	Circle of Willis anomalies in Turner syndrome: Absent A1 segment of the anterior cerebral artery. Birth Defects Research, 2019, 111, 1584-1588.	0.8	2
27	ADGRL3 (LPHN3) variants predict substance use disorder. Translational Psychiatry, 2019, 9, 42.	2.4	29
28	Onward and upward. American Journal of Medical Genetics, Part A, 2019, 179, 1119-1121.	0.7	1
29	Tuberous sclerosis in a patient from Nigeria. American Journal of Medical Genetics, Part A, 2019, 179, 1423-1425.	0.7	4
30	Introducing in AJMG Part A : Genetic Syndromes in Adults. American Journal of Medical Genetics, Part A, 2019, 179, 1413-1414.	0.7	3
31	A CCR4-NOT Transcription Complex, Subunit 1, CNOT1, Variant Associated with Holoprosencephaly. American Journal of Human Genetics, 2019, 104, 990-993.	2.6	30
32	Diversity and dysmorphology. Current Opinion in Pediatrics, 2019, 31, 702-707.	1.0	17
33	Low-level parental mosaicism affects the recurrence risk of holoprosencephaly. Genetics in Medicine, 2019, 21, 1015-1020.	1.1	9
34	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	0.7	40
35	Looking back and looking forward. Molecular Genetics & Genomic Medicine, 2018, 6, 3-8.	0.6	1
36	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	0.7	55

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37	Clinical and Demographic Evaluation of a Holoprosencephaly Cohort From the Kyoto Collection of Human Embryos. Anatomical Record, 2018, 301, 973-986.	0.8	13
38	Loss-of-function mutations in FGF8 can be independent risk factors for holoprosencephaly. Human Molecular Genetics, 2018, 27, 1989-1998.	1.4	14
39	In-depth investigations of adolescents and adults with holoprosencephaly identify unique characteristics. Genetics in Medicine, 2018, 20, 14-23.	1.1	15
40	SIX3 deletions and incomplete penetrance in families affected by holoprosencephaly. Congenital Anomalies (discontinued), 2018, 58, 29-32.	0.3	12
41	Introducing in AJMG Part A: Case reports in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1547-1548.	0.7	4
42	Introduction. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 113-116.	0.7	0
43	Clinical epidemiology of congenital heart disease in Nigerian children, 2012â€“2017. Birth Defects Research, 2018, 110, 1233-1240.	0.8	15
44	Molecular testing in holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 187-193.	0.7	22
45	Genomics and Epigenomics of Congenital Heart Defects: Expert Review and Lessons Learned in Africa. OMICS A Journal of Integrative Biology, 2018, 22, 301-321.	1.0	18
46	Extracephalic manifestations of nonchromosomal, nonsyndromic holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 246-257.	0.7	3
47	Holoprosencephaly in the genomics era. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 165-174.	0.7	40
48	Syndromes associated with holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 229-237.	0.7	45
49	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.	0.7	5
50	Challenging issues arising in counseling families experiencing holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 238-245.	0.7	4
51	Holoprosencephaly flashcards: An updated summary for the clinician. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 117-121.	0.7	7
52	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.	1.1	25
53	Early inspirations from times gone by. American Journal of Medical Genetics, Part A, 2018, 176, 1797-1798.	0.7	1
54	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	0.7	0

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55	Haploinsufficiency of ZNF462 is associated with craniofacial anomalies, corpus callosum dysgenesis, ptosis, and developmental delay. European Journal of Human Genetics, 2017, 25, 946-951.	1.4	33
56	Loss of function inROBO1is associated with tetralogy of Fallot and septal defects. Journal of Medical Genetics, 2017, 54, 825-829.	1.5	27
57	22q11.2 deletion syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 879-888.	0.7	103
58	Cover Image, Volume 173A, Number 4, April 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
59	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.	3.1	207
60	Standing on the shoulders of giants. American Journal of Medical Genetics, Part A, 2017, 173, 13-15.	0.7	5
61	Down syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 42-53.	0.7	75
62	Cover Image, Volume 173A, Number 9, September 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
63	BOC is a modifier gene in holoprosencephaly. Human Mutation, 2017, 38, 1464-1470.	1.1	27
64	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	0.7	68
65	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.	0.6	6
66	Human germline hedgehog pathway mutations predispose to fatty liver. Journal of Hepatology, 2017, 67, 809-817.	1.8	24
67	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.	1.8	26
68	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.	0.6	10
69	Neural Plasticity in Obesity and Psychiatric Disorders. Neural Plasticity, 2016, 2016, 1-3.	1.0	1
70	Mentors without Borders. Molecular Genetics & Genomic Medicine, 2016, 4, 489-493.	0.6	5
71	An Ultraconserved Brain-Specific Enhancer Within ADGRL3 (LPHN3) Underpins Attention-Deficit/Hyperactivity Disorder Susceptibility. Biological Psychiatry, 2016, 80, 943-954.	0.7	48
72	Infantile Cirrhosis, Growth Impairment, and Neurodevelopmental Anomalies Associated with Deficiency of PPP1R15B. Journal of Pediatrics, 2016, 179, 144-149.e2.	0.9	8

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73	Muenke syndrome: An international multicenter natural history study. American Journal of Medical Genetics, Part A, 2016, 170, 918-929.	0.7	37
74	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
75	<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.	0.6	35
76	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.	2.6	111
77	Towards a more representative morphology: clinical and ethical considerations for including diverse populations in diagnostic genetic atlases. Genetics in Medicine, 2016, 18, 1069-1074.	1.1	27
78	Dominant-negative kinase domain mutations inFGFR1can explain the clinical severity of Hartsfield syndrome. Human Molecular Genetics, 2016, 25, 1912-1922.	1.4	46
79	An electronic atlas of human malformation syndromes in diverse populations. Genetics in Medicine, 2016, 18, 1085-1087.	1.1	44
80	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.	0.6	17
81	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.	0.7	38
82	The Genetic Workup for Congenital Structural Heart Disease: From Clinical to Genetic Evaluation. , 2015, , 238-256.		1
83	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.	1.5	40
84	Executive Function and Adaptive Behavior in Muenke Syndrome. Journal of Pediatrics, 2015, 167, 428-434.	0.9	9
85	Expanding the Phenotypic Expression of Sonic Hedgehog Mutations Beyond Holoprosencephaly. Journal of Craniofacial Surgery, 2015, 26, 3-5.	0.3	15
86	Muenke syndrome. Middle East Journal of Medical Genetics, 2015, 4, 1-6.	0.0	1
87	Holoprosencephaly: ZIC2 mutation in a case with panhypopituitarism. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 777-81.	0.4	4
88	Hearing loss in syndromic craniosynostoses: Otologic manifestations and clinical findings. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 2037-2047.	0.4	26
89	Genetics and genomic medicine around the world. Molecular Genetics & Genomic Medicine, 2014, 2, 1-2.	0.6	3
90	Hearing Loss in Syndromic Craniosynostoses: Introduction and Consideration of Mechanisms. American Journal of Audiology, 2014, 23, 135-141.	0.5	12

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91	Pathogenic mutations inGLI2cause a specific phenotype that is distinct from holoprosencephaly. Journal of Medical Genetics, 2014, 51, 413-418.	1.5	53
92	Noonan syndrome. American Family Physician, 2014, 89, 37-43.	0.1	48
93	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.	0.7	15
94	Symptomatology of autism spectrum disorder in a population with neurofibromatosis type 1. Developmental Medicine and Child Neurology, 2013, 55, 131-138.	1.1	109
95	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.	0.3	35
96	Individualized genomics and the future of translational medicine. Molecular Genetics & Genomic Medicine, 2013, 1, 1-3.	0.6	6
97	Genotypic and phenotypic analysis of 396 individuals with mutations inSonic Hedgehog. Journal of Medical Genetics, 2012, 49, 473-479.	1.5	67
98	Holoprosencephalyâ€“polydactyly/pseudotrisomy 13. Clinical Dysmorphology, 2012, 21, 183-190.	0.1	9
99	Evidence for SHH as a candidate gene for encephalocele. Clinical Dysmorphology, 2012, 21, 148-151.	0.1	6
100	Palatal and Oral Manifestations of Muenke Syndrome (FGFR3-Related Craniosynostosis). Journal of Craniofacial Surgery, 2012, 23, 664-668.	0.3	25
101	Impact of genetics on the diagnosis and clinical management of syndromic craniosynostoses. Child's Nervous System, 2012, 28, 1447-1463.	0.6	70
102	Phenotype profile of a genetic mouse model for Muenke syndrome. Child's Nervous System, 2012, 28, 1483-1493.	0.6	20
103	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.	0.5	48
104	Molecular analysis of the Noggin (NOG) gene in holoprosencephaly patients. Molecular Genetics and Metabolism, 2012, 106, 241-243.	0.5	5
105	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
106	Epilepsy in Muenke Syndrome: FGFR3-Related Craniosynostosis. Pediatric Neurology, 2012, 47, 355-361.	1.0	20
107	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
108	The Healing Energy of Breath in Traditional Chinese Medicine and Other Eastern Traditions. , 2012, , 301-323.		2

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109	Unique Alterations of an Ultraconserved Non-Coding Element in the 3â€²UTR of ZIC2 in Holoprosencephaly. PLoS ONE, 2012, 7, e39026.	1.1	8
110	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.	0.7	0
111	A common genetic network underlies substance use disorders and disruptive or externalizing disorders. Human Genetics, 2012, 131, 917-929.	1.8	60
112	The genetics of addiction. Human Genetics, 2012, 131, 773-777.	1.8	30
113	2011 William Allan Award Introduction: John M. Opitz 1. American Journal of Human Genetics, 2012, 90, 390-391.	2.6	1
114	New Syndrome of Congenital Circumferential Skin Folds Associated with Multiple Congenital Anomalies. Pediatric Dermatology, 2012, 29, 89-95.	0.5	7
115	Missense substitutions in the GAS1 protein present in holoprosencephaly patients reduce the affinity for its ligand, SHH. Human Genetics, 2012, 131, 301-310.	1.8	52
116	When to suspect a genetic syndrome. American Family Physician, 2012, 86, 826-33.	0.1	21
117	Minimal evidence for a direct involvement of twisted gastrulation homolog 1 (TWSG1) gene in human holoprosencephalyâ˜†. Molecular Genetics and Metabolism, 2011, 102, 470-480.	0.5	10
118	Clinical utility gene card for: Holoprosencephaly. European Journal of Human Genetics, 2011, 19, 3-3.	1.4	15
119	Copy-Number Variations Involving the IHH Locus Are Associated with Syndactyly and Craniosynostosis. American Journal of Human Genetics, 2011, 88, 70-75.	2.6	89
120	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.	2.6	116
121	Geneticâ€“environmental interaction in a unique case of Muenke syndrome with intracranial hypertension. Child's Nervous System, 2011, 27, 2183-2186.	0.6	0
122	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
123	Holoprosencephaly: A guide to diagnosis and clinical management. Indian Pediatrics, 2011, 48, 457-466.	0.2	51
124	Holoprosencephaly in a family segregating novel variants in ZIC2 and GLI2. , 2011, 155, 860-864.		15
125	A broad range of ophthalmologic anomalies is part of the holoprosencephaly spectrum. American Journal of Medical Genetics, Part A, 2011, 155, 2713-2720.	0.7	35
126	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.	1.1	29

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127	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.	1.1	49
128	Holoprosencephaly: recommendations for diagnosis and management. Current Opinion in Pediatrics, 2010, 22, 687-695.	1.0	44
129	Heterozygous mutations in SIX3 and SHH are associated with schizencephaly and further expand the clinical spectrum of holoprosencephaly. Human Genetics, 2010, 127, 555-561.	1.8	51
130	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
131	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.	0.7	2
132	Holoprosencephaly due to numeric chromosome abnormalities. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 146-148.	0.7	56
133	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.	0.7	16
134	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.	0.7	36
135	The molecular genetics of holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 52-61.	0.7	220
136	Analysis of genotypeâ€“phenotype correlations in human holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 133-141.	0.7	139
137	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.	0.7	40
138	Abnormal sterol metabolism in holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 102-108.	0.7	35
139	Holoprosencephaly flashcards: A summary for the clinician. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 3-7.	0.7	23
140	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.	0.7	15
141	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.	0.7	62
142	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.	0.7	5
143	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.	1.5	75
144	A novel SIX3 mutation segregates with holoprosencephaly in a large family. American Journal of Medical Genetics, Part A, 2009, 149A, 919-925.	0.7	35

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145	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.	0.7	12
146	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.	0.7	80
147	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.	1.1	56
148	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.	1.1	77
149	Truncating loss-of-function mutations of DISP1 contribute to holoprosencephaly-like microform features in humans. Human Genetics, 2009, 125, 393-400.	1.8	61
150	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
151	ADHD latent class clusters: DSM-IV subtypes and comorbidity. Psychiatry Research, 2009, 170, 192-198.	1.7	42
152	Cumulative ligand activity of NODAL mutations and modifiers are linked to human heart defects and holoprosencephaly. Molecular Genetics and Metabolism, 2009, 98, 225-234.	0.5	67
153	Holoprosencephaly in an 8.5-week triploidy gestation. Clinical Dysmorphology, 2009, 18, 166-167.	0.1	1
154	Additional EFNB1 mutations in craniofrontonasal syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 2008-2012.	0.7	34
155	Locus homogeneity between syndactyly type 1A and craniosynostosis Philadelphia type?. American Journal of Medical Genetics, Part A, 2008, 146A, 2308-2311.	0.7	6
156	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.	0.7	148
157	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.	1.1	160
158	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.	2.6	153
159	Regulation of a remote Shh forebrain enhancer by the Six3 homeoprotein. Nature Genetics, 2008, 40, 1348-1353.	9.4	182
160	Holoprosencephalyâ€“Polydactyly syndrome: In search of an etiology. European Journal of Medical Genetics, 2008, 51, 106-112.	0.7	11
161	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.	0.3	73
162	Mutations in the human SIX3 gene in holoprosencephaly are loss of function. Human Molecular Genetics, 2008, 17, 3919-3928.	1.4	56

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163	Review: Genetics of Attention Deficit/Hyperactivity Disorder. Journal of Pediatric Psychology, 2008, 33, 1085-1099.	1.1	69
164	Adverse Birth Outcome Among Mothers With Low Serum Cholesterol. Obstetrical and Gynecological Survey, 2008, 63, 81-82.	0.2	0
165	Adverse Birth Outcome Among Mothers With Low Serum Cholesterol. Pediatrics, 2007, 120, 723-733.	1.0	188
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