Maximilian Muenke

List of Publications by Year in Descending Order

252 papers	14,702 citations	63 h-index	115 g-index
279 ext. papers	16,237 ext. citations	7.2 avg, IF	6.11 L-index

#	Paper	IF	Citations
252	HOLOPROSENCEPHALY **2021**, 487-503		
251	The 2019 US medical genetics workforce: a focus on clinical genetics. *Genetics in Medicine*, **2021**, 23, 1458-1464	8.1	8
250	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. *Genetics in Medicine*, **2021**, 23, 1952-1960	8.1	1
249	Identifying environmental risk factors and gene-environment interactions in holoprosencephaly. *Birth Defects Research*, **2021**, 113, 63-76	2.9	8
248	Exome Sequencing and Congenital Heart Disease in Sub-Saharan Africa. *Circulation Genomic and Precision Medicine*, **2021**, 14, e003108	5.2	5
247	Quantitative Craniofacial Analysis and Generation of Human Induced Pluripotent Stem Cells for Muenke Syndrome: A Case Report. *Journal of Developmental Biology*, **2021**, 9,	3.5	0
246	Prenatal exposure to pesticides and risk for holoprosencephaly: a case-control study. *Environmental Health*, **2020**, 19, 65	6	13
245	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	11.5	27
244	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	1.6	1
243	PRDM15 loss of function links NOTCH and WNT/PCP signaling to patterning defects in holoprosencephaly. *Science Advances*, **2020**, 6, eaax9852	14.3	6
242	Comorbidity of congenital heart defects and holoprosencephaly is likely genetically driven and gene-specific. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2020**, 184, 154-158	3.1	4
241	Biallelic variants in KYNU cause a multisystemic syndrome with hand hyperphalangism. *Bone*, **2020**, 133, 115219	4.7	5
240	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	11	14
239	A call for global action for rare diseases in Africa. *Nature Genetics*, **2020**, 52, 21-26	36.3	11
238	Turner syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2020**, 182, 303-313	2.5	8
237	Rubinstein-Taybi syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2939-2950	2.5	4
236	Mutations in sphingolipid metabolism genes are associated with ADHD. *Translational Psychiatry*, **2020**, 10, 231	8.6	3

235	Rare hypomorphic human variation in the heptahelical domain of SMO contributes to holoprosencephaly phenotypes. *Human Mutation*, **2020**, 41, 2105-2118	4.7	3
234	Functional analysis of Sonic Hedgehog variants associated with holoprosencephaly in humans using a CRISPR/Cas9 zebrafish model. *Human Mutation*, **2020**, 41, 2155-2166	4.7	2
233	Identification of a novel PCNT founder pathogenic variant in the Israeli Druze population. *European Journal of Medical Genetics*, **2020**, 63, 103643	2.6	5
232	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotype-phenotype correlations, and molecular basis. *Genetics in Medicine*, **2020**, 22, 389-397	8.1	22
231	ADGRL3 (LPHN3) variants predict substance use disorder. *Translational Psychiatry*, **2019**, 9, 42	8.6	13
230	Tuberous sclerosis in a patient from Nigeria. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1423-1425	2.5	3
229	A CCR4-NOT Transcription Complex, Subunit 1, CNOT1, Variant Associated with Holoprosencephaly. *American Journal of Human Genetics*, **2019**, 104, 990-993	11	22
228	Novel heterozygous variants in KMT2D associated with holoprosencephaly. *Clinical Genetics*, **2019**, 96, 266-270	4	12
227	Cohesin complex-associated holoprosencephaly. *Brain*, **2019**, 142, 2631-2643	11.2	31
226	Phenotype delineation of ZNF462 related syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2075-2082	2.5	11
225	Circle of Willis anomalies in Turner syndrome: Absent A1 segment of the anterior cerebral artery. *Birth Defects Research*, **2019**, 111, 1584-1588	2.9	
224	Diversity and dysmorphology. *Current Opinion in Pediatrics*, **2019**, 31, 702-707	3.2	7
223	Low-level parental mosaicism affects the recurrence risk of holoprosencephaly. *Genetics in Medicine* , **2019**, 21, 1015-1020	8.1	7
222	Cornelia de Lange syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2019**, 179, 150-158	2.5	25
221	Williams-Beuren syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1128-1136	2.5	31
220	Clinical and Demographic Evaluation of a Holoprosencephaly Cohort From the Kyoto Collection of Human Embryos. *Anatomical Record*, **2018**, 301, 973-986	2.1	10
219	Loss-of-function mutations in FGF8 can be independent risk factors for holoprosencephaly. *Human Molecular Genetics*, **2018**, 27, 1989-1998	5.6	12
218	In-depth investigations of adolescents and adults with holoprosencephaly identify unique characteristics. *Genetics in Medicine*, **2018**, 20, 14-23	8.1	13

217	Extracephalic manifestations of nonchromosomal, nonsyndromic holoprosencephaly. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 246-257	3.1	3
216	Holoprosencephaly in the genomics era. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 165-174	3.1	36
215	Syndromes associated with holoprosencephaly. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 229-237	3.1	37
214	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	3.1	5
213	Challenging issues arising in counseling families experiencing holoprosencephaly. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 238-245	3.1	3
212	Holoprosencephaly flashcards: An updated summary for the clinician. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 117-121	3.1	6
211	Common genetic causes of holoprosencephaly are limited to a small set of evolutionarily conserved driver genes of midline development coordinated by TGF-□hedgehog, and FGF signaling. *Human Mutation*, **2018**, 39, 1416-1427	4.7	18
210	SIX3 deletions and incomplete penetrance in families affected by holoprosencephaly. *Congenital Anomalies (discontinued)*, **2018**, 58, 29-32	1.1	11
209	Introduction. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 113-114	3.1	6
208	Clinical epidemiology of congenital heart disease in Nigerian children, 2012-2017. *Birth Defects Research*, **2018**, 110, 1233-1240	2.9	8
207	Molecular testing in holoprosencephaly. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 187-193	3.1	19
206	Genomics and Epigenomics of Congenital Heart Defects: Expert Review and Lessons Learned in Africa. *OMICS A Journal of Integrative Biology*, **2018**, 22, 301-321	3.8	11
205	Haploinsufficiency of ZNF462 is associated with craniofacial anomalies, corpus callosum dysgenesis, ptosis, and developmental delay. *European Journal of Human Genetics*, **2017**, 25, 946-951	5.3	13
204	Loss of function in is associated with tetralogy of Fallot and septal defects. *Journal of Medical Genetics*, **2017**, 54, 825-829	5.8	13
203	22q11.2 deletion syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2017**, 173, 879-888	2.5	67
202	Young adult outcomes in the follow-up of the multimodal treatment study of attention-deficit/hyperactivity disorder: symptom persistence, source discrepancy, and height suppression. *Journal of Child Psychology and Psychiatry and Allied Disciplines*, **2017**, 58, 663-678	7.9	138
201	Down syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2017**, 173, 42-53	2.5	45
200	BOC is a modifier gene in holoprosencephaly. *Human Mutation*, **2017**, 38, 1464-1470	4.7	23

199	Noonan syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2323-2334	2.5	43
198	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	2.3	3
197	Human germline hedgehog pathway mutations predispose to fatty liver. *Journal of Hepatology*, **2017**, 67, 809-817	13.4	18
196	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	5.6	19
195	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	2.3	6
194	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	3.1	2
193	ADGRL3 (LPHN3) variants are associated with a refined phenotype of ADHD in the MTA study. *Molecular Genetics & Genomic Medicine*, **2016**, 4, 540-7	2.3	24
192	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	11	68
191	Towards a more representative morphology: clinical and ethical considerations for including diverse populations in diagnostic genetic atlases. *Genetics in Medicine*, **2016**, 18, 1069-1074	8.1	12
190	Dominant-negative kinase domain mutations in FGFR1 can explain the clinical severity of Hartsfield syndrome. *Human Molecular Genetics*, **2016**, 25, 1912-1922	5.6	32
189	An electronic atlas of human malformation syndromes in diverse populations. *Genetics in Medicine*, **2016**, 18, 1085-1087	8.1	23
188	Mentors without Borders. *Molecular Genetics & Genomic Medicine*, **2016**, 4, 489-93	2.3	2
187	An Ultraconserved Brain-Specific Enhancer Within ADGRL3 (LPHN3) Underpins Attention-Deficit/Hyperactivity Disorder Susceptibility. *Biological Psychiatry*, **2016**, 80, 943-954	7.9	35
186	Infantile Cirrhosis, Growth Impairment, and Neurodevelopmental Anomalies Associated with Deficiency of PPP1R15B. *Journal of Pediatrics*, **2016**, 179, 144-149.e2	3.6	7
185	Muenke syndrome: An international multicenter natural history study. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 918-29	2.5	29
184	Executive Function and Adaptive Behavior in Muenke Syndrome. *Journal of Pediatrics*, **2015**, 167, 428-34	3.6	6
183	Expanding the phenotypic expression of Sonic Hedgehog mutations beyond holoprosencephaly. *Journal of Craniofacial Surgery*, **2015**, 26, 3-5	1.2	15
182	Muenke syndrome. *Middle East Journal of Medical Genetics*, **2015**, 4, 1-6		1

181	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, 424-32	2.3	13
180	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**, 167A, 2657-63	2.5	28
179	Mutations in SPECC1L, 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, 104-10	5.8	26
178	Pathogenic mutations in GLI2 cause a specific phenotype that is distinct from holoprosencephaly. *Journal of Medical Genetics*, **2014**, 51, 413-8	5.8	45
177	Holoprosencephaly: ZIC2 mutation in a case with panhypopituitarism. *Journal of Pediatric Endocrinology and Metabolism*, **2014**, 27, 777-81	1.6	3
176	Hearing loss in syndromic craniosynostoses: otologic manifestations and clinical findings. *International Journal of Pediatric Otorhinolaryngology*, **2014**, 78, 2037-47	1.7	14
175	Genetics and genomic medicine around the world. *Molecular Genetics & Genomic Medicine*, **2014**, 2, 1-2	2.3	1
174	Hearing loss in syndromic craniosynostoses: introduction and consideration of mechanisms. *American Journal of Audiology*, **2014**, 23, 135-41	1.8	9
173	Noonan syndrome. *American Family Physician*, **2014**, 89, 37-43	1.3	32
172	Talocalcaneal coalition in Muenke syndrome: report of a patient, review of the literature in FGFR-related craniosynostoses, and consideration of mechanism. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 453-60	2.5	13
171	Symptomatology of autism spectrum disorder in a population with neurofibromatosis type 1. *Developmental Medicine and Child Neurology*, **2013**, 55, 131-8	3.3	78
170	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, 458-68	1.2	26
169	Individualized genomics and the future of translational medicine. *Molecular Genetics & Genomic Medicine*, **2013**, 1, 1-3	2.3	4
168	Missense substitutions in the GAS1 protein present in holoprosencephaly patients reduce the affinity for its ligand, SHH. *Human Genetics*, **2012**, 131, 301-10	6.3	44
167	Impact of genetics on the diagnosis and clinical management of syndromic craniosynostoses. *Childs Nervous System*, **2012**, 28, 1447-63	1.7	54
166	Phenotype profile of a genetic mouse model for Muenke syndrome. *Childs Nervous System*, **2012**, 28, 1483-93	1.7	19
165	Utilizing prospective sequence analysis of SHH, ZIC2, SIX3 and TGIF in holoprosencephaly probands to describe the parameters limiting the observed frequency of mutant gene gene interactions. *Molecular Genetics and Metabolism*, **2012**, 105, 658-64	3.7	45
164	Molecular analysis of the Noggin (NOG) gene in holoprosencephaly patients. *Molecular Genetics and Metabolism*, **2012**, 106, 241-3	3.7	4

163	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, 912-7		5
162	Epilepsy in Muenke syndrome: FGFR3-related craniosynostosis. *Pediatric Neurology*, **2012**, 47, 355-61	2.9	15
161	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, 205-12	3.1	18
160	The Healing Energy of Breath in Traditional Chinese Medicine and Other Eastern Traditions **2012**, 301-323		2
159	Unique alterations of an ultraconserved non-coding element in the 3'UTR of ZIC2 in holoprosencephaly. *PLoS ONE*, **2012**, 7, e39026	3.7	8
158	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	2.5	
157	A common genetic network underlies substance use disorders and disruptive or externalizing disorders. *Human Genetics*, **2012**, 131, 917-29	6.3	46
156	2011 William Allan Award introduction: John M. Opitz. *American Journal of Human Genetics*, **2012**, 90, 390-1	11	
155	New syndrome of congenital circumferential skin folds associated with multiple congenital anomalies. *Pediatric Dermatology*, **2012**, 29, 89-95	1.9	5
154	Genotypic and phenotypic analysis of 396 individuals with mutations in Sonic Hedgehog. *Journal of Medical Genetics*, **2012**, 49, 473-9	5.8	60
153	Holoprosencephaly-polydactyly/pseudotrisomy 13: a presentation of two new cases and a review of the literature. *Clinical Dysmorphology*, **2012**, 21, 183-190	0.9	7
152	Evidence for SHH as a candidate gene for encephalocele. *Clinical Dysmorphology*, **2012**, 21, 148-151	0.9	5
151	Palatal and oral manifestations of Muenke syndrome (FGFR3-related craniosynostosis). *Journal of Craniofacial Surgery*, **2012**, 23, 664-8	1.2	23
150	When to suspect a genetic syndrome. *American Family Physician*, **2012**, 86, 826-33	1.3	14
149	Minimal evidence for a direct involvement of twisted gastrulation homolog 1 (TWSG1) gene in human holoprosencephaly. *Molecular Genetics and Metabolism*, **2011**, 102, 470-80	3.7	9
148	Clinical utility gene card for: Holoprosencephaly. *European Journal of Human Genetics*, **2011**, 19, preceeding 118-20	5.3	14
147	Copy-number variations involving the IHH locus are associated with syndactyly and craniosynostosis. *American Journal of Human Genetics*, **2011**, 88, 70-5	11	76
146	Mutations in CDON, encoding a hedgehog receptor, result in holoprosencephaly and defective interactions with other hedgehog receptors. *American Journal of Human Genetics*, **2011**, 89, 231-40	11	103

145	Genetic-environmental interaction in a unique case of Muenke syndrome with intracranial hypertension. *Childs Nervous System*, **2011**, 27, 2183-6	1.7	
144	Potential cognitive endophenotypes in multigenerational families: segregating ADHD from a genetic isolate. *ADHD Attention Deficit and Hyperactivity Disorders*, **2011**, 3, 291-9	3.1	12
143	Holoprosencephaly: a guide to diagnosis and clinical management. *Indian Pediatrics*, **2011**, 48, 457-66	1.2	41
142	Holoprosencephaly in a family segregating novel variants in ZIC2 and GLI2. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 860-4	2.5	12
141	A broad range of ophthalmologic anomalies is part of the holoprosencephaly spectrum. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2713-20	2.5	27
140	From the black widow spider to human behavior: Latrophilins, a relatively unknown class of G protein-coupled receptors, are implicated in psychiatric disorders. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2011**, 156B, 1-10	3.5	27
139	Screening of human LPHN3 for variants with a potential impact on ADHD susceptibility. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2011**, 156B, 11-8	3.5	40
138	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, 513-24	5.8	63
137	Holoprosencephaly: recommendations for diagnosis and management. *Current Opinion in Pediatrics* , **2010**, 22, 687-95	3.2	38
136	Holoprosencephaly **2010**, 441-460		
135	Heterozygous mutations in SIX3 and SHH are associated with schizencephaly and further expand the clinical spectrum of holoprosencephaly. *Human Genetics*, **2010**, 127, 555-61	6.3	36
134	Toward a better understanding of ADHD: LPHN3 gene variants and the susceptibility to develop ADHD. *ADHD Attention Deficit and Hyperactivity Disorders*, **2010**, 2, 139-47	3.1	26
133	Deletion of 8q24 in an adult with mild dysmorphic features, developmental delay, and ketotic hypoglycemia. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1545-9	2.5	2
132	Holoprosencephaly due to numeric chromosome abnormalities. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2010**, 154C, 146-8	3.1	47
131	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, 176-82	3.1	12
130	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, 158-69	3.1	29
129	The molecular genetics of holoprosencephaly. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2010**, 154C, 52-61	3.1	186
128	Analysis of genotype-phenotype correlations in human holoprosencephaly. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2010**, 154C, 133-41	3.1	114

127	Cyclopia (synophthalmia) in Smith-Lemli-Opitz syndrome: First reported case and consideration of mechanism. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2010**, 154C, 142-5	3.1	37
126	Abnormal sterol metabolism in holoprosencephaly. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2010**, 154C, 102-8	3.1	33
125	Holoprosencephaly flashcards: A summary for the clinician. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2010**, 154C, 3-7	3.1	18
124	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, 170-5	3.1	14
123	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	3.1	54
122	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	3.1	3
121	A novel SIX3 mutation segregates with holoprosencephaly in a large family. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 919-25	2.5	30
120	Maternally inherited heterozygous sequence change in the sonic hedgehog gene in a male patient with bilateral closed-lip schizencephaly and partial absence of the corpus callosum. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1592-4	2.5	9
119	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, 2543-6	2.5	69
118	The full spectrum of holoprosencephaly-associated mutations within the ZIC2 gene in humans predicts loss-of-function as the predominant disease mechanism. *Human Mutation*, **2009**, 30, E541-54	4.7	52
117	The mutational spectrum of holoprosencephaly-associated changes within the SHH gene in humans predicts loss-of-function through either key structural alterations of the ligand or its altered synthesis. *Human Mutation*, **2009**, 30, E921-35	4.7	64
116	Truncating loss-of-function mutations of DISP1 contribute to holoprosencephaly-like microform features in humans. *Human Genetics*, **2009**, 125, 393-400	6.3	56
115	Polymorphisms in the neural nicotinic acetylcholine receptor 4 subunit (CHRNA4) are associated with ADHD in a genetic isolate. *ADHD Attention Deficit and Hyperactivity Disorders*, **2009**, 1, 19-24	3.1	16
114	ADHD latent class clusters: DSM-IV subtypes and comorbidity. *Psychiatry Research*, **2009**, 170, 192-8	9.9	32
113	Cumulative ligand activity of NODAL mutations and modifiers are linked to human heart defects and holoprosencephaly. *Molecular Genetics and Metabolism*, **2009**, 98, 225-34	3.7	61
112	Holoprosencephaly in an 8.5-week triploidy gestation. *Clinical Dysmorphology*, **2009**, 18, 166-167	0.9	1
111	Regulation of a remote Shh forebrain enhancer by the Six3 homeoprotein. *Nature Genetics*, **2008**, 40, 1348-53	36.3	153
110	Holoprosencephaly-Polydactyly syndrome: in search of an etiology. *European Journal of Medical Genetics*, **2008**, 51, 106-12	2.6	10

109	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	7.2	65
108	Mutations in the human SIX3 gene in holoprosencephaly are loss of function. *Human Molecular Genetics*, **2008**, 17, 3919-28	5.6	48
107	Review: Genetics of attention deficit/hyperactivity disorder. *Journal of Pediatric Psychology*, **2008**, 33, 1085-99	3.2	57
106	Additional EFNB1 mutations in craniofrontonasal syndrome. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2008-12	2.5	25
105	Locus homogeneity between syndactyly type 1A and craniosynostosis Philadelphia type?. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2308-11	2.5	6
104	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, 2501-11	2.5	107
103	Meta-analysis of genome-wide linkage scans of attention deficit hyperactivity disorder. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2008**, 147B, 1392-8	3.5	131
102	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	11	137
101	Attention-deficit/hyperactivity disorder and comorbid disruptive behavior disorders: evidence of pleiotropy and new susceptibility loci. *Biological Psychiatry*, **2007**, 61, 1329-39	7.9	63
100	Muenke syndrome (FGFR3-related craniosynostosis): expansion of the phenotype and review of the literature. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 3204-15	2.5	83
99	Adverse birth outcome among mothers with low serum cholesterol. *Pediatrics*, **2007**, 120, 723-33	7.4	156
98	Functional analysis of mutations in TGIF associated with holoprosencephaly. *Molecular Genetics and Metabolism*, **2007**, 90, 97-111	3.7	58
97	SIX3 mutations with holoprosencephaly. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2577-83	2.5	22
96	Increased prevalence of ADHD in Turner syndrome with no evidence of imprinting effects. *Journal of Pediatric Psychology*, **2006**, 31, 945-55	3.2	91
95	A functional screen for sonic hedgehog regulatory elements across a 1 Mb interval identifies long-range ventral forebrain enhancers. *Development (Cambridge)*, **2006**, 133, 761-72	6.6	178
94	Reply to "Statin Drugs and Congenital Anomalies" by Gibb and Scialli. *American Journal of Medical Genetics, Part A*, **2005**, 135A, 232-234	2.5	1
93	A previously unidentified amino-terminal domain regulates transcriptional activity of wild-type and disease-associated human GLI2. *Human Molecular Genetics*, **2005**, 14, 2181-8	5.6	139
92	Attention deficit/hyperactivity disorder (ADHD): complex phenotype, simple genotype?. *Genetics in Medicine*, **2004**, 6, 1-15	8.1	51

91	Central nervous system and limb anomalies in case reports of first-trimester statin exposure. *New England Journal of Medicine*, **2004**, 350, 1579-82	59.2	165
90	FISH diagnosis of the common 57-kb deletion in CTNS causing cystinosis. *Human Genetics*, **2004**, 115, 510-4	6.3	30
89	Anomalies of the forebrain with radial limb defects: Garcia-Lurie-Steinfeld syndrome?. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2004**, 70, 537-44		10
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