Maximilian Muenke

List of Publications by Citations

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	Mutations in the human Sonic Hedgehog gene cause holoprosencephaly. *Nature Genetics*, **1996**, 14, 357-60	36.3	973
251	A common mutation in the fibroblast growth factor receptor 1 gene in Pfeiffer syndrome. *Nature Genetics*, **1994**, 8, 269-74	36.3	526
250	Holoprosencephaly due to mutations in ZIC2, a homologue of Drosophila odd-paired. *Nature Genetics*, **1998**, 20, 180-3	36.3	408
249	Mutations in the homeodomain of the human SIX3 gene cause holoprosencephaly. *Nature Genetics*, **1999**, 22, 196-8	36.3	360
248	Mutations in TGIF cause holoprosencephaly and link NODAL signalling to human neural axis determination. *Nature Genetics*, **2000**, 25, 205-8	36.3	337
247	Overexpression of an osteogenic morphogen in fibrodysplasia ossificans progressiva. *New England Journal of Medicine*, **1996**, 335, 555-61	59.2	330
246	Familial dementia caused by polymerization of mutant neuroserpin. *Nature*, **1999**, 401, 376-9	50.4	306
245	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, 285-91	36.3	296
244	Loss-of-function mutations in the EGF-CFC gene CFC1 are associated with human left-right laterality defects. *Nature Genetics*, **2000**, 26, 365-9	36.3	288
243	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, 13424-9	11.5	279
242	Identical mutations in three different fibroblast growth factor receptor genes in autosomal dominant craniosynostosis syndromes. *Nature Genetics*, **1996**, 14, 174-6	36.3	270
241	Multiple hits during early embryonic development: digenic diseases and holoprosencephaly. *American Journal of Human Genetics*, **2002**, 71, 1017-32	11	269
240	Fibroblast-growth-factor receptor mutations in human skeletal disorders. *Trends in Genetics*, **1995**, 11, 308-13	8.5	266
239	Genetics of ventral forebrain development and holoprosencephaly. *Current Opinion in Genetics and Development*, **2000**, 10, 262-9	4.9	221
238	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, 472-86	11	203
237	Mutations in PATCHED-1, the receptor for SONIC HEDGEHOG, are associated with holoprosencephaly. *Human Genetics*, **2002**, 110, 297-301	6.3	192
236	Cbfbeta interacts with Runx2 and has a critical role in bone development. *Nature Genetics*, **2002**, 32, 639-44	36.3	187

235	The molecular genetics of holoprosencephaly. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2010**, 154C, 52-61	3.1	186
234	Mutations in FGFR1 and FGFR2 cause familial and sporadic Pfeiffer syndrome. *Human Molecular Genetics*, **1995**, 4, 323-8	5.6	183
233	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
232	Mutations in holoprosencephaly. *Human Mutation*, **2000**, 16, 99-108	4.7	175
231	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
230	CFC1 mutations in patients with transposition of the great arteries and double-outlet right ventricle. *American Journal of Human Genetics*, **2002**, 70, 776-80	11	164
229	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	11	163
228	Holoprosencephaly in RSH/Smith-Lemli-Opitz syndrome: does abnormal cholesterol metabolism affect the function of Sonic Hedgehog?. *American Journal of Medical Genetics Part A*, **1996**, 66, 478-84		163
227	Adverse birth outcome among mothers with low serum cholesterol. *Pediatrics*, **2007**, 120, 723-33	7.4	156
226	Regulation of a remote Shh forebrain enhancer by the Six3 homeoprotein. *Nature Genetics*, **2008**, 40, 1348-53	36.3	153
225	Mechanistic and epidemiologic considerations in the evaluation of adverse birth outcomes following gestational exposure to statins. *American Journal of Medical Genetics Part A*, **2004**, 131, 287-98		140
224	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
223	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
222	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		138
221	Localization of DNA sequences required for human centromere function through an analysis of rearranged Y chromosomes. *Nature Genetics*, **1993**, 5, 368-75	36.3	138
220	Human developmental disorders and the Sonic hedgehog pathway. *Trends in Molecular Medicine*, **1998**, 4, 343-9		137
219	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
218	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

217	Association between conformational mutations in neuroserpin and onset and severity of dementia. *Lancet, The*, **2002**, 359, 2242-7	40	126
216	Novel mutation in sonic hedgehog in non-syndromic colobomatous microphthalmia. *American Journal of Medical Genetics Part A*, **2003**, 116A, 215-21		121
215	The human osmoregulatory Na+/myo-inositol cotransporter gene (SLC5A3): molecular cloning and localization to chromosome 21. *Genomics*, **1995**, 25, 507-13	4.3	116
214	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
213	Holoprosencephaly: from Homer to Hedgehog. *Clinical Genetics*, **1998**, 53, 155-63	4	109
212	Molecular mechanisms of holoprosencephaly. *Molecular Genetics and Metabolism*, **1999**, 68, 126-38	3.7	109
211	Craniosynostosis syndromes: from genes to premature fusion of skull bones. *Molecular Genetics and Metabolism*, **1999**, 68, 139-51	3.7	108
210	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
209	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
208	Opitz syndrome is genetically heterogeneous, with one locus on Xp22, and a second locus on 22q11.2. *Nature Genetics*, **1995**, 11, 459-61	36.3	96
207	. *Nature*, **1999**, 401, 376-379	50.4	92
206	Increased prevalence of ADHD in Turner syndrome with no evidence of imprinting effects. *Journal of Pediatric Psychology*, **2006**, 31, 945-55	3.2	91
205	A loss-of-function mutation in the CFC domain of TDGF1 is associated with human forebrain defects. *Human Genetics*, **2002**, 110, 422-8	6.3	86
204	Physical mapping of the holoprosencephaly critical region on chromosome 7q36. *Nature Genetics*, **1993**, 3, 247-51	36.3	85
203	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
202	Symptomatology of autism spectrum disorder in a population with neurofibromatosis type 1. *Developmental Medicine and Child Neurology*, **2013**, 55, 131-8	3.3	78
201	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, 714-6	3.6	78
200	Copy-number variations involving the IHH locus are associated with syndactyly and craniosynostosis. *American Journal of Human Genetics*, **2011**, 88, 70-5	11	76

199	Fibrodysplasia ossificans progressiva, a heritable disorder of severe heterotopic ossification, maps to human chromosome 4q27-31. *American Journal of Human Genetics*, **2000**, 66, 128-35	11	75
198	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
197	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
196	22q11.2 deletion syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2017**, 173, 879-888	2.5	67
195	Human enteric defensin genes: chromosomal map position and a model for possible evolutionary relationships. *Genomics*, **1996**, 31, 95-106	4.3	66
194	Opitz G/BBB syndrome: clinical comparisons of families linked to Xp22 and 22q, and a review of the literature. *American Journal of Medical Genetics Part A*, **1996**, 62, 305-17		66
193	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
192	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
191	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
190	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
189	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, 369-78		63
188	Midline and laterality defects: left and right meet in the middle. *BioEssays*, **2001**, 23, 888-900	4.1	62
187	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
186	Genotypic and phenotypic analysis of 396 individuals with mutations in Sonic Hedgehog. *Journal of Medical Genetics*, **2012**, 49, 473-9	5.8	60
185	The interplay of genetic and environmental factors in craniofacial morphogenesis: holoprosencephaly and the role of cholesterol. *Congenital Anomalies (discontinued)*, **2003**, 43, 1-21	1.1	60
184	Phenotype of the fibroblast growth factor receptor 2 Ser351Cys mutation: Pfeiffer syndrome type III. *American Journal of Medical Genetics Part A*, **1998**, 78, 356-60		58
183	Opitz G/BBB syndrome in Xp22: mutations in the MID1 gene cluster in the carboxy-terminal domain. *American Journal of Human Genetics*, **1998**, 63, 703-10	11	58
182	Functional analysis of mutations in TGIF associated with holoprosencephaly. *Molecular Genetics and Metabolism*, **2007**, 90, 97-111	3.7	58

181	How a Hedgehog might see holoprosencephaly. *Human Molecular Genetics*, **2003**, 12 Spec No 1, R15-25	5.6	58
180	Review: Genetics of attention deficit/hyperactivity disorder. *Journal of Pediatric Psychology*, **2008**, 33, 1085-99	3.2	57
179	Genetic approaches to understanding brain development: holoprosencephaly as a model. *Mental Retardation and Developmental Disabilities Research Reviews*, **2000**, 6, 15-21		57
178	Truncating loss-of-function mutations of DISP1 contribute to holoprosencephaly-like microform features in humans. *Human Genetics*, **2009**, 125, 393-400	6.3	56
177	Impact of genetics on the diagnosis and clinical management of syndromic craniosynostoses. *Childs Nervous System*, **2012**, 28, 1447-63	1.7	54
176	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
175	Molecular characterization of breakpoints in patients with holoprosencephaly and definition of the HPE2 critical region 2p21. *Human Molecular Genetics*, **1996**, 5, 223-9	5.6	54
174	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
173	Attention deficit/hyperactivity disorder (ADHD): complex phenotype, simple genotype?. *Genetics in Medicine*, **2004**, 6, 1-15	8.1	51
172	The topographic organization of repetitive DNA in the human nucleolus. *Genomics*, **1993**, 15, 123-32	4.3	49
171	Mutations in the human SIX3 gene in holoprosencephaly are loss of function. *Human Molecular Genetics*, **2008**, 17, 3919-28	5.6	48
170	Holoprosencephaly due to numeric chromosome abnormalities. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2010**, 154C, 146-8	3.1	47
169	Holoprosencephaly as a genetic model for normal craniofacial development. *Seminars in Developmental Biology*, **1994**, 5, 293-301		47
168	A common genetic network underlies substance use disorders and disruptive or externalizing disorders. *Human Genetics*, **2012**, 131, 917-29	6.3	46
167	Linkage of Pfeiffer syndrome to chromosome 8 centromere and evidence for genetic heterogeneity. *Human Molecular Genetics*, **1994**, 3, 2153-8	5.6	46
166	Down syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2017**, 173, 42-53	2.5	45
165	Pathogenic mutations in GLI2 cause a specific phenotype that is distinct from holoprosencephaly. *Journal of Medical Genetics*, **2014**, 51, 413-8	5.8	45
164	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

163	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
162	Noonan syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2323-2334	2.3	43
161	Holoprosencephaly: a guide to diagnosis and clinical management. *Indian Pediatrics*, **2011**, 48, 457-66	1.2	41
160	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, 548-56		41
159	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
158	Identification of novel mutations in SHH and ZIC2 in a South American (ECLAMC) population with holoprosencephaly. *Human Genetics*, **2001**, 109, 1-6	6.3	40
157	Attention-deficit/hyperactivity disorder and comorbidities in 18 Paisa Colombian multigenerational families. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2004**, 43, 1506-15	7.2	39
156	Holoprosencephaly: recommendations for diagnosis and management. *Current Opinion in Pediatrics* , **2010**, 22, 687-95	3.2	38
155	Syndromes associated with holoprosencephaly. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 229-237	3.1	37
154	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
153	SONIC HEDGEHOG mutations causing human holoprosencephaly impair neural patterning activity. *Human Genetics*, **2003**, 113, 170-7	6.3	37
152	Holoprosencephaly in the genomics era. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 165-174	3.1	36
151	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
150	An Ultraconserved Brain-Specific Enhancer Within ADGRL3 (LPHN3) Underpins Attention-Deficit/Hyperactivity Disorder Susceptibility. *Biological Psychiatry*, **2016**, 80, 943-954	7.9	35
149	Abnormal sterol metabolism in holoprosencephaly. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2010**, 154C, 102-8	3.1	33
148	Structure of the human gene encoding the associated microfibrillar protein (MFAP1) and localization to chromosome 15q15-q21. *Genomics*, **1994**, 23, 443-9	4.3	33
147	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
146	ADHD latent class clusters: DSM-IV subtypes and comorbidity. *Psychiatry Research*, **2009**, 170, 192-8	9.9	32

145	Holoprosencephaly: molecular study of a California population. *American Journal of Medical Genetics Part A*, **2000**, 90, 315-9		32
144	Noonan syndrome. *American Family Physician*, **2014**, 89, 37-43	1.3	32
143	Williams-Beuren syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1128-1136	2.5	31
142	Cohesin complex-associated holoprosencephaly. *Brain*, **2019**, 142, 2631-2643	11.2	31
141	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
140	FISH diagnosis of the common 57-kb deletion in CTNS causing cystinosis. *Human Genetics*, **2004**, 115, 510-4	6.3	30
139	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
138	Muenke syndrome: An international multicenter natural history study. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 918-29	2.5	29
137	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
136	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
135	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
134	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
133	Reciprocal mouse and human limb phenotypes caused by gain- and loss-of-function mutations affecting Lmbr1. *Genetics*, **2001**, 159, 715-26	4	27
132	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
131	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
130	Toward a better understanding of ADHD: LPHN3 gene variants and the susceptibility to develop ADHD. *ADHD Attention Deficit and Hyperactivity Disorders*, **2010**, 2, 139-47	3.1	26
129	Clinical characteristics of patients with unicoronal synostosis and mutations of fibroblast growth factor receptor 3: a preliminary report. *Plastic and Reconstructive Surgery*, **2001**, 108, 1849-54	2.7	26
128	Additional EFNB1 mutations in craniofrontonasal syndrome. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2008-12	2.5	25

127	Cornelia de Lange syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2019**, 179, 150-158	2.5	25
126	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
125	An electronic atlas of human malformation syndromes in diverse populations. *Genetics in Medicine*, **2016**, 18, 1085-1087	8.1	23
124	BOC is a modifier gene in holoprosencephaly. *Human Mutation*, **2017**, 38, 1464-1470	4.7	23
123	Palatal and oral manifestations of Muenke syndrome (FGFR3-related craniosynostosis). *Journal of Craniofacial Surgery*, **2012**, 23, 664-8	1.2	23
122	Fibrodysplasia ossificans progressiva in two half-sisters: evidence for maternal mosaicism. *American Journal of Medical Genetics Part A*, **1996**, 61, 320-4		23
121	VACTERL Association With High Prenatal Lead Exposure: Similarities to Animal Models of Lead Teratogenicity. *Pediatrics*, **1991**, 87, 390-392	7.4	23
120	A CCR4-NOT Transcription Complex, Subunit 1, CNOT1, Variant Associated with Holoprosencephaly. *American Journal of Human Genetics*, **2019**, 104, 990-993	11	22
119	SIX3 mutations with holoprosencephaly. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2577-83	2.5	22
118	Mutational analysis of the Sonic Hedgehog gene in 220 newborns with oral clefts in a South American (ECLAMC) population. *American Journal of Medical Genetics Part A*, **2002**, 108, 12-5		22
117	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
116	ARP3beta, the gene encoding a new human actin-related protein, is alternatively spliced and predominantly expressed in brain neuronal cells. *FEBS Journal*, **2000**, 267, 2921-8		20
115	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
114	Phenotype profile of a genetic mouse model for Muenke syndrome. *Childs Nervous System*, **2012**, 28, 1483-93	1.7	19
113	Craniosynostosis, Philadelphia type: a new autosomal dominant syndrome with sagittal craniosynostosis and syndactyly of the fingers and toes. *American Journal of Medical Genetics Part A* , **1996**, 62, 184-91		19
112	Molecular testing in holoprosencephaly. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 187-193	3.1	19
111	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
110	Human germline hedgehog pathway mutations predispose to fatty liver. *Journal of Hepatology*, **2017**, 67, 809-817	13.4	18

109	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
108	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
107	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
106	Expanding the phenotypic expression of Sonic Hedgehog mutations beyond holoprosencephaly. *Journal of Craniofacial Surgery*, **2015**, 26, 3-5	1.2	15
105	Epilepsy in Muenke syndrome: FGFR3-related craniosynostosis. *Pediatric Neurology*, **2012**, 47, 355-61	2.9	15
104	Hearing loss in syndromic craniosynostoses: otologic manifestations and clinical findings. *International Journal of Pediatric Otorhinolaryngology*, **2014**, 78, 2037-47	1.7	14
103	Clinical utility gene card for: Holoprosencephaly. *European Journal of Human Genetics*, **2011**, 19, preceeding 118-20	5.3	14
102	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
101	Rearrangement in the PITX2 and MIPOL1 genes in a patient with a t(4;14) chromosome. *European Journal of Human Genetics*, **2003**, 11, 315-24	5.3	14
100	Sub-band deletion of 7q36.3 in a patient with ring chromosome 7: association with holoprosencephaly. *American Journal of Medical Genetics Part A*, **1996**, 65, 113-6		14
99	When to suspect a genetic syndrome. *American Family Physician*, **2012**, 86, 826-33	1.3	14
98	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
97	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
96	Loss of function in is associated with tetralogy of Fallot and septal defects. *Journal of Medical Genetics*, **2017**, 54, 825-829	5.8	13
95	ADGRL3 (LPHN3) variants predict substance use disorder. *Translational Psychiatry*, **2019**, 9, 42	8.6	13
94	Prenatal exposure to pesticides and risk for holoprosencephaly: a case-control study. *Environmental Health*, **2020**, 19, 65	6	13
93	In-depth investigations of adolescents and adults with holoprosencephaly identify unique characteristics. *Genetics in Medicine*, **2018**, 20, 14-23	8.1	13
92	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

91	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
90	Novel SNP at the common primer site of exon IIIa of FGFR2 gene causes error in molecular diagnosis of craniosynostosis syndrome. *American Journal of Medical Genetics Part A*, **2001**, 102, 282-5		13
89	Loss-of-function mutations in FGF8 can be independent risk factors for holoprosencephaly. *Human Molecular Genetics*, **2018**, 27, 1989-1998	5.6	12
88	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
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