# Maximilian Muenke

List of Publications by Year
in descending order



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|---|---|---|---|
| 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 | Mutations in the human Sonic Hedgehog gene cause holoprosencephaly. Nature Genetics, 1996, 14, 357-360. | 9.4 | 1,075 |
| 2 | A common mutation in the fibroblast growth factor receptor 1 gene in Pfeiffer syndrome. Nature Genetics, 1994, 8, 269-274. | 9.4 | 615 |
| 3 | Holoprosencephaly due to mutations in ZIC2, a homologue of Drosophila odd-paired. Nature Genetics, 1998, 20, 180-183. | 9.4 | 448 |
| 4 | Mutations in the homeodomain of the human SIX3 gene cause holoprosencephaly. Nature Genetics, 1999, 22, 196-198. | 9.4 | 398 |
| 5 | Mutations in TGIF cause holoprosencephaly and link NODAL signalling to human neural axis determination. Nature Genetics, 2000, 25, 205-208. | 9.4 | 368 |
| 6 | Overexpression of an Osteogenic Morphogen in Fibrodysplasia Ossificans Progressiva. New England Journal of Medicine, 1996, 335, 555-561. | 13.9 | 364 |
| 7 | Familial dementia caused by polymerization of mutant neuroserpin. Nature, 1999, 401, 376-379. | 13.7 | 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. | 9.4 | 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. | 9.4 | 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. | 3.3 | 313 |
| 11 | Identical mutations in three different fibroblast growth factor receptor genes in autosomal dominant craniosynostosis syndromes. Nature Genetics, 1996, 14, 174-176. | 9.4 | 306 |
| 12 | Multiple Hits during Early Embryonic Development: Digenic Diseases and Holoprosencephaly. American Journal of Human Genetics, 2002, 71, 1017-1032. | 2.6 | 293 |
| 13 | Fibroblast-growth-factor receptor mutations in human skeletal disorders. Trends in Genetics, 1995, 11, 308-313. | 2.9 | 292 |
| 14 | Genetics of ventral forebrain development and holoprosencephaly. Current Opinion in Genetics and Development, 2000, 10, 262-269. | 1.5 | 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. | 2.6 | 238 |
| 16 | Mutations in FGFR1 and FGFR2 cause familial and sporadic Pfeiffer syndrome. Human Molecular Genetics, 1995, 4, 323-328. | 1.4 | 221 |
| 17 | Mutations in PATCHED-1, the receptor for SONIC HEDGEHOG, are associated with holoprosencephaly. Human Genetics, 2002, 110, 297-301. | 1.8 | 220 |
| 18 | The molecular genetics of holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 52-61. | 0.7 | 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. | 13.9 | 210 |
| 20 | Cbfî² interacts with Runx2 and has a critical role in bone development. Nature Genetics, 2002, 32, 639-644. | 9.4 | 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. | 3.1 | 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. | 1.2 | 198 |
| 23 | Mutations in holoprosencephaly. Human Mutation, 2000, 16, 99-108. | 1.1 | 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. | 2.6 | 192 |
| 26 | Adverse Birth Outcome Among Mothers With Low Serum Cholesterol. Pediatrics, 2007, 120, 723-733. | 1.0 | 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. | 2.6 | 182 |
| 28 | Regulation of a remote Shh forebrain enhancer by the Six3 homeoprotein. Nature Genetics, 2008, 40, 1348-1353. | 9.4 | 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. | 1.1 | 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. | 1.4 | 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. | 2.6 | 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. | 9.4 | 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. | 0.7 | 148 |

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|---|---|---|---|
| 37 | Association between conformational mutations in neuroserpin and onset and severity of dementia. Lancet, The, 2002, 359, 2242-2247. | 6.3 | 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. | 0.7 | 139 |
| 39 | Novel mutation in sonic hedgehog in non-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. | 1.0 | 130 |
| 41 | Craniosynostosis Syndromes: From Genes to Premature Fusion of Skull Bones. Molecular Genetics and Metabolism, 1999, 68, 139-151. | 0.5 | 123 |
| 42 | Molecular Mechanisms of Holoprosencephaly. Molecular Genetics and Metabolism, 1999, 68, 126-138. | 0.5 | 121 |
| 43 | The human osmoregulatory Na+/myo-inositol cotransporter gene (SLC5A3): molecular cloning and localization to chromosome 21. Genomics, 1995, 25, 507-513. | 1.3 | 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. | 2.6 | 116 |
| 45 | Title is missing!. Nature, 1999, 401, 376-379. | 13.7 | 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. | 2.6 | 111 |
| 47 | Increased Prevalence of ADHD in Turner Syndrome with No Evidence of Imprinting Effects. Journal of Pediatric Psychology, 2006, 31, 945-955. | 1.1 | 110 |
| 48 | Symptomatology of autism spectrum disorder in a population with neurofibromatosis type 1. Developmental Medicine and Child Neurology, 2013, 55, 131-138. | 1.1 | 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. | 9.4 | 103 |
| 50 | 22q11.2 deletion syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 879-888. | 0.7 | 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. | 0.7 | 99 |
| 52 | Physical mapping of the holoprosencephaly critical region on chromosome 7q36. Nature Genetics, 1993, 3, 247-251. | 9.4 | 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. | 0.9 | 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. | 1.8 | 93 |

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|---|---|---|---|
| 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. | 3.3 | 91 |
| 56 | 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 |
| 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. | 2.6 | 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. | 0.7 | 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. | 1.1 | 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. | 1.5 | 75 |
| 63 | Down syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 42-53. | 0.7 | 75 |
| 64 | Human Enteric Defensin Genes: Chromosomal Map Position and a Model for Possible Evolutionary Relationships. Genomics, 1996, 31, 95-106. | 1.3 | 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. | 0.3 | 73 |
| 66 | Phenotype of the fibroblast growth factor receptor 2 Ser351Cys mutation: Pfeiffer syndrome type III. , 1998, 78, 356-360. | | 72 |
| 67 | Midline and laterality defects: Left and right meet in the middle. BioEssays, 2001, 23, 888-900. | 1.2 | 70 |
| 68 | Impact of genetics on the diagnosis and clinical management of syndromic craniosynostoses. Child's Nervous System, 2012, 28, 1447-1463. | 0.6 | 70 |
| 69 | The 2019 US medical genetics workforce: a focus on clinical genetics. Genetics in Medicine, 2021, 23, 1458-1464. | 1.1 | 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. | 0.7 | 69 |
| 72 | Review: Genetics of Attention Deficit/Hyperactivity Disorder. Journal of Pediatric Psychology, 2008, 33, 1085-1099. | 1.1 | 69 |

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|---|---|---|---|
| 73 | Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334. | 0.7 | 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. | 0.3 | 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. | 0.5 | 67 |
| 76 | Genotypic and phenotypic analysis of 396 individuals with mutations inSonic Hedgehog. Journal of Medical Genetics, 2012, 49, 473-479. | 1.5 | 67 |
| 77 | Attention deficit/hyperactivity disorder (ADHD): Complex phenotype, simple genotype?. Genetics in Medicine, 2004, 6, 1-15. | 1.1 | 65 |
| 78 | How a Hedgehog might see holoprosencephaly. Human Molecular Genetics, 2003, 12, 15R-25. | 1.4 | 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. | 2.6 | 63 |
| 80 | Functional analysis of mutations in TGIF associated with holoprosencephaly. Molecular Genetics and Metabolism, 2007, 90, 97-111. | 0.5 | 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. | 0.7 | 62 |
| 82 | Truncating loss-of-function mutations of DISP1 contribute to holoprosencephaly-like microform features in humans. Human Genetics, 2009, 125, 393-400. | 1.8 | 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. | 1.4 | 60 |
| 84 | A common genetic network underlies substance use disorders and disruptive or externalizing disorders. Human Genetics, 2012, 131, 917-929. | 1.8 | 60 |
| 85 | REPORTS. Human Molecular Genetics, 1994, 3, 2153-2158. | 1.4 | 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. | 1.4 | 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. | 1.1 | 56 |
| 89 | Holoprosencephaly due to numeric chromosome abnormalities. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 146-148. | 0.7 | 56 |
| 90 | Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136. | 0.7 | 55 |

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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 | 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 |
| 93 | The Topographic Organization of Repetitive DNA in the Human Nucleolus. Genomics, 1993, 15, 123-132. | 1.3 | 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. | 0.3 | 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. | 1.8 | 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. | 1.8 | 51 |
| 97 | Holoprosencephaly: A guide to diagnosis and clinical management. Indian Pediatrics, 2011, 48, 457-466. | 0.2 | 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. | 1.1 | 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. | 0.5 | 48 |
| 100 | An Ultraconserved Brain-Specific Enhancer Within ADGRL3 (LPHN3) Underpins Attention-Deficit/Hyperactivity Disorder Susceptibility. Biological Psychiatry, 2016, 80, 943-954. | 0.7 | 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. | 1.8 | 46 |
| 104 | Dominant-negative kinase domain mutations inFGFR1can explain the clinical severity of Hartsfield syndrome. Human Molecular Genetics, 2016, 25, 1912-1922. | 1.4 | 46 |
| 105 | Syndromes associated with holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 229-237. | 0.7 | 45 |
| 106 | Holoprosencephaly: recommendations for diagnosis and management. Current Opinion in Pediatrics, 2010, 22, 687-695. | 1.0 | 44 |
| 107 | An electronic atlas of human malformation syndromes in diverse populations. Genetics in Medicine, 2016, 18, 1085-1087. | 1.1 | 44 |
| 108 | Holoprosencephaly: Molecular study of a California Population. , 2000, 90, 315-319. | | 43 |

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|---|---|---|---|
| 109 | Cohesin complex-associated holoprosencephaly. Brain, 2019, 142, 2631-2643. | 3.7 | 43 |
| 110 | ADHD latent class clusters: DSM-IV subtypes and comorbidity. Psychiatry Research, 2009, 170, 192-198. | 1.7 | 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. | 0.7 | 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. | 1.5 | 40 |
| 113 | Holoprosencephaly in the genomics era. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 165-174. | 0.7 | 40 |
| 114 | Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158. | 0.7 | 40 |
| 115 | SONIC HEDGEHOG mutations causing human holoprosencephaly impair neural patterning activity. Human Genetics, 2003, 113, 170-177. | 1.8 | 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. | 0.7 | 38 |
| 117 | Muenke syndrome: An international multicenter natural history study. American Journal of Medical Genetics, Part A, 2016, 170, 918-929. | 0.7 | 37 |
| 118 | FISH diagnosis of the common 57-kb deletion in CTNS causing cystinosis. Human Genetics, 2004, 115, 510-514. | 1.8 | 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. | 0.7 | 36 |
| 121 | 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 |
| 122 | Abnormal sterol metabolism in holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 102-108. | 0.7 | 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. | 0.7 | 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. | 0.3 | 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. | 0.6 | 35 |
| 126 | Additional EFNB1 mutations in craniofrontonasal syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 2008-2012. | 0.7 | 34 |

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| 127 | Structure of the Human Gene Encoding the Associated Microfibrillar Protein (MFAP1) and Localization to Chromosome 15q15-q21. Genomics, 1994, 23, 443-449. | 1.3 | 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. | 1.4 | 33 |
| 129 | Reciprocal Mouse and Human Limb Phenotypes Caused by Gain- and Loss-of-Function Mutations Affecting Lmbr1. Genetics, 2001, 159, 715-726. | 1.2 | 32 |
| 130 | A call for global action for rare diseases in Africa. Nature Genetics, 2020, 52, 21-26. | 9.4 | 31 |
| 131 | The genetics of addiction. Human Genetics, 2012, 131, 773-777. | 1.8 | 30 |
| 132 | A CCR4-NOT Transcription Complex, Subunit 1, CNOT1, Variant Associated with Holoprosencephaly. American Journal of Human Genetics, 2019, 104, 990-993. | 2.6 | 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. | 2.6 | 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. | 0.7 | 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. | 1.1 | 29 |
| 137 | ADGRL3 (LPHN3) variants predict substance use disorder. Translational Psychiatry, 2019, 9, 42. | 2.4 | 29 |
| 138 | Fibrodysplasia ossificans progressiva in two half-sisters: Evidence for maternal mosaicism. , 1996, 61, 320-324. | | 28 |
| 139 | Mutational analysis of theSonic Hedgehoggene 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. | 1.0 | 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. | 1.1 | 27 |
| 142 | Loss of function inROBO1is associated with tetralogy of Fallot and septal defects. Journal of Medical Genetics, 2017, 54, 825-829. | 1.5 | 27 |
| 143 | BOC is a modifier gene in holoprosencephaly. Human Mutation, 2017, 38, 1464-1470. | 1.1 | 27 |
| 144 | Hearing loss in syndromic craniosynostoses: Otologic manifestations and clinical findings. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 2037-2047. | 0.4 | 26 |

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|---|---|---|---|
| 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. | 1.8 | 26 |
| 146 | SIX3 mutations with holoprosencephaly. American Journal of Medical Genetics, Part A, 2006, 140A, 2577-2583. | 0.7 | 25 |
| 147 | Palatal and Oral Manifestations of Muenke Syndrome (FGFR3-Related Craniosynostosis). Journal of Craniofacial Surgery, 2012, 23, 664-668. | 0.3 | 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. | 1.1 | 25 |
| 149 | Human germline hedgehog pathway mutations predispose to fatty liver. Journal of Hepatology, 2017, 67, 809-817. | 1.8 | 24 |
| 150 | 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 |
| 151 | Phenotype delineation of ZNF462 related syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2075-2082. | 0.7 | 23 |
| 152 | Molecular testing in holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 187-193. | 0.7 | 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. | 0.6 | 20 |
| 156 | Epilepsy in Muenke Syndrome: FGFR3-Related Craniosynostosis. Pediatric Neurology, 2012, 47, 355-361. | 1.0 | 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 |
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