

Ethylin Wang Jobs

List of Publications by Year
in descending order

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179
papers

14,863
citations

31949

53
h-index

20343

116
g-index

183
all docs

183
docs citations

183
times ranked

14747
citing authors

#	ARTICLE	IF	CITATIONS
1	Exome sequencing identifies the cause of a mendelian disorder. <i>Nature Genetics</i> , 2010, 42, 30-35.	9.4	1,813
2	A mutation in the homeodomain of the human <i>MSX2</i> gene in a family affected with autosomal dominant craniosynostosis. <i>Cell</i> , 1993, 75, 443-450.	13.5	658
3	Human dopamine transporter gene (<i>DAT1</i>) maps to chromosome 5p15.3 and displays a VNTR. <i>Genomics</i> , 1992, 14, 1104-1106.	1.3	655
4	Mutations in <i>TWIST</i> , a basic helix-loop-helix transcription factor, in Saethre-Chotzen syndrome. <i>Nature Genetics</i> , 1997, 15, 36-41.	9.4	628
5	Connexin 43 (<i>GJA1</i>) Mutations Cause the Pleiotropic Phenotype of Oculodentodigital Dysplasia. <i>American Journal of Human Genetics</i> , 2003, 72, 408-418.	2.6	585
6	A genome-wide association study of cleft lip with and without cleft palate identifies risk variants near <i>MAFB</i> and <i>ABCA4</i> . <i>Nature Genetics</i> , 2010, 42, 525-529.	9.4	518
7	Mutant P450 oxidoreductase causes disordered steroidogenesis with and without Antley-Bixler syndrome. <i>Nature Genetics</i> , 2004, 36, 228-230.	9.4	462
8	Jackson-Weiss and Crouzon syndromes are allelic with mutations in fibroblast growth factor receptor 2. <i>Nature Genetics</i> , 1994, 8, 275-279.	9.4	458
9	Fibroblast growth factor receptor 3 (<i>FGFR3</i>) transmembrane mutation in Crouzon syndrome with acanthosis nigricans. <i>Nature Genetics</i> , 1995, 11, 462-464.	9.4	378
10	Advancing age has differential effects on DNA damage, chromatin integrity, gene mutations, and aneuploidies in sperm. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006, 103, 9601-9606.	3.3	363
11	Missense Mutations in <i>GJB2</i> Encoding Connexin-26 Cause the Ectodermal Dysplasia Keratitis-Ichthyosis-Deafness Syndrome. <i>American Journal of Human Genetics</i> , 2002, 70, 1341-1348.	2.6	345
12	Roberts syndrome is caused by mutations in <i>ESCO2</i> , a human homolog of yeast <i>ECO1</i> that is essential for the establishment of sister chromatid cohesion. <i>Nature Genetics</i> , 2005, 37, 468-470.	9.4	334
13	Diversity and Function of Mutations in P450 Oxidoreductase in Patients with Antley-Bixler Syndrome and Disordered Steroidogenesis. <i>American Journal of Human Genetics</i> , 2005, 76, 729-749.	2.6	321
14	<i>GJA1</i> mutations, variants, and connexin 43 dysfunction as it relates to the oculodentodigital dysplasia phenotype. <i>Human Mutation</i> , 2009, 30, 724-733.	1.1	240
15	De Novo Alu-Element Insertions in <i>FGFR2</i> Identify a Distinct Pathological Basis for Apert Syndrome. <i>American Journal of Human Genetics</i> , 1999, 64, 446-461.	2.6	225
16	Genetic Heterogeneity of Saethre-Chotzen Syndrome, Due to <i>TWIST</i> and <i>FGFR</i> Mutations. <i>American Journal of Human Genetics</i> , 1998, 62, 1370-1380.	2.6	202
17	Paternal Origin of <i>FGFR2</i> Mutations in Sporadic Cases of Crouzon Syndrome and Pfeiffer Syndrome. <i>American Journal of Human Genetics</i> , 2000, 66, 768-777.	2.6	191
18	Haploinsufficiency of <i>SF3B4</i> , a Component of the Pre-mRNA Spliceosomal Complex, Causes Nager Syndrome. <i>American Journal of Human Genetics</i> , 2012, 90, 925-933.	2.6	188

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19	Fibroblast growth factor receptor 2 mutations in Beare's Stevenson cutis gyrata syndrome. <i>Nature Genetics</i> , 1996, 13, 492-494.	9.4	181
20	Abnormalities in cartilage and bone development in the Apert syndrome FGFR2+/S252W mouse. <i>Development (Cambridge)</i> , 2005, 132, 3537-3548.	1.2	172
21	The Paternal-Age Effect in Apert Syndrome Is Due, in Part, to the Increased Frequency of Mutations in Sperm. <i>American Journal of Human Genetics</i> , 2003, 73, 939-947.	2.6	164
22	Evidence for gene-environment interaction in a genome wide study of nonsyndromic cleft palate. <i>Genetic Epidemiology</i> , 2011, 35, n/a-n/a.	0.6	145
23	Receptor Tyrosine Kinases Activate Canonical WNT/ β -Catenin Signaling via MAP Kinase/LRP6 Pathway and Direct β -Catenin Phosphorylation. <i>PLoS ONE</i> , 2012, 7, e35826.	1.1	142
24	A genome-wide association study identifies susceptibility loci for nonsyndromic sagittal craniosynostosis near BMP2 and within BBS9. <i>Nature Genetics</i> , 2012, 44, 1360-1364.	9.4	120
25	A novel syndrome caused by the E410K amino acid substitution in the neuronal β -tubulin isotype 3. <i>Brain</i> , 2013, 136, 522-535.	3.7	112
26	The molecular mechanism underlying Roberts syndrome involves loss of ESCO2 acetyltransferase activity. <i>Human Molecular Genetics</i> , 2008, 17, 2172-2180.	1.4	108
27	Association between IRF6 and nonsyndromic cleft lip with or without cleft palate in four populations. <i>Genetics in Medicine</i> , 2007, 9, 219-227.	1.1	107
28	Characterization of the Nucleolar Gene Product, Treacle, in Treacher Collins Syndrome. <i>Molecular Biology of the Cell</i> , 2000, 11, 3061-3071.	0.9	105
29	Functional Characterization of Connexin43 Mutations Found in Patients With Oculodentodigital Dysplasia. <i>Circulation Research</i> , 2005, 96, e83-91.	2.0	104
30	Cytogenomic identification and long-read single molecule real-time (SMRT) sequencing of a Bardet-Biedl Syndrome 9 (BBS9) deletion. <i>Npj Genomic Medicine</i> , 2018, 3, 3.	1.7	97
31	Mutations in HOXD13 Underlie Syndactyly Type V and a Novel Brachydactyly-Syndactyly Syndrome. <i>American Journal of Human Genetics</i> , 2007, 80, 361-371.	2.6	94
32	Oculodentodigital dysplasia connexin43 mutations result in non-functional connexin hemichannels and gap junctions in C6 glioma cells. <i>Journal of Cell Science</i> , 2006, 119, 532-541.	1.2	91
33	Increased risk for developmental delay in Saethre-Chotzen syndrome is associated with TWIST deletions: an improved strategy for TWIST mutation screening. <i>Human Genetics</i> , 2003, 114, 68-76.	1.8	83
34	Willingness to participate in genomics research and desire for personal results among underrepresented minority patients: a structured interview study. <i>Journal of Community Genetics</i> , 2013, 4, 469-482.	0.5	82
35	Effect on splicing of a silent FGFR2 mutation in Crouzon syndrome. <i>Nature Genetics</i> , 1995, 9, 232-233.	9.4	80
36	Reasons for participating and genetic information needs among racially and ethnically diverse biobank participants: a focus group study. <i>Journal of Community Genetics</i> , 2011, 2, 153-163.	0.5	75

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37	A Unique Point Mutation in the PMP22 Gene Is Associated with Charcot-Marie-Tooth Disease and Deafness. <i>American Journal of Human Genetics</i> , 1999, 64, 1580-1593.	2.6	74
38	Mapping the Treacher Collins syndrome locus to 5q31.3â†’q33.3. <i>Genomics</i> , 1991, 11, 193-198.	1.3	73
39	The Pleiotropic Effects of Fibroblast Growth Factor Receptors in Mammalian Development.. <i>Cell Structure and Function</i> , 2000, 25, 85-96.	0.5	72
40	HOXB1 Founder Mutation in Humans Recapitulates the Phenotype of Hoxb1 Mice. <i>American Journal of Human Genetics</i> , 2012, 91, 171-179.	2.6	72
41	Closing the Gap: Genetic and Genomic Continuum from Syndromic to Nonsyndromic Craniosynostoses. <i>Current Genetic Medicine Reports</i> , 2014, 2, 135-145.	1.9	72
42	A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome. <i>Nature Communications</i> , 2017, 8, 16077.	5.8	72
43	Activation of p38 MAPK pathway in the skull abnormalities of Apert syndrome Fgfr2+P253R mice. <i>BMC Developmental Biology</i> , 2010, 10, 22.	2.1	70
44	Genetic basis of potential therapeutic strategies for craniosynostosis. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 3007-3015.	0.7	67
45	Evidence of geneâ€“environment interaction for the IRF6 gene and maternal multivitamin supplementation in controlling the risk of cleft lip with/without cleft palate. <i>Human Genetics</i> , 2010, 128, 401-410.	1.8	65
46	Phenotypic variability in 49 cases of ESCO2 mutations, including novel missense and codon deletion in the acetyltransferase domain, correlates with ESCO2 expression and establishes the clinical criteria for Roberts syndrome. <i>Journal of Medical Genetics</i> , 2010, 47, 30-37.	1.5	65
47	The FaceBase Consortium: A comprehensive resource for craniofacial researchers. <i>Development (Cambridge)</i> , 2016, 143, 2677-88.	1.2	62
48	Toward understanding the pathogenesis of craniosynostosis through clinical and molecular correlates. <i>Clinical Genetics</i> , 1998, 53, 79-86.	1.0	60
49	Beyond the closed suture in apert syndrome mouse models: Evidence of primary effects of FGFR2 signaling on facial shape at birth. <i>Developmental Dynamics</i> , 2010, 239, 3058-3071.	0.8	60
50	Translocations Disrupting PHF21A in the Potocki-Shaffer-Syndrome Region Are Associated with Intellectual Disability and Craniofacial Anomalies. <i>American Journal of Human Genetics</i> , 2012, 91, 56-72.	2.6	59
51	Syndrome of coronal craniosynostosis, Klippel-Feil anomaly, and Sprengel shoulder with and without Pro250Arg mutation in the FGFR3 gene. <i>American Journal of Medical Genetics Part A</i> , 2001, 104, 112-119.	2.4	58
52	<i>OTX2</i> mutations contribute to the otocephaly-dysgnathia complex. <i>Journal of Medical Genetics</i> , 2012, 49, 373-379.	1.5	58
53	Studies of mitotic and centromeric abnormalities in Roberts syndrome: Implications for a defect in the mitotic mechanism. <i>Chromosoma</i> , 1991, 100, 251-261.	1.0	57
54	The CEPH Consortium Linkage Map of Human Chromosome 13. <i>Genomics</i> , 1993, 16, 486-496.	1.3	55

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55	Human and Mouse Chromosomal Mapping of the Myeloid Cell Leukemia-1 Gene: MCL1 Maps to Human Chromosome 1q21, a Region That Is Frequently Altered in Preneoplastic and Neoplastic Disease. <i>Genomics</i> , 1994, 23, 457-463.	1.3	55
56	Analysis of candidate genes on chromosome 2 in oral cleft case-parent trios from three populations. <i>Human Genetics</i> , 2006, 120, 501-518.	1.8	54
57	The Ups and Downs of Mutation Frequencies during Aging Can Account for the Apert Syndrome Paternal Age Effect. <i>PLoS Genetics</i> , 2009, 5, e1000558.	1.5	54
58	Association of a new chromosomal deletion [del(1)(q32q42)] with diaphragmatic hernia: assignment of a human ferritin gene. <i>Human Genetics</i> , 1988, 78, 267-270.	1.8	52
59	Genomic Organization, Expression, and Chromosome Location of the Human SNAIL Gene (SNAI1) and a Related Processed Pseudogene (SNAI1P). <i>Genomics</i> , 1999, 62, 42-49.	1.3	52
60	Saethre-Chotzen syndrome with familial translocation at chromosome 7p22. <i>American Journal of Medical Genetics Part A</i> , 1993, 47, 637-639.	2.4	51
61	FGF/FGFR Signaling Coordinates Skull Development by Modulating Magnitude of Morphological Integration: Evidence from Apert Syndrome Mouse Models. <i>PLoS ONE</i> , 2011, 6, e26425.	1.1	51
62	Tissue-specific responses to aberrant FGF signaling in complex head phenotypes. <i>Developmental Dynamics</i> , 2013, 242, 80-94.	0.8	51
63	Ten-year experience of more than 35,000 orofacial clefts in Africa. <i>BMC Pediatrics</i> , 2015, 15, 8.	0.7	51
64	Development and preliminary evaluation of an online educational video about whole-genome sequencing for research participants, patients, and the general public. <i>Genetics in Medicine</i> , 2016, 18, 501-512.	1.1	51
65	Genetic Heterogeneity among Craniosynostosis Syndromes: Mapping the Saethre-Chotzen Syndrome Locus between D7S513 and D7S516 and Exclusion of Jackson-Weiss and Crouzon Syndrome Loci from 7p. <i>Genomics</i> , 1994, 19, 115-119.	1.3	50
66	Two craniosynostotic patients with 11q deletions, and review of 48 cases. <i>American Journal of Medical Genetics Part A</i> , 1995, 59, 193-198.	2.4	48
67	Ocular anterior chamber dysgenesis in craniosynostosis syndromes with a fibroblast growth factor receptor 2 mutation. <i>American Journal of Medical Genetics Part A</i> , 1999, 85, 160-170.	2.4	48
68	Maternal transmission effects of the PAX genes among cleft case-parent trios from four populations. <i>European Journal of Human Genetics</i> , 2009, 17, 831-839.	1.4	48
69	Dear Old Dad. <i>Science of Aging Knowledge Environment: SAGE KE</i> , 2004, 2004, 1re-1.	0.9	48
70	Associations between periconceptual alcohol consumption and craniosynostosis, omphalocele, and gastroschisis. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2011, 91, 623-630.	1.6	47
71	p38 Inhibition ameliorates skin and skull abnormalities in Fgfr2 Beare-Stevenson mice. <i>Journal of Clinical Investigation</i> , 2012, 122, 2153-2164.	3.9	47
72	Human PRRX1 and PRRX2 genes: cloning, expression, genomic localization, and exclusion as disease genes for Nager syndrome. <i>Mammalian Genome</i> , 2000, 11, 1000-1005.	1.0	44

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73	Evidence that TGFA influences risk to cleft lip with/without cleft palate through unconventional genetic mechanisms. <i>Human Genetics</i> , 2009, 126, 385-394.	1.8	44
74	Structure of the human spermidine/spermine N1-acetyltransferase gene. <i>Biochemical and Biophysical Research Communications</i> , 1992, 187, 1493-1502.	1.0	43
75	Autosomal Dominant Postaxial Polydactyly, Nail Dystrophy, and Dental Abnormalities Map to Chromosome 4p16, in the Region Containing the Ellis-van Creveld Syndrome Locus. <i>American Journal of Human Genetics</i> , 1997, 61, 1405-1412.	2.6	43
76	Human SLUG Gene Organization, Expression, and Chromosome Map Location on 8q. <i>Genomics</i> , 1998, 51, 468-471.	1.3	43
77	A novel heterozygous deletion in the EVC2 gene causes Weyers acrofacial dysostosis. <i>Human Genetics</i> , 2006, 119, 199-205.	1.8	42
78	Brain phenotypes in two FGFR2 mouse models for Apert syndrome. <i>Developmental Dynamics</i> , 2010, 239, 987-997.	0.8	42
79	Integrated Transcriptome and Network Analysis Reveals Spatiotemporal Dynamics of Calvarial Suturogenesis. <i>Cell Reports</i> , 2020, 32, 107871.	2.9	42
80	Genetic and Physical Mapping of the Treacher Collins Syndrome Locus with Respect to Loci in the Chromosome 5q3 Region. <i>Genomics</i> , 1993, 18, 7-13.	1.3	40
81	Prenatal ultrasonographic and molecular diagnosis of apert syndrome. , 1997, 17, 1081-1084.		39
82	Cochleosaccular Dysplasia Associated With a Connexin 26 Mutation in Keratitis/Ichthyosis/Deafness Syndrome. <i>Laryngoscope</i> , 2006, 116, 1404-1408.	1.1	39
83	The FGF and FGFR Gene Family and Risk of Cleft Lip with or Without Cleft Palate. <i>Cleft Palate-Craniofacial Journal</i> , 2013, 50, 96-103.	0.5	39
84	Physical and Genetic Mapping of a Human Apical Epithelial Na ⁺ /H ⁺ Exchanger (NHE3) Isoform to Chromosome 5p15.3. <i>Genomics</i> , 1993, 15, 668-672.	1.3	38
85	Human cDNA clones transcribed from an unusually high-molecular-weight RNA encode a new collagen chain. <i>Gene</i> , 1993, 123, 211-217.	1.0	37
86	Genome wide study of maternal and parental origin effects on the etiology of orofacial clefts. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 784-794.	0.7	37
87	Craniofacial divergence by distinct prenatal growth patterns in Fgfr2 mutant mice. <i>BMC Developmental Biology</i> , 2014, 14, 8.	2.1	37
88	The study of abnormal bone development in the Apert syndrome Fgfr2 +/S252W mouse using a 3D hydrogel culture model. <i>Bone</i> , 2008, 43, 55-63.	1.4	36
89	Gene expression in pharyngeal arch 1 during human embryonic development. <i>Human Molecular Genetics</i> , 2005, 14, 903-912.	1.4	35
90	Morphological comparison of the craniofacial phenotypes of mouse models expressing the Apert FGFR2 S252W mutation in neural crest- or mesoderm-derived tissues. <i>Bone</i> , 2014, 63, 101-109.	1.4	35

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91	Alphoid DNA polymorphisms for chromosome 21 can be distinguished from those of chromosome 13 using probes homologous to both. <i>Genomics</i> , 1991, 9, 141-146.	1.3	34
92	Two Craniosynostotic Syndrome Loci, Crouzon and Jackson-Weiss, Map to Chromosome 10q23-q26. <i>Genomics</i> , 1994, 22, 418-424.	1.3	33
93	Parental origin of mutations in sporadic cases of Treacher Collins syndrome. <i>European Journal of Human Genetics</i> , 2003, 11, 718-722.	1.4	33
94	Differential parental transmission of markers in <i>RUNX2</i> among cleft case-parent trios from four populations. <i>Genetic Epidemiology</i> , 2008, 32, 505-512.	0.6	33
95	Evidence of Gene-Environment Interaction for Two Genes on Chromosome 4 and Environmental Tobacco Smoke in Controlling the Risk of Nonsyndromic Cleft Palate. <i>PLoS ONE</i> , 2014, 9, e88088.	1.1	33
96	Chromosomal assignment of a gene encoding a new collagen type (COL15A1) to 9q21-q22. <i>Genomics</i> , 1992, 14, 220-224.	1.3	32
97	Localization of the Human Mxi1 Transcription Factor Gene (MX11) to Chromosome 10q24-q25. <i>Genomics</i> , 1994, 21, 669-672.	1.3	32
98	Genomic, cDNA and embryonic expression analysis of zebrafish IRF6, the gene mutated in the human oral clefting disorders Van der Woude and popliteal pterygium syndromes. <i>Gene Expression Patterns</i> , 2005, 5, 629-638.	0.3	32
99	Linkage Analysis Narrows the Critical Region for Oculodentodigital Dysplasia to Chromosome 6q22-q23. <i>Genomics</i> , 1999, 58, 34-40.	1.3	31
100	Quantitative Assessment of Facial Asymmetry Using Three-Dimensional Surface Imaging in Adults: Validating the Precision and Repeatability of a Global Approach. <i>Cleft Palate-Craniofacial Journal</i> , 2016, 53, 126-131.	0.5	30
101	Postnatal brain and skull growth in an Apert syndrome mouse model. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 745-757.	0.7	29
102	From shape to cells: mouse models reveal mechanisms altering palate development in Apert syndrome. <i>DMM Disease Models and Mechanisms</i> , 2013, 6, 768-79.	1.2	29
103	A Novel ZRS Mutation Leads to Preaxial Polydactyly Type 2 in a Heterozygous Form and Werner Mesomelic Syndrome in a Homozygous Form. <i>Human Mutation</i> , 2014, 35, 945-948.	1.1	29
104	Chromosomal deletion 4p15.32-p14 in a treacher collins syndrome patient: Exclusion of the disease locus from and mapping of anonymous DNA sequences to this region. <i>Genomics</i> , 1991, 11, 188-192.	1.3	28
105	A YAC Contig of Approximately 3 Mb from Human Chromosome 5q31-q33. <i>Genomics</i> , 1994, 19, 470-477.	1.3	28
106	Evidence of gene-environment interaction for the <i>RUNX2</i> gene and environmental tobacco smoke in controlling the risk of cleft lip with/without cleft palate. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2012, 94, 76-83.	1.6	28
107	Characterization of ocular motor deficits in congenital facial weakness: Moebius and related syndromes. <i>Brain</i> , 2014, 137, 1068-1079.	3.7	28
108	De MinimisRisk: A Proposal for a New Category of Research Risk. <i>American Journal of Bioethics</i> , 2011, 11, 1-7.	0.5	27

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109	Mutation Screening of Candidate Genes in Patients with Nonsyndromic Sagittal Craniosynostosis. <i>Plastic and Reconstructive Surgery</i> , 2016, 137, 952-961.	0.7	27
110	tgf β 23 regulation of chondrogenesis and osteogenesis in zebrafish is mediated through formation and survival of a subpopulation of the cranial neural crest. <i>Mechanisms of Development</i> , 2010, 127, 329-344.	1.7	26
111	Oral Clefting in China Over the Last Decade. <i>Plastic and Reconstructive Surgery - Global Open</i> , 2014, 2, e236.	0.3	26
112	Roberts/Pseudothalidomide Syndrome and Normal Intelligence: Approaches to Diagnosis and Management. <i>Developmental Medicine and Child Neurology</i> , 2008, 34, 534-539.	1.1	25
113	Association between genes on chromosome 4p16 and non-syndromic oral clefts in four populations. <i>European Journal of Human Genetics</i> , 2010, 18, 726-732.	1.4	25
114	FaceBase 3: analytical tools and FAIR resources for craniofacial and dental research. <i>Development (Cambridge)</i> , 2020, 147, .	1.2	25
115	Partial gonadal dysgenesis in a patient with a marker Y chromosome. <i>American Journal of Medical Genetics Part A</i> , 1992, 42, 807-812.	2.4	24
116	Cloning and chromosomal localization of the human BARX2 homeobox protein gene. <i>Gene</i> , 2000, 250, 171-180.	1.0	24
117	BMP4 Was Associated with NSCL/P in an Asian Population. <i>PLoS ONE</i> , 2012, 7, e35347.	1.1	24
118	Tetrasomy 21 in an infant with down syndrome and congenital leukemia. <i>American Journal of Medical Genetics Part A</i> , 1982, 12, 91-95.	2.4	23
119	A centromere-based genetic map of the short arm of human chromosome 6. <i>Genomics</i> , 1991, 9, 420-428.	1.3	23
120	Midface and upper airway dysgenesis in FGFR2-craniosynostosis involves multiple tissue-specific and cell cycle effects. <i>Development (Cambridge)</i> , 2018, 145, .	1.2	22
121	Identification of anHMGB3Frameshift Mutation in a Family With an X-linked Colobomatous Microphthalmia Syndrome Using Whole-Genome and X-Exome Sequencing. <i>JAMA Ophthalmology</i> , 2014, 132, 1215.	1.4	21
122	Novel Genes Mapping to the Critical Region of the 5q α ⁺ Syndrome. <i>Genomics</i> , 1997, 45, 88-96.	1.3	20
123	Genomic, cDNA, and embryonic expression analysis of zebrafish transforming growth factor beta 3 (tgf β 3). <i>Developmental Dynamics</i> , 2005, 232, 1021-1030.	0.8	20
124	Genetic and Lifestyle Causal Beliefs about Obesity and Associated Diseases among Ethnically Diverse Patients: A Structured Interview Study. <i>Public Health Genomics</i> , 2013, 16, 83-93.	0.6	20
125	Specific functional pathologies of Cx43 mutations associated with oculodentodigital dysplasia. <i>Molecular Biology of the Cell</i> , 2016, 27, 2172-2185.	0.9	20
126	Prenatal identification of small mosaic markers of different chromosomal origins. <i>Prenatal Diagnosis</i> , 1992, 12, 83-91.	1.1	19

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127	Mandibular dysmorphology due to abnormal embryonic osteogenesis in FGFR2-related craniosynostosis mice. <i>DMM Disease Models and Mechanisms</i> , 2019, 12, .	1.2	19
128	Mouse TCOF1 Is Expressed Widely, Has Motifs Conserved in Nucleolar Phosphoproteins, and Maps to Chromosome 18. <i>Biochemical and Biophysical Research Communications</i> , 1997, 238, 1-6.	1.0	18
129	Embryonic craniofacial bone volume and bone mineral density in <i>Fgfr2</i> ^{+P253R} and nonmutant mice. <i>Developmental Dynamics</i> , 2014, 243, 541-551.	0.8	18
130	Chromosomal localization of genes required for the terminal steps of oxidative metabolism: β and γ subunits of ATP synthase and the phosphate carrier. <i>Human Genetics</i> , 1994, 93, 600-2.	1.8	17
131	TCOF1 mutations excluded from a role in other first and second branchial arch-related disorders. <i>American Journal of Medical Genetics Part A</i> , 2002, 111, 324-327.	2.4	17
132	Mosaicism of aTCOF1 mutation in an individual clinically unaffected with treacher collins syndrome. <i>American Journal of Medical Genetics Part A</i> , 2004, 126A, 84-88.	2.4	17
133	Terminal deletion(4)(q33) in a male infant. <i>Clinical Genetics</i> , 1982, 21, 125-129.	1.0	17
134	Joint Testing of Genotypic and Gene-Environment Interaction Identified Novel Association for BMP4 with Non-Syndromic CL/P in an Asian Population Using Data from an International Cleft Consortium. <i>PLoS ONE</i> , 2014, 9, e109038.	1.1	17
135	Cleft Lip and/or Palate. <i>Journal of Craniofacial Surgery</i> , 2014, 25, 1601-1609.	0.3	16
136	Mutational Screening of FGFR1, CER1, and CDON in a Large Cohort of Trigenocephalic Patients. <i>Cleft Palate-Craniofacial Journal</i> , 2006, 43, 148-151.	0.5	15
137	Two different structural abnormalities of chromosome 13 in offspring of chromosomally normal parents with two fragile sites. <i>Clinical Genetics</i> , 1983, 23, 380-385.	1.0	15
138	Phylogenetic and evolutionary relationships and developmental expression patterns of the zebrafish twist gene family. <i>Development Genes and Evolution</i> , 2009, 219, 289-300.	0.4	15
139	A twisted hand: bHLH protein phosphorylation and dimerization regulate limb development. <i>BioEssays</i> , 2005, 27, 1102-1106.	1.2	14
140	Single maxillary central incisor and coloboma in hypomelanosis of Ko. <i>Clinical Genetics</i> , 1987, 31, 370-373.	1.0	14
141	Choanal Atresia and Craniosynostosis: Development and Disease. <i>Plastic and Reconstructive Surgery</i> , 2018, 141, 156-168.	0.7	14
142	Nonsyndromic craniosynostosis: novel coding variants. <i>Pediatric Research</i> , 2019, 85, 463-468.	1.1	14
143	Single-cell analysis identifies a key role for Hhip in murine coronal suture development. <i>Nature Communications</i> , 2021, 12, 7132.	5.8	14
144	Characterization of reiterated human DNA with respect to mammalian X chromosome homology. <i>Somatic Cell and Molecular Genetics</i> , 1984, 10, 93-103.	0.7	13

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148	TUBB3 Arg262His causes a recognizable syndrome including CFEOM3, facial palsy, joint contractures, and early-onset peripheral neuropathy. <i>Human Genetics</i> , 2021, 140, 1709-1731.	1.8	13
149	Novel Molecular Pathways Elicited by Mutant FGFR2 May Account for Brain Abnormalities in Apert Syndrome. <i>PLoS ONE</i> , 2013, 8, e60439.	1.1	12
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161	Tyrosine-dependent basolateral targeting of human connexin43-eYFP in Madin-Darby canine kidney cells can be disrupted by the oculodentodigital dysplasia mutation L90V. <i>FEBS Journal</i> , 2009, 276, 6992-7005.	2.2	5
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164	Aural atresia associated with multiple congenital anomalies and mental retardation: A new syndrome. <i>Journal of Pediatrics</i> , 1987, 110, 747-750.	0.9	3
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