Cynthia C Morton

List of Publications by Year
in descending order

184	15,184	17440 63	20358 116
papers	citations	h-index	g-index

194	194	194	15008
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Newborn Hearing Screening â€" A Silent Revolution. New England Journal of Medicine, 2006, 354, 2151-2164.	27.0	1,267
2	Mutations in the gene encoding B1 subunit of H+-ATPase cause renal tubular acidosis with sensorineural deafness. Nature Genetics, 1999, 21, 84-90.	21.4	633
3	Disruption of Neurexin 1 Associated with Autism Spectrum Disorder. American Journal of Human Genetics, 2008, 82, 199-207.	6.2	545
4	Sequencing Chromosomal Abnormalities Reveals Neurodevelopmental Loci that Confer Risk across Diagnostic Boundaries. Cell, 2012, 149, 525-537.	28.9	534
5	Association of Unconventional Myosin MYO15 Mutations with Human Nonsyndromic Deafness DFNB3. Science, 1998, 280, 1447-1451.	12.6	419
6	Disruption of the architectural factor HMGI-C: DNA-binding AT hook motifs fused in lipomas to distinct transcriptional regulatory domains. Cell, 1995, 82, 57-65.	28.9	415
7	Mutation of a Gene Encoding a Protein with Extracellular Matrix Motifs in Usher Syndrome Type IIa. Science, 1998, 280, 1753-1757.	12.6	366
8	Mutation in Transcription Factor POU4F3 Associated with Inherited Progressive Hearing Loss in Humans. Science, 1998, 279, 1950-1954.	12.6	322
9	Mutations in a novel cochlear gene cause DFNA9, a human nonsyndromic deafness with vestibular dysfunction. Nature Genetics, 1998, 20, 299-303.	21.4	317
10	Integration of cytogenetic landmarks into the draft sequence of the human genome. Nature, 2001, 409, 953-958.	27.8	302
11	Mutations in the novel protocadherin PCDH15 cause Usher syndrome type 1F. Human Molecular Genetics, 2001, 10, 1709-1718.	2.9	257
12	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	21.4	251
13	Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. Nature Genetics, 2012, 44, 390-397.	21.4	229
14	Human Î³-chain genes are rearranged in leukaemic T cells and map to the short arm of chromosome 7. Nature, 1985, 316, 549-552.	27.8	212
15	Mutations in the transcriptional activator EYA4 cause late-onset deafness at the DFNA10 locus. Human Molecular Genetics, 2001, 10, 195-200.	2.9	210
16	Assessment of 2q23.1 Microdeletion Syndrome Implicates MBD5 as a Single Causal Locus of Intellectual Disability, Epilepsy, and Autism Spectrum Disorder. American Journal of Human Genetics, 2011, 89, 551-563.	6.2	195
17	Disruption of ROBO2 Is Associated with Urinary Tract Anomalies and Confers Risk of Vesicoureteral Reflux. American Journal of Human Genetics, 2007, 80, 616-632.	6.2	189
18	Isolation of Novel and Known Genes from a Human Fetal Cochlear cDNA Library Using Subtractive Hybridization and Differential Screening. Genomics, 1994, 23, 42-50.	2.9	187

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19	RARE TRISOMY MOSAICISM DIAGNOSED IN AMNIOCYTES, INVOLVING AN AUTOSOME OTHER THAN CHROMOSOMES 13, 18, 20, AND 21: KARYOTYPE/PHENOTYPE CORRELATIONS. Prenatal Diagnosis, 1997, 17, 201-242.	2.3	185
20	Clinical Diagnosis by Whole-Genome Sequencing of a Prenatal Sample. New England Journal of Medicine, 2012, 367, 2226-2232.	27.0	174
21	Purification of the human NF-E2 complex: cDNA cloning of the hematopoietic cell-specific subunit and evidence for an associated partner.. Molecular and Cellular Biology, 1993, 13, 5604-5612.	2.3	172
22	SUMO1 Haploinsufficiency Leads to Cleft Lip and Palate. Science, 2006, 313, 1751-1751.	12.6	165
23	Next-Generation Sequencing Strategies Enable Routine Detection of Balanced Chromosome Rearrangements for Clinical Diagnostics and Genetic Research. American Journal of Human Genetics, 2011, 88, 469-481.	6.2	154
24	Analysis of androgen receptor DNA reveals the independent clonal origins of uterine leiomyomata and the secondary nature of cytogenetic aberrations in the development of leiomyomata. Genes Chromosomes and Cancer, 1994, 11, 1-6.	2.8	149
25	Molecular pathogenesis of uterine smooth muscle tumors from transcriptional profiling. Genes Chromosomes and Cancer, 2004, 40, 97-108.	2.8	145
26	High Prevalence of Symptoms of Meniere's Disease in three Families With a Mutation in the COCH Gene. Human Molecular Genetics, 1999, 8, 1425-1429.	2.9	144
27	Proteomics Reveal Cochlin Deposits Associated with Glaucomatous Trabecular Meshwork. Journal of Biological Chemistry, 2005, 280, 6080-6084.	3.4	140
28	Mapping and Characterization of a Novel Cochlear Gene in Human and in Mouse: A Positional Candidate Gene for a Deafness Disorder, DFNA9. Genomics, 1997, 46, 345-354.	2.9	139
29	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	21.4	131
30	HMGIC expression in human adult and fetal tissues and in uterine leiomyomata. Genes Chromosomes and Cancer, 1999, 25, 316-322.	2.8	125
31	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	2.4	125
32	A Pro51Ser mutation in the COCH gene is associated with late onset autosomal dominant progressive sensorineural hearing loss with vestibular defects. Human Molecular Genetics, 1999, 8, 361-366.	2.9	124
33	Cytogenetic abnormalities in uterine leiomyomata. Obstetrics and Gynecology, 1991, 77, 923-6.	2.4	121
34	Constitutional Rearrangement of the Architectural Factor HMGA2: A Novel Human Phenotype Including Overgrowth and Lipomas. American Journal of Human Genetics, 2005, 76, 340-348.	6.2	116
35	Cytogenetic evidence of clonality in a case of pigmented villonodular synovitis. Cancer, 1991, 67, 121-125.	4.1	113
36	Genetics of uterine leiomyomata. Genes Chromosomes and Cancer, 2000, 28, 235-245.	2.8	111

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37	Genetics, genomics and gene discovery in the auditory system. Human Molecular Genetics, 2002, 11, 1229-1240.	2.9	110
38	Chromosomal translocation t(8;12) induces aberrantHMGIC expression in aggressive angiomyxoma of the vulva. Genes Chromosomes and Cancer, 2001, 32, 172-176.	2.8	109
39	Uterine Leiomyomata with t(10;17) Disrupt the Histone Acetyltransferase MORF. Cancer Research, 2004, 64, 5570-5577.	0.9	106
40	Translocation breakpoints upstream of theHMGIC gene in uterine leiomyomata suggest dysregulation of this gene by a mechanism different from that in lipomas. , 1996, 17, 1-6.		102
41	GWAS Identifies 44 Independent Associated Genomic Loci for Self-Reported Adult Hearing Difficulty in UK Biobank. American Journal of Human Genetics, 2019, 105, 788-802.	6.2	101
42	Cochlin immunostaining of inner ear pathologic deposits and proteomic analysis in DFNA9 deafness and vestibular dysfunction. Human Molecular Genetics, 2006, 15, 1071-1085.	2.9	100
43	NFIA Haploinsufficiency Is Associated with a CNS Malformation Syndrome and Urinary Tract Defects. PLoS Genetics, 2007, 3, e80.	3.5	100
44	Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project. American Journal of Human Genetics, 2008, 82, 712-722.	6.2	95
45	MATR3 disruption in human and mouse associated with bicuspid aortic valve, aortic coarctation and patent ductus arteriosus. Human Molecular Genetics, 2015, 24, 2375-2389.	2.9	90
46	Genome-wide association and epidemiological analyses reveal common genetic origins between uterine leiomyomata and endometriosis. Nature Communications, 2019, 10, 4857.	12.8	90
47	Trisomy 5 and trisomy 7 are nonrandom aberrations in pigmented villonodular synovitis: Confirmation of trisomy 7 in uncultured cells. Genes Chromosomes and Cancer, 1992, 4, 264-266.	2.8	88
48	Increased Frequencies of Cochlin-Specific T Cells in Patients with Autoimmune Sensorineural Hearing Loss. Journal of Immunology, 2006, 177, 4203-4210.	0.8	88
49	Cytogenetic findings in pediatric adipose tumors: Consistent rearrangement of chromosome 8 in lipoblastoma. Genes Chromosomes and Cancer, 1993, 6, 24-29.	2.8	87
50	Cytogenetic abnormalities in uterine myomas are associated with myoma size. Molecular Human Reproduction, 1998, 4, 83-86.	2.8	85
51	Genome-wide Linkage and Association Analyses Implicate FASN in Predisposition to Uterine Leiomyomata. American Journal of Human Genetics, 2012, 91, 621-628.	6.2	83
52	The impact of race as a risk factor for symptom severity and age at diagnosis of uterine leiomyomata among affected sisters. American Journal of Obstetrics and Gynecology, 2008, 198, 168.e1-168.e9.	1.3	81
53	Fusion transcripts involving HMGA2 are not a common molecular mechanism in uterine leiomyomata with rearrangements in 12q15. Cancer Research, 2003, 63, 1351-8.	0.9	79
54	Subcellular localisation, secretion, and post-translational processing of normal cochlin, and of mutants causing the sensorineural deafness and vestibular disorder, DFNA9. Journal of Medical Genetics, 2003, 40, 479-486.	3.2	76

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55	The human ortholog of rhesus mannose-binding protein-A gene is an expressed pseudogene that localizes to Chromosome 10. Mammalian Genome, 1998, 9, 246-249.	2.2	71
56	Disseminated peritoneal leiomyomatosis. Clonality analysis by X chromosome inactivation and cytogenetics of a clinically benign smooth muscle proliferation. American Journal of Pathology, 1997, 150, 2153-66.	3.8	71
57	Human cochlear expressed sequence tags provide insight into cochlear gene expression and identify candidate genes for deafness. Human Molecular Genetics, 1999, 8, 439-452.	2.9	70
58	Intravenous leiomyomatosis is characterized by a der(14)t(12;14)(q15;q24). Genes Chromosomes and Cancer, 2003, 36, 205-206.	2.8	69
59	Genetic heterogeneity among uterine leiomyomata: insights into malignant progression. Human Molecular Genetics, 2007, 16, R7-R13.	2.9	69
60	Chromosome aberrations in uterine smooth muscle tumors: potential diagnostic relevance of cytogenetic instability. Cancer Research, 1990, 50, 4092-7.	0.9	68
61	ORC5L, a New Member of the Human Origin Recognition Complex, Is Deleted in Uterine Leiomyomas and Malignant Myeloid Diseases. Journal of Biological Chemistry, 1998, 273, 27137-27145.	3.4	67
62	Disseminated peritoneal leiomyomatosis after laparoscopic supracervical hysterectomy with characteristic molecular cytogenetic findings of uterine leiomyoma. Genes Chromosomes and Cancer, 2010, 49, 1152-1160.	2.8	67
63	HMGI(Y) expression in human uterine leiomyomata. Involvement of another high-mobility group architectural factor in a benign neoplasm. American Journal of Pathology, 1997, 150, 911-8.	3.8	67
64	Genome Sequencing Explores Complexity of Chromosomal Abnormalities in Recurrent Miscarriage. American Journal of Human Genetics, 2019, 105, 1102-1111.	6.2	66
65	SYCP2 Translocation-Mediated Dysregulation and Frameshift Variants Cause Human Male Infertility. American Journal of Human Genetics, 2020, 106, 41-57.	6.2	66
66	HMGA2 expression in uterine leiomyomata and myometrium: Quantitative analysis and tissue culture studies. Genes Chromosomes and Cancer, 2003, 38, 68-79.	2.8	65
67	Intravenous leiomyomatosis: an unusual intermediate between benign and malignant uterine smooth muscle tumors. Modern Pathology, 2016, 29, 500-510.	5.5	65
68	Involvement of fumarate hydratase in nonsyndromic uterine leiomyomas: Genetic linkage analysis and FISH studies. Genes Chromosomes and Cancer, 2004, 41, 183-190.	2.8	64
69	Low-pass genome sequencing versus chromosomal microarray analysis: implementation in prenatal diagnosis. Genetics in Medicine, 2020, 22, 500-510.	2.4	64
70	Clonal 6p21 rearrangement is restricted to the mesenchymal component of an endometrial polyp. Genes Chromosomes and Cancer, 1992, 5, 260-263.	2.8	63
71	Novel SSBP2â€JAK2 fusion gene resulting from a t(5;9)(q14.1;p24.1) in preâ€B acute lymphocytic leukemia. Genes Chromosomes and Cancer, 2008, 47, 884-889.	2.8	63
72	A proposal for comprehensive newborn hearing screening to improve identification of deaf and hard-of-hearing children. Genetics in Medicine, 2019, 21, 2614-2630.	2.4	63

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73	Consistent cytogenetic aberrations in hepatoblastoma: A common pathway of genetic alterations in embryonal liver and skeletal muscle malignancies?. Genes Chromosomes and Cancer, 1991, 3, 37-43.	2.8	62
74	Uterine leiomyomata with deletions of Ip represent a distinct cytogenetic subgroup associated with unusual histologic features. Genes Chromosomes and Cancer, 2006, 45, 304-312.	2.8	62
75	Disruption of a Large Intergenic Noncoding RNA in Subjects with Neurodevelopmental Disabilities. American Journal of Human Genetics, 2012, 91, 1128-1134.	6.2	61
76	Haploinsufficiency of KDM6A is associated with severe psychomotor retardation, global growth restriction, seizures and cleft palate. Human Genetics, 2013, 132, 537-552.	3.8	60
77	Purification of the Human NF-E2 Complex: cDNA Cloning of the Hematopoietic Cell-Specific Subunit and Evidence for an Associated Partner. Molecular and Cellular Biology, 1993, 13, 5604-5612.	2.3	59
78	A novelDFNA9mutation in the vWFA2 domain ofCOCHalters a conserved cysteine residue and intrachain disulfide bond formation resulting in progressive hearing loss and site-specific vestibular and central oculomotor dysfunction. American Journal of Medical Genetics, Part A, 2005, 139A, 86-95.	1.2	58
79	The Genetics of Uterine Leiomyomata. Obstetrics and Gynecology, 2006, 107, 917-921.	2.4	58
80	Yield of additional genetic testing after chromosomal microarray for diagnosis of neurodevelopmental disability and congenital anomalies: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2018, 20, 1105-1113.	2.4	57
81	Ion Channel Gene Expression in the Inner Ear. JARO - Journal of the Association for Research in Otolaryngology, 2007, 8, 305-328.	1.8	56
82	Genetics and the Development of Fibroids. Clinical Obstetrics and Gynecology, 2001, 44, 335-349.	1.1	55
83	Fluorescencein situ hybridization (FISH) for rapid detection of aneuploidy: experience in 911 prenatal cases. Prenatal Diagnosis, 2001, 21, 262-269.	2.3	55
84	Hypogonadotropic hypogonadism and cleft lip and palate caused by a balanced translocation producing haploinsufficiency for FGFR1. Journal of Medical Genetics, 2005, 42, 666-672.	3.2	55
85	The del(7q) subgroup in uterine leiomyomata: Genetic and biologic characteristics. Cancer Genetics and Cytogenetics, 1997, 98, 69-74.	1.0	54
86	Disruption of MBD5 contributes to a spectrum of psychopathology and neurodevelopmental abnormalities. Molecular Psychiatry, 2014, 19, 368-379.	7.9	54
87	Structural Chromosomal Rearrangements Require Nucleotide-Level Resolution: Lessons from Next-Generation Sequencing in Prenatal Diagnosis. American Journal of Human Genetics, 2016, 99, 1015-1033.	6.2	53
88	Identification of balanced chromosomal rearrangements previously unknown among participants in the 1000 Genomes Project: implications for interpretation of structural variation in genomes and the future of clinical cytogenetics. Genetics in Medicine, 2018, 20, 697-707.	2.4	52
89	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. American Journal of Human Genetics, 2017, 101, 206-217.	6.2	51
90	Identification of a YAC spanning the translocation breakpoints in uterine leiomyomata, pulmonary chondroid hamartoma, and lipoma: physical mapping of the 12q14â€“q15 breakpoint region in uterine leiomyomata. Genomics, 1995, 26, 265-271.	2.9	50

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91	Historical and Clinical Perspectives on Chromosomal Translocations. Advances in Experimental Medicine and Biology, 2018, 1044, 1-14.	1.6	50
92	Amplification ofAML1 in childhood acute lymphoblastic leukemias. Genes Chromosomes and Cancer, 2001, 30, 407-409.	2.8	49
93	Expression of HMGIY in Three Uterine Leiomyomata with Complex Rearrangements of Chromosome 6. Cancer Genetics and Cytogenetics, 1999, 114, 9-16.	1.0	48
94	Isolation from Cochlea of a Novel Human Intronless Gene with Predominant Fetal Expression. JARO - Journal of the Association for Research in Otolaryngology, 2004, 5, 185-202.	1.8	48
95	Identification of Pathogenic Mechanisms ofCOCHMutations, Abolished Cochlin Secretion, and Intracellular Aggregate Formation: Genotype-Phenotype Correlations in DFNA9 Deafness and Vestibular Disorder. Human Mutation, 2014, 35, 1506-1513.	2.5	48
96	HMGIC expression in human adult and fetal tissues and in uterine leiomyomata. Genes Chromosomes and Cancer, 1999, 25, 316-22.	2.8	47
97	The Iceberg under Water: Unexplored Complexity of Chromoanagenesis in Congenital Disorders. American Journal of Human Genetics, 2019, 104, 565-577.	6.2	46
98	Localization and expression of the human estrogen receptor beta gene in uterine leiomyomata. , 1998, 23, 361-366.		45
99	A targeted Coch missense mutation: a knock-in mouse model for DFNA9 late-onset hearing loss and vestibular dysfunction. Human Molecular Genetics, 2008, 17, 3426-3434.	2.9	44
100	Expression profiling of uterine leiomyomata cytogenetic subgroups reveals distinct signatures in matched myometrium: transcriptional profiling of the t(12;14) and evidence in support of predisposing genetic heterogeneity. Human Molecular Genetics, 2012, 21, 2312-2329.	2.9	44
101	Genomic structure, functional comparison, and tissue distribution of mouse Cd59a and Cd59b. Mammalian Genome, 2001, 12, 582-589.	2.2	43
102	A Novel Conserved Cochlear Gene, OTOR: Identification, Expression Analysis, and Chromosomal Mapping. Genomics, 2000, 66, 242-248.	2.9	42
103	Describing Sequencing Results of Structural Chromosome Rearrangements with a Suggested Next-Generation Cytogenetic Nomenclature. American Journal of Human Genetics, 2014, 94, 695-709.	6.2	42
104	NR2F1 deletion in a patient with a de novo paracentric inversion, inv(5)(q15q33.2), and syndromic deafness. American Journal of Medical Genetics, Part A, 2009, 149A, 931-938.	1.2	41
105	Uterine leiomyomata and decreased height: a common HMGA2 predisposition allele. Human Genetics, 2009, 125, 257-263.	3.8	40
106	Implication of LRRC4C and DPP6 in neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2017, 173, 395-406.	1.2	40
107	Nationwide population genetic screening improves outcomes of newborn screening for hearing loss in China. Genetics in Medicine, 2019, 21, 2231-2238.	2.4	40
108	Translocations in 7q22 define a critical region in uterine leiomyomata. Cancer Genetics and Cytogenetics, 1994, 77, 65-68.	1.0	39

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109	A multi-stage genome-wide association study of uterine fibroids in African Americans. Human Genetics, 2017, 136, 1363-1373.	3.8	39
110	An Ancient Conserved Gene Expressed in the Human Inner Ear: Identification, Expression Analysis, and Chromosomal Mapping of Human and Mouse Antiquitin (ATQ1). Genomics, 1997, 46, 191-199.	2.9	38
111	Audiometric, Vestibular, and Genetic Aspects of a DFNA9 Family with a G88E COCH Mutation. Otology and Neurotology, 2005, 26, 926-933.	1.3	38
112	The Genetic Heterogeneity of Uterine Leiomyomata. Obstetrics and Gynecology Clinics of North America, 2006, 33, 13-39.	1.9	38
113	Hearing and vestibular deficits in the Coch null mouse model: Comparison to the Coch mouse and to DFNA9 hearing and balance disorder. Hearing Research, 2011, 272, 42-48.	2.0	37
114	A newly established metastatic breast tumor cell line with integrated amplified copies ofERBB2 and double minute chromosomes. Genes Chromosomes and Cancer, 1989, 1, 48-58.	2.8	33
115	Norrie disease gene mutation in a large Costa Rican kindred with a novel phenotype including venous insufficiency. Human Mutation, 1997, 9, 402-408.	2.5	33
116	Targeted disruption of mouse Coch provides functional evidence that DFNA9 hearing loss is not a COCH haploinsufficiency disorder. Human Genetics, 2005, 118, 29-34.	3.8	33
117	Deletion of an enhancer near DLX5 and DLX6 in a family with hearing loss, craniofacial defects, and an inv(7)(q21.3q35). Human Genetics, 2010, 127, 19-31.	3.8	32
118	Prenatal diagnosis of chromothripsis, with nine breaks characterized by karyotyping, FISH, microarray and wholeâ€genome sequencing. Prenatal Diagnosis, 2015, 35, 299-301.	2.3	30
119	Actin capping protein CAPZB regulates cell morphology, differentiation, and neural crest migration in craniofacial morphogenesis. Human Molecular Genetics, 2016, 25, 1255-1270.	2.9	30
120	Cochlin in the eye: Functional implications. Progress in Retinal and Eye Research, 2007, 26, 453-469.	15.5	29
121	Identifying the molecular signature of the interstitial deletion 7q subgroup of uterine leiomyomata using a paired analysis. Genes Chromosomes and Cancer, 2009, 48, 865-885.	2.8	29
122	Mechanisms for Structural Variation in the Human Genome. Current Genetic Medicine Reports, 2013, 1, 81-90.	1.9	29
123	Dysregulation ofHMGIC in a uterine lipoleiomyoma with a complex rearrangement including chromosomes 7, 12, and 14. Genes Chromosomes and Cancer, 2000, 27, 209-215.	2.8	28
124	Genome-wide association meta-analysis identifies 48 risk variants and highlights the role of the stria vascularis in hearing loss. American Journal of Human Genetics, 2022, 109, 1077-1091.	6.2	27
125	Gene Discovery in the Auditory System: Characterization of Additional Cochlear-Expressed Sequences. JARO - Journal of the Association for Research in Otolaryngology, 2002, 3, 45-53.	1.8	26
126	Screening of deafness-causing DNA variants that are common in patients of European ancestry using a microarray-based approach. PLoS ONE, 2017, 12, e0169219.	2.5	26

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127	Generation of protective pneumococcal-specific nasal resident memory CD4+ T cells via parenteral immunization. Mucosal Immunology, 2020, 13, 172-182.	6.0	26
128	Molecular and Cytogenetic Characterization of Plexiform Leiomyomata Provide Further Evidence for Genetic Heterogeneity Underlying Uterine Fibroids. American Journal of Pathology, 2008, 172, 1403-1410.	3.8	24
129	A t(2;19)(p13;p13.2) in a giant invasive cardiac lipoma from a patient with multiple lipomatosis. , 2000, 28, 133-137.		22
130	Mapping by chromosome sorting of several gene probes, including C-myc, to the derivative chromosomes of a 3;8 translocation associated with familial renal cancer. Cytometry, 1986, 7, 589-594.	1.8	21
131	Training the Future Leaders in Personalized Medicine. Journal of Personalized Medicine, 2016, 6, 1.	2.5	21
132	A novel COCH mutation associated with autosomal dominant nonsyndromic hearing loss disrupts the structural stability of the vWFA2 domain. Journal of Molecular Medicine, 2012, 90, 1321-1331.	3.9	20
133	Expression studies of osteoglycin/mimecan (OGN) in the cochlea and auditory phenotype of Ogn-deficient mice. Hearing Research, 2008, 237, 57-65.	2.0	18
134	Histopathology of the Human Inner Ear in the p.L114P COCH Mutation (DFNA9). Audiology and Neuro-Otology, 2016, 21, 88-97.	1.3	18
135	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212.	2.4	18
136	Cochlin in Normal Middle Ear and Abnormal Middle Ear Deposits in DFNA9 and Coch G88E/G88E Mice. JARO - Journal of the Association for Research in Otolaryngology, 2014, 15, 961-974.	1.8	17
137	Section E6.5â€“6.8 of the ACMG technical standards and guidelines: chromosome studies of lymph node and solid tumorâ€“acquired chromosomal abnormalities. Genetics in Medicine, 2016, 18, 643-648.	2.4	17
138	Genetic Approaches to the Study of Uterine Leiomyomata. Environmental Health Perspectives, 2000, 108, 775-778.	6.0	17
139	Beginning of a molecular era in hearing and deafness. Clinical Genetics, 1999, 55, 149-159.	2.0	16
140	Low-pass genome sequencingâ€“based detection of absence of heterozygosity: validation in clinical cytogenetics. Genetics in Medicine, 2021, 23, 1225-1233.	2.4	16
141	Many Tumors and Many Genes. American Journal of Pathology, 1998, 153, 1015-1020.	3.8	15
142	Loss of LDAH associated with prostate cancer and hearing loss. Human Molecular Genetics, 2018, 27, 4194-4203.	2.9	14
143	Characterization of an Abundant COL9A1 Transcript in the Cochlea with a Novel 3â€² UTR: Expression Studies and Detection of miRNA Target Sequence. JARO - Journal of the Association for Research in Otolaryngology, 2006, 7, 160-172.	1.8	13
144	Genetic Association Studies in Uterine Fibroids: Risk Alleles Presage the Path to Personalized Therapies. Seminars in Reproductive Medicine, 2016, 34, 235-241.	1.1	13

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145	Identification of genetically aberrant cell lineages in Wilms' tumors. Genes Chromosomes and Cancer, 1994, 10, 40-48.	2.8	12
146	Methylthioadenosine phosphorylase (MTAP) in hearing: Gene disruption by chromosomal rearrangement in a hearing impaired individual and model organism analysis. American Journal of Medical Genetics, Part A, 2007, 143A, 1630-1639.	1.2	12
147	Estrogen-related receptor gamma implicated in a phenotype including hearing loss and mild developmental delay. European Journal of Human Genetics, 2016, 24, 1622-1626.	2.8	12
148	Clinical, pathologic, cytogenetic, and molecular profiling in self-identified black women with uterine leiomyomata. Cancer Genetics, 2018, 222-223, 1-8.	0.4	12
149	Cochlin Deficiency Protects Against Noise-Induced Hearing Loss. Frontiers in Molecular Neuroscience, 2021, 14, 670013.	2.9	11
150	Balanced Chromosomal Rearrangement Detection by Lowâ€Pass Wholeâ€Genome Sequencing. Current Protocols in Human Genetics, 2018, 96, 8.18.1-8.18.16.	3.5	10
151	1q42â^¼q44 is rarely cytogenetically involved in sporadic uterine leiomyomata. Cancer Genetics and Cytogenetics, 2002, 138, 92-93.	1.0	9
152	Pelvic and pulmonary benign metastasizing leiomyoma: A case report. Case Reports in Women's Health, 2018, 18, e00061.	0.5	9
153	Comprehensive clinically oriented workflow for nucleotide level resolution and interpretation in prenatal diagnosis of de novo apparently balanced chromosomal translocations in their genomic landscape. Human Genetics, 2020, 139, 531-543.	3.8	9
154	Long-range cis-regulatory elements controlling GDF6 expression are essential for ear development. Journal of Clinical Investigation, 2020, 130, 4213-4217.	8.2	9
155	Continuing to break the sound barrier: genes in hearing. Current Opinion in Genetics and Development, 2002, 12, 343-348.	3.3	8
156	Phenotypic interpretation of complex chromosomal rearrangements informed by nucleotide-level resolution and structural organization of chromatin. European Journal of Human Genetics, 2018, 26, 374-381.	2.8	8
157	A synonymous variant in MYO15A enriched in the Ashkenazi Jewish population causes autosomal recessive hearing loss due to abnormal splicing. European Journal of Human Genetics, 2021, 29, 988-997.	2.8	8
158	Position effects of 22q13 rearrangements on candidate genes in Phelan-McDermid syndrome. PLoS ONE, 2021, 16, e0253859.	2.5	8
159	Primary Parauterine Leiomyoma With a t(6;14). , 1999, 26, 385-386.		6
160	Gene discovery in the auditory system using a tissue specific approach. American Journal of Medical Genetics Part A, 2004, 130A, 26-28.	2.4	6
161	Next-Generation Newborn Hearing Screening. Monographs in Human Genetics, 2016, , 30-39.	0.5	6
162	3C-PCR: a novel proximity ligation-based approach to phase chromosomal rearrangement breakpoints with distal allelic variants. Human Genetics, 2018, 137, 55-62.	3.8	5

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163	Prenatal diagnosis and molecular cytogenetics in a case of partial trisomy 14 and monosomy 21. American Journal of Medical Genetics Part A, 2001, 100, 246-250.	2.4	4
164	Experience of Low-Pass Whole-Genome Sequencing-Based Copy Number Variant Analysis: A Survey of Chinese Tertiary Hospitals. Diagnostics, 2022, 12, 1098.	2.6	4
165	Complex cytogenetic rearrangements at the DURS 1 locus in syndromic Duane retraction syndrome. Clinical Case Reports (discontinued), 2013, 1, 30-37.	0.5	3
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