List of Publications by Year in descending order

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		41344	29157
133	11,664	49	104
papers	citations	h-index	g-index
141	141	141	10473
all docs	docs citations	times ranked	citing authors

#	Article	IF	CITATIONS
1	The DNA sequence of human chromosome 22. Nature, 1999, 402, 489-495.	27.8	1,086
2	22q11.2 deletion syndrome. Nature Reviews Disease Primers, 2015, 1, 15071.	30.5	954
3	TBX1 Is Responsible for Cardiovascular Defects in Velo-Cardio-Facial/DiGeorge Syndrome. Cell, 2001, 104, 619-629.	28.9	884
4	Schizophrenia susceptibility associated with interstitial deletions of chromosome 22q11 Proceedings of the National Academy of Sciences of the United States of America, 1995, 92, 7612-7616.	7.1	591
5	Meiotic Pachytene Arrest in MLH1-Deficient Mice. Cell, 1996, 85, 1125-1134.	28.9	528
6	Psychotic Illness in Patients Diagnosed with Velo-Cardio-Facial Syndrome and Their Relatives. Journal of Nervous and Mental Disease, 1994, 182, 476-477.	1.0	394
7	A common molecular basis for rearrangement disorders on chromosome 22q11. Human Molecular Genetics, 1999, 8, 1157-1167.	2.9	385
8	Low-Copy Repeats Mediate the Common 3-Mb Deletion in Patients with Velo-cardio-facial Syndrome. American Journal of Human Genetics, 1999, 64, 1076-1086.	6.2	340
9	VEGF: A modifier of the del22q11 (DiGeorge) syndrome?. Nature Medicine, 2003, 9, 173-182.	30.7	288
10	Association of codon 108/158 catechol-O-methyltransferase gene polymorphism with the psychiatric manifestations of velo-cardio-facial syndrome. American Journal of Medical Genetics Part A, 1996, 67, 468-472.	2.4	259
11	Genomic Disorders on 22q11. American Journal of Human Genetics, 2002, 70, 1077-1088.	6.2	228
12	Microduplication and Triplication of 22q11.2: A Highly Variable Syndrome. American Journal of Human Genetics, 2005, 76, 865-876.	6.2	221
13	Abraham's Children in the Genome Era: Major Jewish Diaspora Populations Comprise Distinct Genetic Clusters with Shared Middle Eastern Ancestry. American Journal of Human Genetics, 2010, 86, 850-859.	6.2	217
14	Full spectrum of malformations in velo-cardio-facial syndrome/DiGeorge syndrome mouse models by altering Tbx1 dosage. Human Molecular Genetics, 2004, 13, 1577-1585.	2.9	214
15	AT-Rich Palindromes Mediate the Constitutional t(11;22) Translocation. American Journal of Human Genetics, 2001, 68, 1-13.	6.2	175
16	Suppression of neural fate and control of inner ear morphogenesis by <i>Tbx1</i> . Development (Cambridge), 2004, 131, 1801-1812.	2.5	150
17	Inactivation of <i>Tbx1</i> in the pharyngeal endoderm results in 22q11DS malformations. Development (Cambridge), 2006, 133, 977-987.	2.5	146
18	Velo-cardio-facial syndrome: Frequency and extent of 22q1l deletions. American Journal of Medical Genetics Part A, 1995, 57, 514-522.	2.4	134

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19	Tbx1 affects asymmetric cardiac morphogenesis by regulating <i>Pitx2</i> in the secondary heart field. Development (Cambridge), 2006, 133, 1565-1573.	2.5	132
20	ldentification of downstream genetic pathways of Tbx1 in the second heart field. Developmental Biology, 2008, 316, 524-537.	2.0	124
21	A Tbx1-Six1/Eya1-Fgf8 genetic pathway controls mammalian cardiovascular and craniofacial morphogenesis. Journal of Clinical Investigation, 2011, 121, 1585-1595.	8.2	123
22	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. New England Journal of Medicine, 2017, 376, 742-754.	27.0	120
23	Shuffling of Genes Within Low-Copy Repeats on 22q11 (LCR22) by Alu-Mediated Recombination Events During Evolution. Genome Research, 2003, 13, 2519-2532.	5.5	115
24	Molecular and clinical study of 183 patients with conotruncal anomaly face syndrome. Human Genetics, 1998, 103, 70-80.	3.8	114
25	Comparative mapping of the human 22q11 chromosomal region and the orthologous region in mice reveals complex changes in gene organization. Proceedings of the National Academy of Sciences of the United States of America, 1997, 94, 14608-14613.	7.1	104
26	Identification of a New Human Catenin Gene Family Member (ARVCF) from the Region Deleted in Velo–Cardio–Facial Syndrome. Genomics, 1997, 41, 75-83.	2.9	103
27	A model for transcription termination by RNA polymerase I. Cell, 1994, 79, 527-534.	28.9	102
28	Dual embryonic origin of the mammalian otic vesicle forming the inner ear. Development (Cambridge), 2011, 138, 5403-5414.	2.5	102
29	Identification, Characterization, and Precise Mapping of a Human Gene Encoding a Novel Membrane-Spanning Protein from the 22q11 Region Deleted in Velo–Cardio–Facial Syndrome. Genomics, 1997, 42, 245-251.	2.9	96
30	Molecular genetics of 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2070-2081.	1.2	96
31	Tissue-specific roles of Tbx1 in the development of the outer, middle and inner ear, defective in 22q11DS patients. Human Molecular Genetics, 2006, 15, 1629-1639.	2.9	91
32	Using common genetic variation to examine phenotypic expression and risk prediction in 22q11.2 deletion syndrome. Nature Medicine, 2020, 26, 1912-1918.	30.7	90
33	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510.	7.9	87
34	The Role of Neural Crest during Cardiac Development in a Mouse Model of DiGeorge Syndrome. Developmental Biology, 2002, 251, 157-166.	2.0	85
35	Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. American Journal of Psychiatry, 2017, 174, 1054-1063.	7.2	77
36	Behavior of mice with mutations in the conserved region deleted in velocardiofacial/DiGeorge syndrome. Neurogenetics, 2006, 7, 247-257.	1.4	70

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37	The 22q11 deletion syndrome candidate gene Tbx1 determines thyroid size and positioning. Human Molecular Genetics, 2007, 16, 276-285.	2.9	67
38	Copy-Number Variation of the Glucose Transporter Gene SLC2A3 and Congenital Heart Defects in the 22q11.2 Deletion Syndrome. American Journal of Human Genetics, 2015, 96, 753-764.	6.2	62
39	Mouse and Human CRKL Is Dosage Sensitive for Cardiac Outflow Tract Formation. American Journal of Human Genetics, 2015, 96, 235-244.	6.2	58
40	A Common Breakpoint on 11q23 in Carriers of the Constitutional t(11;22) Translocation. American Journal of Human Genetics, 1999, 65, 1608-1616.	6.2	57
41	T-genes and limb bud development. American Journal of Medical Genetics, Part A, 2006, 140A, 1407-1413.	1.2	57
42	Genotype and cardiovascular phenotype correlations with TBX1 in 1,022 velo-cardio-facial/digeorge/22q11.2 deletion syndrome patients. Human Mutation, 2011, 32, 1278-1289.	2.5	57
43	Isolation of a new clathrin heavy chain gene with muscle-specific expression from the region commonly deleted in velo-cardio-facial syndrome. Human Molecular Genetics, 1996, 5, 617-624.	2.9	55
44	Over-expression of a human chromosome 22q11.2 segment including TXNRD2, COMT and ARVCF developmentally affects incentive learning and working memory in mice. Human Molecular Genetics, 2009, 18, 3914-3925.	2.9	53
45	Enhanced Maternal Origin of the 22q11.2 Deletion in Velocardiofacial and DiGeorge Syndromes. American Journal of Human Genetics, 2013, 92, 439-447.	6.2	53
46	Mesodermal Tbx1 is required for patterning the proximal mandible in mice. Developmental Biology, 2010, 344, 669-681.	2.0	52
47	Detection of an Atypical 22q11 Deletion That Has No Overlap with the DiGeorge Syndrome Critical Region. American Journal of Human Genetics, 1997, 60, 1544-1547.	6.2	52
48	Detection of an Atypical 22q11 Deletion That Has No Overlap with the DiGeorge Syndrome Critical Region. American Journal of Human Genetics, 1997, 60, 1544-1548.	6.2	51
49	Identification of Critical Regions and Candidate Genes for Cardiovascular Malformations and Cardiomyopathy Associated with Deletions of Chromosome 1p36. PLoS ONE, 2014, 9, e85600.	2.5	51
50	A Pedigree-Based Map of Recombination in the Domestic Dog Genome. G3: Genes, Genomes, Genetics, 2016, 6, 3517-3524.	1.8	51
51	Changes in the guanine nucleotide-binding proteins, Giand go, during differentiation of 3T3-L1 cells. FEBS Letters, 1986, 199, 103-106.	2.8	50
52	Der(22) Syndrome and Velo-Cardio-Facial Syndrome/DiGeorge Syndrome Share a 1.5-Mb Region of Overlap on Chromosome 22q11. American Journal of Human Genetics, 1999, 64, 747-758.	6.2	50
53	Mutations in <i>GJB2</i> , <i>GJB6</i> , and mitochondrial DNA are rare in African American and Caribbean Hispanic individuals with hearing impairment. American Journal of Medical Genetics, Part A, 2007, 143A, 830-838.	1.2	50
54	Refinement of causative genes in monosomy 1p36 through clinical and molecular cytogenetic characterization of small interstitial deletions. American Journal of Medical Genetics, Part A, 2010, 152A, 1951-1959.	1.2	50

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55	Frequent translocations occur between low copy repeats on chromosome 22q11.2 (LCR22s) and telomeric bands of partner chromosomes. Human Molecular Genetics, 2003, 12, 1823-1837.	2.9	49
56	Histone Modifier Genes Alter Conotruncal Heart Phenotypes in 22q11.2 Deletion Syndrome. American Journal of Human Genetics, 2015, 97, 869-877.	6.2	49
57	Genetic modifiers of the physical malformations in veloâ€cardioâ€facial syndrome/DiGeorge syndrome. Developmental Disabilities Research Reviews, 2008, 14, 19-25.	2.9	48
58	Genotype–phenotype correlation in interstitial 6q deletions: a report of 12 new cases. Neurogenetics, 2012, 13, 31-47.	1.4	48
59	Dissection of Tbx1 and Fgf interactions in mouse models of 22q11DS suggests functional redundancy. Human Molecular Genetics, 2006, 15, 3219-3228.	2.9	47
60	Canonical Wnt signaling modulates <i>Tbx1</i> , <i>Eya1</i> , and <i>Six1</i> expression, restricting neurogenesis in the otic vesicle. Developmental Dynamics, 2010, 239, 1708-1722.	1.8	47
61	Cooperative Function of Tbx1 and Brn4 in the Periotic Mesenchyme is Necessary for Cochlea Formation. JARO - Journal of the Association for Research in Otolaryngology, 2008, 9, 33-43.	1.8	46
62	Tbx1 and Brn4regulate retinoic acid metabolic genes during cochlear morphogenesis. BMC Developmental Biology, 2009, 9, 31.	2.1	46
63	A 200-kb region of human chromosome 22q11.2 confers antipsychotic-responsive behavioral abnormalities in mice. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 19132-19137.	7.1	44
64	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. Human Genetics, 2016, 135, 273-285.	3.8	43
65	Gene expression profile of trisomy 21 placentas: A potential approach for designing noninvasive techniques of prenatal diagnosis. American Journal of Obstetrics and Gynecology, 2002, 187, 457-462.	1.3	42
66	Complete Sequence of the 22q11.2 Allele in 1,053 Subjects with 22q11.2 Deletion Syndrome Reveals Modifiers of Conotruncal Heart Defects. American Journal of Human Genetics, 2020, 106, 26-40.	6.2	42
67	The 22q11 low copy repeats are characterized by unprecedented size and structural variability. Genome Research, 2019, 29, 1389-1401.	5.5	39
68	REGULATION OF GROWTH AND DIFFERENTIATION OF EPITHELIAL CELLS BY HORMONES, GROWTH FACTORS, AND SUBSTRATES OF EXTRACELLULAR MATRIX. Annals of the New York Academy of Sciences, 1981, 372, 354-370.	3.8	36
69	New cases and refinement of the critical region in the 1q41q42 microdeletion syndrome. European Journal of Medical Genetics, 2011, 54, 42-49.	1.3	36
70	Mammalian TBX1 Preferentially Binds and Regulates Downstream Targets Via a Tandem T-site Repeat. PLoS ONE, 2014, 9, e95151.	2.5	33
71	Variance of IQ is partially dependent on deletion type among 1,427 22q11.2 deletion syndrome subjects. American Journal of Medical Genetics, Part A, 2018, 176, 2172-2181.	1.2	33
72	Hominoid lineage specific amplification of low-copy repeats on 22q11.2 (LCR22s) associated with velo-cardio-facial/digeorge syndrome. Human Molecular Genetics, 2007, 16, 2560-2571.	2.9	32

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73	Tbx1 is required autonomously for cell survival and fate in the pharyngeal core mesoderm to form the muscles of mastication. Human Molecular Genetics, 2014, 23, 4215-4231.	2.9	31
74	A Novel C-Terminal CIB2 (Calcium and Integrin Binding Protein 2) Mutation Associated with Non-Syndromic Hearing Loss in a Hispanic Family. PLoS ONE, 2015, 10, e0133082.	2.5	31
75	Single cell multi-omic analysis identifies a Tbx1-dependent multilineage primed population in murine cardiopharyngeal mesoderm. Nature Communications, 2021, 12, 6645.	12.8	31
76	Traffic of genetic information between segmental duplications flanking the typical 22q11.2 deletion in velo-cardio-facial syndrome/DiGeorge syndrome. Genome Research, 2005, 15, 1487-1495.	5.5	30
77	AT-rich repeats associated with chromosome 22q11.2 rearrangement disorders shape human genome architecture on Yq12. Genome Research, 2007, 17, 451-460.	5.5	30
78	Agnathia–otocephaly complex: A case report and examination of the OTX2 and PRRX1 genes. Gene, 2012, 494, 124-129.	2.2	28
79	Association between autism spectrum disorder in individuals with velocardiofacial (22q11.2 deletion) syndrome and PRODH and COMT genotypes. Psychiatric Genetics, 2014, 24, 269-272.	1.1	28
80	The Role of mGluR Copy Number Variation in Genetic and Environmental Forms of Syndromic Autism Spectrum Disorder. Scientific Reports, 2016, 6, 19372.	3.3	28
81	Incontinentia pigmenti in a newborn male infant with DNA confirmation. , 1998, 75, 159-163.		27
82	Reduced dosage of β-catenin provides significant rescue of cardiac outflow tract anomalies in a Tbx1 conditional null mouse model of 22q11.2 deletion syndrome. PLoS Genetics, 2017, 13, e1006687.	3.5	27
83	Tbx1 and Foxi3 genetically interact in the pharyngeal pouch endoderm in a mouse model for 22q11.2 deletion syndrome. PLoS Genetics, 2019, 15, e1008301.	3.5	27
84	Characterization of the past and current duplication activities in the human 22q11.2 region. BMC Genomics, 2011, 12, 71.	2.8	25
85	Characterization and Mutation Analysis of Goosecoid-like (GSCL), a Homeodomain-Containing Gene That Maps to the Critical Region for VCFS/DGS on 22q11. Genomics, 1997, 46, 364-372.	2.9	24
86	Endodermâ€specific deletion of <i>Tbx1</i> reveals an FGFâ€independent role for Tbx1 in pharyngeal apparatus morphogenesis. Developmental Dynamics, 2014, 243, 1143-1151.	1.8	24
87	Isolation and Characterization of a Human Gene Containing a Nuclear Localization Signal from the Critical Region for Velo–Cardio–Facial Syndrome on 22q11. Genomics, 1998, 53, 146-154.	2.9	22
88	Spectrum of elastin sequence variants and cardiovascular phenotypes in 49 patients with Williams–Beuren syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 527-533.	1.2	22
89	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the <i>GPR98</i> Locus on 5q14.3. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	22
90	Deletion size analysis of 1680 22q11.2DS subjects identifies a new recombination hotspot on chromosome 22q11.2. Human Molecular Genetics, 2018, 27, 1150-1163.	2.9	22

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91	Mutational analysis of HOXA2 and SIX2 in a Bronx population with isolated microtia. International Journal of Pediatric Otorhinolaryngology, 2010, 74, 878-882.	1.0	21
92	GJB2 mutation spectrum in 209 hearing impaired individuals of predominantly Caribbean Hispanic and African descent. International Journal of Pediatric Otorhinolaryngology, 2010, 74, 611-618.	1.0	20
93	Overt cleft palate phenotype and <i>TBX1</i> genotype correlations in veloâ€cardioâ€facial/DiGeorge/22q11.2 deletion syndrome patients. American Journal of Medical Genetics, Part A, 2012, 158A, 2781-2787.	1.2	20
94	Variant discovery and breakpoint region prediction for studying the human 22q11.2 deletion using BAC clone and whole genome sequencing analysis. Human Molecular Genetics, 2016, 25, 3754-3767.	2.9	20
95	Optical mapping of the 22q11.2DS region reveals complex repeat structures and preferred locations for non-allelic homologous recombination (NAHR). Scientific Reports, 2020, 10, 12235.	3.3	20
96	Cleft palate, retrognathia and congenital heart disease in velo-cardio-facial syndrome: A phenotype correlation study. International Journal of Pediatric Otorhinolaryngology, 2011, 75, 1167-1172.	1.0	18
97	Whole-Genome Sequencing and Integrative Genomic Analysis Approach on Two 22q11.2 Deletion Syndrome Family Trios for Genotype to Phenotype Correlations. Human Mutation, 2015, 36, 797-807.	2.5	16
98	Dysregulation of TBX1 dosage in the anterior heart field results in congenital heart disease resembling the 22q11.2 duplication syndrome. Human Molecular Genetics, 2018, 27, 1847-1857.	2.9	16
99	Use of polyestradiol phosphate and anti-17β estradiol antibodies for the localization of estrogen receptors in target tissues: A critique. Cancer, 1980, 46, 2872-2879.	4.1	15
100	Isolation and Characterization of a Novel Gene Containing WD40 Repeats from the Region Deleted in Velo–cardio–facial/ DiGeorge Syndrome on Chromosome 22q11. Genomics, 2001, 73, 264-271.	2.9	15
101	Development of a Targeted Multi-Disorder High-Throughput Sequencing Assay for the Effective Identification of Disease-Causing Variants. PLoS ONE, 2015, 10, e0133742.	2.5	15
102	Gene-based genome-wide association studies and meta-analyses of conotruncal heart defects. PLoS ONE, 2019, 14, e0219926.	2.5	15
103	Identification of putative retinoic acid target genes downstream of mesenchymal <i>Tbx1</i> during inner ear development. Developmental Dynamics, 2012, 241, 563-573.	1.8	14
104	Genetic analysis of nonalcoholic fatty liver disease within a Caribbean–Hispanic population. Molecular Genetics & Genomic Medicine, 2015, 3, 558-569.	1.2	14
105	A candidate gene approach to identify modifiers of the palatal phenotype in 22q11.2 deletion syndrome patients. International Journal of Pediatric Otorhinolaryngology, 2013, 77, 123-127.	1.0	13
106	Expression of Cdcrel-1 (Pnutl1), a gene frequently deleted in velo-cardio-facial syndrome/DiGeorge syndrome. Mechanisms of Development, 2000, 96, 121-124.	1.7	12
107	Conditional and constitutive expression of a Tbx1-GFP fusion protein in mice. BMC Developmental Biology, 2013, 13, 33.	2.1	12
108	A normative chart for cognitive development in a genetically selected population. Neuropsychopharmacology, 2022, 47, 1379-1386.	5.4	12

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109	Gene targeting in mammalian cells by homologous recombination. Current Opinion in Biotechnology, 1993, 4, 577-582.	6.6	11
110	Genetic dosage compensation in a family with velo ardioâ€facial/DiGeorge/22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 548-554.	1.2	10
111	Molecular characterization of an interstitial deletion of 1p31.3 in a patient with obesity and psychiatric illness and a review of the literature. , 2011, 155, 825-832.		10
112	Biallelic expression of <i>Tbx1</i> protects the embryo from developmental defects caused by increased receptor tyrosine kinase signaling. Developmental Dynamics, 2012, 241, 1310-1324.	1.8	9
113	LPA receptor activity is basal specific and coincident with early pregnancy and involution during mammary gland postnatal development. Scientific Reports, 2016, 6, 35810.	3.3	9
114	Gene-based analyses of the maternal genome implicate maternal effect genes as risk factors for conotruncal heart defects. PLoS ONE, 2020, 15, e0234357.	2.5	8
115	Genetic evaluation of American minority pediatric cochlear implant recipients. International Journal of Pediatric Otorhinolaryngology, 2009, 73, 195-203.	1.0	7
116	<i>Tbx1</i> and <i>Jag1</i> act in concert to modulate the fate of neurosensory cells of the mouse otic vesicle. Biology Open, 2017, 6, 1472-1482.	1.2	7
117	Integrated rare variant-based risk gene prioritization in disease case-control sequencing studies. PLoS Genetics, 2017, 13, e1007142.	3.5	7
118	Atypical chromosome 22q11.2 deletions are complex rearrangements and have different mechanistic origins. Human Molecular Genetics, 2019, 28, 3724-3733.	2.9	7
119	Two Functional Copies of the <i>DGCR6</i> Gene Are Present on Human Chromosome 22q11 Due to a Duplication of an Ancestral Locus. Genome Research, 2001, 11, 208-217.	5.5	6
120	A Novel, Single Nucleotide Polymorphism-Based Assay to Detect 22q11 Deletions. Genetic Testing and Molecular Biomarkers, 2007, 11, 91-100.	1.7	5
121	Interstitial duplication of 22q13.2 in a girl with short stature, impaired speech and language, and dysmorphism. Journal of Pediatric Genetics, 2015, 01, 047-053.	0.7	5
122	NOTCH maintains developmental cardiac gene network through WNT5A. Journal of Molecular and Cellular Cardiology, 2018, 125, 98-105.	1.9	4
123	Copy number variations in individuals with conotruncal heart defects reveal some shared developmental pathways irrespective of 22q11.2 deletion status. Birth Defects Research, 2019, 111, 888-905.	1.5	3
124	The Prevalence of Ultrarapid Metabolizers of Codeine in a Diverse Urban Population. Otolaryngology - Head and Neck Surgery, 2019, 160, 420-425.	1.9	3
125	Crk and Crkl have shared functions in neural crest cells for cardiac outflow tract septation and vascular smooth muscle differentiation. Human Molecular Genetics, 2021, , .	2.9	3
126	Common Variation in Cytoskeletal Genes Is Associated with Conotruncal Heart Defects. Genes, 2021, 12, 655.	2.4	2

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127	Genome-Wide Association Studies of Conotruncal Heart Defects with Normally Related Great Vessels in the United States. Genes, 2021, 12, 1030.	2.4	1
128	A Tbx1-Six1/Eya1-Fgf8 genetic pathway controls mammalian cardiovascular and craniofacial morphogenesis. Journal of Clinical Investigation, 2011, 121, 2060-2060.	8.2	0
129	Chromosome 22q11.2 Rearrangement Disorders. , 2006, , 193-206.		0
130	Title is missing!. , 2020, 15, e0234357.		0
131	Title is missing!. , 2020, 15, e0234357.		0
132	Title is missing!. , 2020, 15, e0234357.		0
133	Title is missing!. , 2020, 15, e0234357.		0