Niels Tommerup

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8284135/publications.pdf

Version: 2024-02-01

277 papers	19,707 citations	20036 63 h-index	14779 131 g-index
283 all docs	283 docs citations	283 times ranked	29321 citing authors

#	Article	IF	Citations
1	Autosomal sex reversal and campomelic dysplasia are caused by mutations in and around the SRY-related gene SOX9. Cell, 1994, 79, 1111-1120.	13.5	1,417
2	Rubinstein-Taybi syndrome caused by mutations in the transcriptional co-activator CBP. Nature, 1995, 376, 348-351.	13.7	1,140
3	Chromosome instability and immunodeficiency syndrome caused by mutations in a DNA methyltransferase gene. Nature, 1999, 402, 187-191.	13.7	1,056
4	A human phenome-interactome network of protein complexes implicated in genetic disorders. Nature Biotechnology, 2007, 25, 309-316.	9.4	871
5	Ancient human genome sequence of an extinct Palaeo-Eskimo. Nature, 2010, 463, 757-762.	13.7	750
6	Isolation of a candidate gene for Menkes disease that encodes a potential heavy metal binding protein. Nature Genetics, 1993, 3, 14-19.	9.4	708
7	Direct Diagnosis by DNA Analysis of the Fragile X Syndrome of Mental Retardation. New England Journal of Medicine, 1991, 325, 1673-1681.	13.9	642
8	BAP1: a novel ubiquitin hydrolase which binds to the BRCA1 RING finger and enhances BRCA1-mediated cell growth suppression. Oncogene, 1998, 16, 1097-1112.	2.6	636
9	JARID2 regulates binding of the Polycomb repressive complexâ€‰2 to target genes in ES cells. Nature, 2010, 464, 306-310.	13.7	499
10	MicroRNA expression in the adult mouse central nervous system. Rna, 2008, 14, 432-444.	1.6	427
11	Deletions of a differentially methylated CpG island at the SNRPN gene define a putative imprinting control region. Nature Genetics, 1994, 8, 52-58.	9.4	418
12	Molecular Identification of a Novel Candidate Sorting Receptor Purified from Human Brain by Receptor-associated Protein Affinity Chromatography. Journal of Biological Chemistry, 1997, 272, 3599-3605.	1.6	368
13	Disruption of the Serine/Threonine Kinase 9 Gene Causes Severe X-Linked Infantile Spasms and Mental Retardation. American Journal of Human Genetics, 2003, 72, 1401-1411.	2.6	265
14	The phenotypic spectrum of SCN8A encephalopathy. Neurology, 2015, 84, 480-489.	1.5	246
15	Molecular Characterization of a Novel Human Hybrid-type Receptor That Binds the Î±2-Macroglobulin Receptor-associated Protein. Journal of Biological Chemistry, 1996, 271, 31379-31383.	1.6	224
16	Recurrent Reciprocal Genomic Rearrangements of 17q12 Are Associated with Renal Disease, Diabetes, and Epilepsy. American Journal of Human Genetics, 2007, 81, 1057-1069.	2.6	222
17	Detection of microRNAs in frozen tissue sections by fluorescence in situ hybridization using locked nucleic acid probes and tyramide signal amplification. Nature Protocols, 2007, 2, 2520-2528.	5.5	221
18	Obstetrical and gynecological complications in fragile X carriers: A multicenter study. American Journal of Medical Genetics Part A, 1994, 51, 400-402.	2.4	202

#	Article	IF	Citations
19	Assignment of an autosomal sex reversaâ€“ locus (SRA1) and campomelic dysplasia (CMPD1) to 17q24.3â€“q25.1. Nature Genetics, 1993, 4, 170-174.	9.4	196
20	Mutations inSYNGAP1Cause Intellectual Disability, Autism, and a Specific Form of Epilepsy by Inducing Haploinsufficiency. Human Mutation, 2013, 34, 385-394.	1.1	196
21	A Functional Link between the Histone Demethylase PHF8 and the Transcription Factor ZNF711 in X-Linked Mental Retardation. Molecular Cell, 2010, 38, 165-178.	4.5	186
22	Single-molecule denaturation mapping of DNA in nanofluidic channels. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 13294-13299.	3.3	183
23	Jarid1b targets genes regulating development and is involved in neural differentiation. EMBO Journal, 2011, 30, 4586-4600.	3.5	183
24	Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. Annals of Neurology, 2016, 79, 428-436.	2.8	159
25	The Human Intrinsic Factor-Vitamin B12 Receptor,Cubilin: Molecular Characterization and Chromosomal Mapping of the Gene to 10p Within the Autosomal Recessive Megaloblastic Anemia (MGA1) Region. Blood, 1998, 91, 3593-3600.	0.6	158
26	Autoimmune diseases in women with Turner's Syndrome. Arthritis and Rheumatism, 2010, 62, 658-666.	6.7	147
27	Acute and persistent symptoms in non-hospitalized PCR-confirmed COVID-19 patients. Scientific Reports, 2021, 11, 13153.	1.6	147
28	Truncation of the Down Syndrome Candidate Gene DYRK1A in Two Unrelated Patients with Microcephaly. American Journal of Human Genetics, 2008, 82, 1165-1170.	2.6	145
29	RESTâ€“Mediated Recruitment of Polycomb Repressor Complexes in Mammalian Cells. PLoS Genetics, 2012, 8, e1002494.	1.5	140
30	Duplications Involving a Conserved Regulatory Element Downstream of BMP2 Are Associated with Brachydactyly Type A2. American Journal of Human Genetics, 2009, 84, 483-492.	2.6	139
31	A balanced chromosomal translocation disruptingARHGEF9is associated with epilepsy, anxiety, aggression, and mental retardation. Human Mutation, 2009, 30, 61-68.	1.1	131
32	Corpus callosum abnormalities, intellectual disability, speech impairment, and autism in patients with haploinsufficiency of ARID1B. Clinical Genetics, 2012, 82, 248-255.	1.0	126
33	Linkage Mapping in 29 Bardetâ€“Biedl Syndrome Families Confirms Loci in Chromosomal Regions 11q13, 15q22.3â€“q23, and 16q21. Genomics, 1997, 41, 93-99.	1.3	125
34	Isolation and Fine Mapping of 16 Novel Human Zinc Finger-Encoding cDNAs Identify Putative Candidate Genes for Developmental and Malignant Disorders. Genomics, 1995, 27, 259-264.	1.3	123
35	Genetic and environmental risk factors in congenital heart disease functionally converge in protein networks driving heart development. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 14035-14040.	3.3	117
36	Mutations in autism susceptibility candidate 2 (AUTS2) in patients with mental retardation. Human Genetics, 2007, 121, 501-509.	1.8	116

#	Article	IF	Citations
37	High frequency of submicroscopic genomic aberrations detected by tiling path array comparative genome hybridisation in patients with isolated congenital heart disease. Journal of Medical Genetics, 2008, 45, 704-709.	1.5	110
38	Distinct CDH3 mutations cause ectodermal dysplasia, ectrodactyly, macular dystrophy (EEM) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 702	1.5	108
39	Gene Panel Testing in Epileptic Encephalopathies and Familial Epilepsies. Molecular Syndromology, 2016, 7, 210-219.	0.3	103
40	Haploinsufficiency of TAB2 Causes Congenital Heart Defects in Humans. American Journal of Human Genetics, 2010, 86, 839-849.	2.6	97
41	Haploinsufficiency of novel FOXG1B variants in a patient with severe mental retardation, brain malformations and microcephaly. Human Genetics, 2005, 117, 536-544.	1.8	96
42	The Genetic Basis of the Pierre Robin Sequence. Cleft Palate-Craniofacial Journal, 2006, 43, 155-159.	0.5	95
43	Pierre Robin sequence may be caused by dysregulation of SOX9 and KCNJ2. Journal of Medical Genetics, 2007, 44, 381-386.	1.5	91
44	Disruption of Netrin G1 by a balanced chromosome translocation in a girl with Rett syndrome. European Journal of Human Genetics, 2005, 13, 921-927.	1.4	90
45	Genetic Association Studies in Lumbar Disc Degeneration: A Systematic Review. PLoS ONE, 2012, 7, e49995.	1.1	90
46	Hypomorphic Mutations in PGAP2, Encoding a GPI-Anchor-Remodeling Protein, Cause Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2013, 92, 575-583.	2.6	87
47	Mutations in GABRB3. Neurology, 2017, 88, 483-492.	1.5	87
48	Novel Connexin 43 (GJA1) mutation causes oculo-dento-digital dysplasia with curly hair. American Journal of Medical Genetics Part A, 2004, 127A, 152-157.	2.4	86
49	The small RNA content of human sperm reveals pseudogene-derived piRNAs complementary to protein-coding genes. Rna, 2015, 21, 1085-1095.	1.6	83
50	Assignment of Human Elongation Factor 1Î± Genes:EEF1AMaps to Chromosome 6q14 andEEF1A2to 20q13.3. Genomics, 1996, 36, 359-361.	1.3	82
51	Comparative genomics beyond sequence-based alignments: RNA structures in the ENCODE regions. Genome Research, 2008, 18, 242-251.	2.4	82
52	Hedgehog signaling in small-cell lung cancer: Frequent in vivo but a rare event in vitro. Lung Cancer, 2006, 52, 281-290.	0.9	80
53	Dissecting spatioâ€temporal protein networks driving human heart development and related disorders. Molecular Systems Biology, 2010, 6, 381.	3.2	80
54	Genome-wide detection of chromosomal rearrangements, indels, and mutations in circular chromosomes by short read sequencing. Genome Research, 2011, 21, 1388-1393.	2.4	79

#	Article	IF	Citations
55	Non-random X chromosome inactivation in an affected twin in a monozygotic twin pair discordant for Wiedemann-Beckwith syndrome. American Journal of Medical Genetics Part A, 1995, 56, 210-214.	2.4	76
56	Exclusion of SNRPN as a major determinant of Prader-Willi syndrome by a translocation breakpoint. Nature Genetics, 1996, 12, 452-454.	9.4	74
57	X-Linked mental retardation with fragile X. a pedigree showing transmission by apparently unaffected males and partial expression in female carriers. Human Genetics, 1981, 59, 23-25.	1.8	71
58	Aberrant expression of miRâ€218 and miRâ€204 in human mesial temporal lobe epilepsy and hippocampal sclerosisâ€"Convergence on axonal guidance. Epilepsia, 2014, 55, 2017-2027.	2.6	71
59	The role of <scp>SLC</scp>2A1 mutations in myoclonic astatic epilepsy and absence epilepsy, and the estimated frequency of <scp>GLUT</scp>1 deficiency syndrome. Epilepsia, 2015, 56, e203-8.	2.6	71
60	The identification and functional annotation of RNA structures conserved in vertebrates. Genome Research, 2017, 27, 1371-1383.	2.4	71
61	A familial reciprocal translocation t(3;7) (p21.1;p13) associated with the Greig polysyndactyly-craniofacial anomalies syndrome. American Journal of Medical Genetics Part A, 1983, 16, 313-321.	2.4	68
62	Disruption of the CNTNAP2 gene in a t(7;15) translocation family without symptoms of Gilles de la Tourette syndrome. European Journal of Human Genetics, 2007, 15, 711-713.	1.4	68
63	Genome-wide Analysis of CDX2 Binding in Intestinal Epithelial Cells (Caco-2). Journal of Biological Chemistry, 2010, 285, 25115-25125.	1.6	68
64	Isolated and syndromic forms of congenital anosmia. Clinical Genetics, 2012, 81, 210-215.	1.0	66
65	Nationwide germline whole genome sequencing of 198 consecutive pediatric cancer patients reveals a high incidence of cancer prone syndromes. PLoS Genetics, 2020, 16, e1009231.	1.5	64
66	Filter-grown TR146 cells as an in vitro model of human buccal epithelial permeability. European Journal of Oral Sciences, 1999, 107, 138-146.	0.7	63
67	Mutations in NRXN1 in a family multiply affected with brain disorders: NRXN1 mutations and brain disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 354-358.	1.1	63
68	MECP2 mutations in Danish patients with Rett syndrome: High frequency of mutations but no consistent correlations with clinical severity or with the X chromosome inactivation pattern. European Journal of Human Genetics, 2001, 9, 178-184.	1.4	61
69	Ciao 1 Is a Novel WD40 Protein That Interacts with the Tumor Suppressor Protein WT1. Journal of Biological Chemistry, 1998, 273, 10880-10887.	1.6	60
70	Translocations Disrupting PHF21A in the Potocki-Shaffer-Syndrome Region Are Associated with Intellectual Disability and Craniofacial Anomalies. American Journal of Human Genetics, 2012, 91, 56-72.	2.6	59
71	Exonâ€disrupting deletions of <scp>NRXN1</scp> in idiopathic generalized epilepsy. Epilepsia, 2013, 54, 256-264.	2.6	59
72	FISHing with locked nucleic acids (LNA): evaluation of different LNA/DNA mixmers. Molecular and Cellular Probes, 2003, 17, 165-169.	0.9	56

#	Article	IF	Citations
73	An excess of chromosome 1 breakpoints in male infertility. European Journal of Human Genetics, 2004, 12, 993-1000.	1.4	56
74	Molecular cytogenetic characterization of ring chromosome 15 in three unrelated patients. , 2004, 130A, 340-344.		56
75	Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. Human Genetics, 2006, 118, 578-590.	1.8	55
76	Additional chromosomal abnormalities in patients with a previously detected abnormal karyotype, mental retardation, and dysmorphic features. American Journal of Medical Genetics, Part A, 2006, 140A, 2180-2187.	0.7	54
77	Profiling microRNAs in lung tissue from pigs infected with Actinobacillus pleuropneumoniae. BMC Genomics, 2012, 13, 459.	1.2	54
78	Marker X chromosome induction in fibroblasts by FUdR. American Journal of Medical Genetics Part A, 1981, 9, 263-264.	2.4	53
79	Preaxial polydactyly/triphalangeal thumb is associated with changed transcription factor-binding affinity in a family with a novel point mutation in the long-range cis-regulatory element ZRS. European Journal of Human Genetics, 2010, 18, 733-736.	1.4	53
80	Psoriasis Upregulated Phorbolin-1 Shares Structural but not Functional Similarity to the mRNA-Editing Protein Apobec-1. Journal of Investigative Dermatology, 1999, 113, 162-169.	0.3	51
81	LNA-modified oligonucleotides are highly efficient as FISH probes. Cytogenetic and Genome Research, 2004, 107, 32-37.	0.6	51
82	Nextâ€generation sequencing: proof of concept for antenatal prediction of the fetal <scp>K</scp>ell blood group phenotype from cellâ€free fetal <scp>DNA</scp> in maternal plasma. Transfusion, 2013, 53, 2892-2898.	0.8	51
83	Reduced ceramide synthase 2 activity causes progressive myoclonic epilepsy. Annals of Clinical and Translational Neurology, 2014, 1, 88-98.	1.7	50
84	Germline Chromothripsis Driven by L1-Mediated Retrotransposition and Alu/Alu Homologous Recombination. Human Mutation, 2016, 37, 385-395.	1.1	50
85	Macroorchidism and fragile X in mentally retarded males. Human Genetics, 1982, 61, 113-117.	1.8	49
86	A neocentromere on human chromosome 3 without detectable Î±-satellite DNA forms morphologically normal kinetochores. Chromosoma, 1998, 107, 359-365.	1.0	49
87	A zinc-finger gene ZNF141 mapping at 4p16.3/D4S90 is a candidate gene for the Wolf-Hirschhorn (4p-) syndrome. Human Molecular Genetics, 1993, 2, 1571-1575.	1.4	48
88	Mapping of the Human PAWR (par-4) Gene to Chromosome 12q21. Genomics, 1998, 53, 241-243.	1.3	48
89	Genetic instability of cell lines derived from a single human small cell carcinoma of the lung. European Journal of Cancer & Clinical Oncology, 1985, 21, 815-824.	0.9	47
90	The strength of combined cytogenetic and mate-pair sequencing techniques illustrated by a germline chromothripsis rearrangement involving FOXP2. European Journal of Human Genetics, 2014, 22, 338-343.	1.4	46

#	Article	IF	Citations
91	High resolution comparative genomic hybridisation analysis reveals imbalances in dyschromosomal patients with normal or apparently balanced conventional karyotypes. European Journal of Human Genetics, 2000, 8, 661-668.	1.4	45
92	Dysfunction of the Heteromeric KV7.3/KV7.5 Potassium Channel is Associated with Autism Spectrum Disorders. Frontiers in Genetics, 2013, 4, 54.	1.1	45
93	CHEMOATTRACTION INTETRAHYMENA: ON THE ROLE OF CHEMOKINESIS. Biological Bulletin, 1986, 170, 357-367.	0.7	44
94	Tentative assignment of a locus for Rubinstein-Taybi syndrome to 16p13.3 by a de novo reciprocal translocation, t(7;16)(q34;p13.3). American Journal of Medical Genetics Part A, 1992, 44, 237-241.	2.4	44
95	Breakpoints around the HOXD cluster result in various limb malformations. Journal of Medical Genetics, 2005, 43, 111-118.	1.5	44
96	Mutational analysis of the human FATE gene in 144 infertile men. Human Genetics, 2003, 113, 195-201.	1.8	41
97	Genetic heterogeneity in Pakistani microcephaly families. Clinical Genetics, 2013, 83, 446-451.	1.0	41
98	Sequence assembly. Computational Biology and Chemistry, 2009, 33, 121-136.	1.1	39
99	Identification of human candidate genes for male infertility by digital differential display. Molecular Human Reproduction, 2001, 7, 11-20.	1.3	38
100	Interstitial deletion 9q22.32-q33.2 associated with additional familial translocation t(9;17)(q34.11;p11.2) in a patient with Gorlin-Goltz syndrome and features of Nail-Patella syndrome. , 2004, 124A, 179-191.		38
101	Non-disjunction of chromosome 13. Human Molecular Genetics, 2007, 16, 2004-2010.	1.4	38
102	Deletion of 7q34â€“q36.2 in two siblings with mental retardation, language delay, primary amenorrhea, and dysmorphic features. American Journal of Medical Genetics, Part A, 2010, 152A, 3115-3119.	0.7	37
103	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. European Journal of Human Genetics, 2016, 24, 1761-1770.	1.4	36
104	Sequence Variants in the Human Cocaine and Amphetamineâ€Regulated Transcript (CART) Gene in Subjects with Early Onset Obesity. Obesity, 1999, 7, 532-536.	4.0	35
105	Tetrasomy 18p de novo: Parental Origin and Different Mechanisms of Formation. European Journal of Human Genetics, 1996, 4, 160-167.	1.4	35
106	Interstitial deletion 13q. Further delineation of the syndrome by clinical and high-resolution chromosome analysis of five patients. American Journal of Medical Genetics Part A, 1988, 29, 739-753.	2.4	34
107	Characterization of a 1.0 Mb YAC contig spannning two chromosome breakpoints related to Menkes disease. Human Molecular Genetics, 1992, 1, 483-489.	1.4	34
108	Haploinsufficiency of CELF4 at 18q12.2 is associated with developmental and behavioral disorders, seizures, eye manifestations, and obesity. European Journal of Human Genetics, 2012, 20, 1315-1319.	1.4	34

#	Article	IF	Citations
109	RRP7A links primary microcephaly to dysfunction of ribosome biogenesis, resorption of primary cilia, and neurogenesis. Nature Communications, 2020, 11, 5816.	5.8	34
110	Small supernumerary marker chromosomes: A legacy of trisomy rescue?. Human Mutation, 2019, 40, 193-200.	1.1	33
111	The human hedgehog-interacting protein gene: Structure and chromosome mapping to 4q31.21â†'q31.3. Cytogenetic and Genome Research, 2001, 92, 300-303.	0.6	32
112	Delineation of an interstitial 9q22 deletion in basal cell nevus syndrome. , 2005, 132A, 324-328.		32
113	Congenital olfactory impairment is linked to cortical changes in prefrontal and limbic brain regions. Brain Imaging and Behavior, 2018, 12, 1569-1582.	1.1	32
114	Compound heterozygous ASPM mutations in Pakistani MCPH families. American Journal of Medical Genetics, Part A, 2009, 149A, 926-930.	0.7	31
115	The myosin chaperone UNC45B is involved in lens development and autosomal dominant juvenile cataract. European Journal of Human Genetics, 2014, 22, 1290-1297.	1.4	31
116	Accurate Breakpoint Mapping in Apparently Balanced Translocation Families with Discordant Phenotypes Using Whole Genome Mate-Pair Sequencing. PLoS ONE, 2017, 12, e0169935.	1.1	31
117	HOXD13 polyalanine tract expansion in classical synpolydactyly type Vordingborg. American Journal of Medical Genetics Part A, 2002, 110, 116-121.	2.4	30
118	A mutation in the receptor binding site of GDF5 causes Mohr-Wriedt brachydactyly type A2. Journal of Medical Genetics, 2005, 43, 225-231.	1.5	30
119	Metaphase FISH on a Chip: Miniaturized Microfluidic Device for Fluorescence in situ Hybridization. Sensors, 2010, 10, 9831-9846.	2.1	30
120	Genetic linkage of autosomal dominant primary open angle glaucoma to chromosome 3q in a Greek pedigree. European Journal of Human Genetics, 2001, 9, 452-457.	1.4	29
121	Delineation of a 2.2 Mb microdeletion at 5q35 associated with microcephaly and congenital heart disease. American Journal of Medical Genetics, Part A, 2006, 140A, 427-433.	0.7	29
122	Identification of the BRD1 interaction network and its impact on mental disorder risk. Genome Medicine, 2016, 8, 53.	3.6	29
123	Risks and Recommendations in Prenatally Detected De Novo Balanced Chromosomal Rearrangements from Assessment of Long-Term Outcomes. American Journal of Human Genetics, 2018, 102, 1090-1103.	2.6	29
124	Structural genomic variation in childhood epilepsies with complex phenotypes. European Journal of Human Genetics, 2014, 22, 896-901.	1.4	28
125	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. PLoS Genetics, 2018, 14, e1007780.	1.5	28
126	Screening of congenital heart disease patients using multiplex ligationâ€dependent probe amplification: Early diagnosis of syndromic patients. American Journal of Medical Genetics, Part A, 2012, 158A, 720-725.	0.7	27

#	Article	IF	Citations
127	Genome-wide DNA methylation analysis of transient neonatal diabetes type 1 patients with mutations in ZFP57. BMC Medical Genetics, 2016, 17, 29.	2.1	27
128	Localization of the Human Gene for Advanced Glycosylation End Product-Specific Receptor (AGER) to Chromosome 6p21.3. Genomics, 1994, 24, 606-608.	1.3	26
129	Human FATE is a novel X-linked gene expressed in fetal and adult testis. Molecular and Cellular Endocrinology, 2001, 184, 25-32.	1.6	26
130	The Hedgehog signaling pathway â€“ implications for drug targets in cancer and neurodegenerative disorders. Pharmacogenomics, 2003, 4, 411-429.	0.6	26
131	A balanced translocation disruptsSYNGAP1in a patient with intellectual disability, speech impairment, and epilepsy with myoclonic absences (EMA). Epilepsia, 2011, 52, e190-e193.	2.6	26
132	Neurodevelopmental disorders associated with dosage imbalance ofZBTB20correlate with the morbidity spectrum of ZBTB20 candidate target genes. Journal of Medical Genetics, 2014, 51, 605-613.	1.5	26
133	Investigation of deletions at 7q11.23 in 44 patients referred for Williams-Beuren syndrome, using FISH and four DNA polymorphisms. Human Genetics, 1996, 99, 56-61.	1.8	25
134	Subtelomeric study of 132 patients with mental retardation reveals 9 chromosomal anomalies and contributes to the delineation of submicroscopic deletions of 1pter, 2qter, 4pter, 5qter and 9qter. BMC Medical Genetics, 2005, 6, 21.	2.1	25
135	Expression analyses of human cleft palate tissue suggest a role for osteopontin and immune related factors in palatal development. Experimental and Molecular Medicine, 2009, 41, 77.	3.2	25
136	The first mutation in <scp>CNGA2</scp> in two brothers with anosmia. Clinical Genetics, 2015, 88, 293-296.	1.0	25
137	Prader-Willi syndrome in a brother and sister without cytogenetic or detectable molecular genetic abnormality at chromosome 15q11q13. American Journal of Medical Genetics Part A, 1992, 44, 534-538.	2.4	24
138	Screening of 99 Danish Patients with Congenital Heart Disease for GATA4 Mutations. Genetic Testing and Molecular Biomarkers, 2006, 10, 277-280.	1.7	24
139	High frequency of rare copy number variants affecting functionally related genes in patients with structural brain malformations. Human Mutation, 2011, 32, 1427-1435.	1.1	24
140	Breakpoint Cloning and Haplotype Analysis Indicate a Single Origin of the Common Inv(10)(p11.2q21.2) Mutation among Northern Europeans. American Journal of Human Genetics, 2006, 78, 878-883.	2.6	23
141	Regional differences in expression of specific markers for human embryonic stem cells. Reproductive BioMedicine Online, 2007, 15, 89-98.	1.1	23
142	Global gene expression analysis in fetal mouse ovaries with and without meiosis and comparison of selected genes with meiosis in the testis. Cell and Tissue Research, 2007, 328, 207-221.	1.5	23
143	De novo unbalanced translocations have a complex history/aetiology. Human Genetics, 2018, 137, 817-829.	1.8	23
144	Assignment of Human KH-Box-Containing Genes byin SituHybridization:HNRNPKMaps to 9q21.32â€“q21.33,PCBP1to 2p12â€“p13, andPCBP2to 12q13.12â€“q13.13, Distal toFRA12A. Genomics, 1996, 32, 297-298.	2.3	22

#	Article	IF	Citations
145	Identification and Characterization of an Inner Ear-Expressed Human Melanoma Inhibitory Activity (MIA)-like Gene (MIAL) with a Frequent Polymorphism That Abolishes Translation. Genomics, 2001, 71, 40-52.	1.3	22
146	Assignment of the Human Genes Encoding 14-3-3 Eta (YWHAH) to 22q12, 14-3-3 Zeta (YWHAZ) to 2p25.1â€“p25.2, and 14-3-3 Beta (YWHAB) to 20q13.1 byin SituHybridization. Genomics, 1996, 33, 149-150.	1.3	21
147	Occurrence of Cancer in a Cohort of 183 Persons with Constitutional Chromosome 7 Abnormalities. Cancer Genetics and Cytogenetics, 1998, 105, 39-42.	1.0	21
148	A 72-year-old Danish puzzle resolvedâ€”comparative analysis of phenotypes in families with different-sizedHOXD13 polyalanine expansions. American Journal of Medical Genetics, Part A, 2005, 138A, 328-339.	0.7	20
149	Characterization of a t(5;8)(q31;q21) translocation in a patient with mental retardation and congenital heart disease: implications for involvement of RUNX1T1 in human brain and heart development. European Journal of Human Genetics, 2009, 17, 1010-1018.	1.4	20
150	Stones in the lacrimal gland: a rare condition. Acta Ophthalmologica, 2009, 87, 672-675.	0.6	20
151	Cost-effective multiplexing before capture allows screening of 25â€‰000 clinically relevant SNPs in childhood acute lymphoblastic leukemia. Leukemia, 2011, 25, 1001-1006.	3.3	20
152	Neural correlates of taste perception in congenital olfactory impairment. Neuropsychologia, 2014, 62, 297-305.	0.7	20
153	Regulatory variants of FOXG1 in the context of its topological domain organisation. European Journal of Human Genetics, 2018, 26, 186-196.	1.4	20
154	A mutation in the FOXE3 gene causes congenital primary aphakia in an autosomal recessive consanguineous Pakistani family. Molecular Vision, 2010, 16, 549-55.	1.1	20
155	SCREENING TEST FOR ATAXIA TELANGIECTASIA. Lancet, The, 1987, 330, 1398-1399.	6.3	19
156	Fine mapping of a de novo interstitial 10q22â€“q23 duplication in a patient with congenital heart disease and microcephaly. European Journal of Medical Genetics, 2008, 51, 81-86.	0.7	19
157	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. Human Molecular Genetics, 2014, 23, 6163-6176.	1.4	19
158	Very short DNA segments can be detected and handled by the repair machinery during germline chromothriptic chromosome reassembly. Human Mutation, 2018, 39, 709-716.	1.1	19
159	Craniosynostosisâ€microcephaly with chromosomal breakage and other abnormalities is caused by a truncating MCPH1 mutation and is allelic to premature chromosomal condensation syndrome and primary autosomal recessive microcephaly type 1. American Journal of Medical Genetics, Part A, 2010, 152A, 495-497.	0.7	18
160	A Cohort of Balanced Reciprocal Translocations Associated with Dyslexia: Identification of Two Putative Candidate Genes at DYX1. Behavior Genetics, 2011, 41, 125-133.	1.4	18
161	Validation of Genome-Wide Intervertebral Disk Calcification Associations in Dachshund and Further Investigation of the Chromosome 12 Susceptibility Locus. Frontiers in Genetics, 2012, 3, 225.	1.1	18
162	A Novel Locus Harbouring a Functional CD164 Nonsense Mutation Identified in a Large Danish Family with Nonsyndromic Hearing Impairment. PLoS Genetics, 2015, 11, e1005386.	1.5	18

#	Article	IF	Citations
163	Optical mapping of single-molecule human DNA in disposable, mass-produced all-polymer devices. Journal of Micromechanics and Microengineering, 2015, 25, 105002.	1.5	18
164	Chromothripsis and DNA Repair Disorders. Journal of Clinical Medicine, 2020, 9, 613.	1.0	18
165	Molecular cytogenetic detection of 9q34 breakpoints associated with nail patella syndrome. European Journal of Human Genetics, 1999, 7, 68-76.	1.4	17
166	Genomic structure, chromosome mapping and expression analysis of the human AVIL gene, and its exclusion as a candidate for locus for inflammatory bowel disease at 12q13â€“14 (IBD2). Gene, 2002, 288, 179-185.	1.0	17
167	Systematic re-examination of carriers of balanced reciprocal translocations: a strategy to search for candidate regions for common and complex diseases. European Journal of Human Genetics, 2006, 14, 410-417.	1.4	17
168	Investigation of 4qâ€deletion in two unrelated patients using array CGH. American Journal of Medical Genetics, Part A, 2008, 146A, 2431-2434.	0.7	17
169	X-linked congenital ptosis and associated intellectual disability, short stature, microcephaly, cleft palate, digital and genital abnormalities define novel Xq25q26 duplication syndrome. Human Genetics, 2014, 133, 625-638.	1.8	17
170	No evidence for pathogenic variants or maternal effect of ZFP57 as the cause of Beckwithâ€“Wiedemann Syndrome. European Journal of Human Genetics, 2012, 20, 119-121.	1.4	16
171	Complete re-sequencing of a 2Mb topological domain encompassing the FTO/IRXB genes identifies a novel obesity-associated region upstream of IRX5. Genome Medicine, 2015, 7, 126.	3.6	16
172	A novel splice site mutation in CEP135 is associated with primary microcephaly in a Pakistani family. Journal of Human Genetics, 2016, 61, 271-273.	1.1	16
173	Second trimester prenatal diagnosis of the fragile X. American Journal of Medical Genetics Part A, 1986, 23, 313-324.	2.4	15
174	Cytogenetically invisible microdeletions involving PITX2 in Rieger syndrome. Clinical Genetics, 2007, 72, 464-470.	1.0	15
175	A 3.2â€‰Mb deletion on 18q12 in a patient with childhood autism and high-grade myopia. European Journal of Human Genetics, 2008, 16, 312-319.	1.4	15
176	Mowatâ€“Wilson syndrome: an underdiagnosed syndrome?. Clinical Genetics, 2008, 73, 579-584.	1.0	15
177	Phenotypic subregions within the split-hand/foot malformation 1 locus. Human Genetics, 2016, 135, 345-357.	1.8	15
178	Macroorchidism, Mental Retardation, and the Fragile X. New England Journal of Medicine, 1981, 305, 1348-1348.	13.9	14
179	Apparent homozygosity for the fragile site at Xq28 in a normal female. Human Genetics, 1982, 61, 60-62.	1.8	14
180	Chromosomal breakage, endomitosis, endoreduplication, and hypersensitivity toward radiomimetric and alkylating agents: A possible new autosomal recessive mutation in a girl with craniosynostosis and microcephaly. Human Genetics, 1993, 92, 339-346.	1.8	14

#	Article	IF	Citations
181	A folate sensitive heritable fragile site at 19p13. Clinical Genetics, 1985, 27, 510-514.	1.0	14
182	Position effect, cryptic complexity, and direct gene disruption as disease mechanisms in de novo apparently balanced translocation cases. PLoS ONE, 2018, 13, e0205298.	1.1	14
183	Human CCS gene: genomic organization and exclusion as a candidate for amyotrophic lateral sclerosis (ALS). BMC Genetics, 2002, 3, 5.	2.7	13
184	Population-based study of cancer among carriers of a constitutional structural chromosomal rearrangement. Genes Chromosomes and Cancer, 2006, 45, 231-246.	1.5	13
185	Partial deletion 11q: report of a case with a large terminal deletion 11q21â€qter without loss of telomeric sequences, and review of the literature. Clinical Genetics, 1995, 47, 231-235.	1.0	13
186	The role of SLC2A1 in early onset and childhood absence epilepsies. Epilepsy Research, 2013, 105, 229-233.	0.8	13
187	Abdominal Wall Defects in Greenland 1989â€“2015. Birth Defects Research, 2017, 109, 836-842.	0.8	13
188	Genome-Wide Supported Risk Variants inMIR137,CACNA1C,CSMD1,DRD2, andGRM3Contribute to Schizophrenia Susceptibility in Pakistani Population. Psychiatry Investigation, 2017, 14, 687.	0.7	13
189	Detection of illegitimate rearrangement within the immunoglobulin locus on 14q32.3 in B-cell malignancies using end-sequenced probes. Genes Chromosomes and Cancer, 2001, 32, 265-274.	1.5	12
190	A novel mutation in IRF6 resulting in VWSâ€“PPS spectrum disorder with renal aplasia. American Journal of Medical Genetics, Part A, 2008, 146A, 1605-1608.	0.7	12
191	Genetic studies in congenital anterior midline cervical cleft. American Journal of Medical Genetics, Part A, 2012, 158A, 2021-2026.	0.7	12
192	Two rare deletions upstream of the NRXN1 gene (2p16.3) affecting the non-coding mRNA AK127244 segregate with diverse psychopathological phenotypes in a family. European Journal of Medical Genetics, 2015, 58, 650-653.	0.7	12
193	Challenges for the Sustainability of University-Run Biobanks. Biopreservation and Biobanking, 2018, 16, 312-321.	0.5	12
194	Haploinsufficiency of ARHGAP42 is associated with hypertension. European Journal of Human Genetics, 2019, 27, 1296-1303.	1.4	12
195	Hydatidiform moles: Methods for culture and cytogenetic analyses. Cancer Genetics and Cytogenetics, 1986, 22, 19-27.	1.0	11
196	Human rab11a: transcription, chromosome mapping and effect on the expression levels of host GTP-binding proteins. FEBS Letters, 1998, 429, 359-364.	1.3	11
197	Genetic counseling in adult carriers of a balanced chromosomal rearrangement ascertained in childhood: Experiences from a nationwide reexamination of translocation carriers. Genetics in Medicine, 2007, 9, 185-187.	1.1	11
198	Folic acid metabolism in patient with fragile X. Clinical Genetics, 1983, 24, 153-155.	1.0	11

#	Article	IF	Citations
199	GLI1 Is Involved in Cell Cycle Regulation and Proliferation of NT2 Embryonal Carcinoma Stem Cells. DNA and Cell Biology, 2008, 27, 251-256.	0.9	11
200	Sequence analysis of 17 NRXN1 deletions. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 52-61.	1.1	11
201	Cryptic breakpoint identified by whole-genome mate-pair sequencing in a rare paternally inherited complex chromosomal rearrangement. Molecular Cytogenetics, 2018, 11, 34.	0.4	11
202	EXPRESSION AND POST-TRANSLATIONAL MODIFICATION OF HUMAN 4-HYDROXY-PHENYLPYRUVATE DIOXYGENASE. Cell Biology International, 2002, 26, 615-625.	1.4	10
203	4q35 deletion and 10p15 duplication associated with immunodeficiency. American Journal of Medical Genetics, Part A, 2006, 140A, 2231-2235.	0.7	10
204	Interstitial deletion of chromosome 4p associated with mild mental retardation, epilepsy and polymicrogyria of the left temporal lobe. Clinical Genetics, 2007, 72, 593-598.	1.0	10
205	Balanced translocation in a patient with severe myoclonic epilepsy of infancy disrupts the sodium channel gene SCN1A. Epilepsia, 2008, 49, 1091-1094.	2.6	10
206	A novel mutation in CDK5RAP2 gene causes primary microcephaly with speech impairment and sparse eyebrows in a consanguineous Pakistani family. European Journal of Medical Genetics, 2017, 60, 627-630.	0.7	10
207	Suggestive linkage to a neighboring region of IRF6 in a cleft lip and palate multiplex family. American Journal of Medical Genetics, Part A, 2007, 143A, 2716-2721.	0.7	9
208	Duplication of MAOA, MAOB, and NDP in a patient with mental retardation and epilepsy. European Journal of Human Genetics, 2011, 19, 1-2.	1.4	9
209	Chromosomal studies in familial polyposis coli. Cancer Genetics and Cytogenetics, 1985, 17, 355-357.	1.0	8
210	Chemotaxis in Tetrahymena. European Journal of Protistology, 1990, 25, 229-233.	0.5	8
211	Molecular characterization of a balanced chromosome translocation in psoriasis vulgaris. Clinical Genetics, 2005, 69, 189-193.	1.0	8
212	A novel primate specific gene, CEI, is located in the homeobox gene IRXA2 promoter in Homo sapiens. Gene, 2006, 371, 167-173.	1.0	8
213	Autoimmune diseases in a Danish cohort of 4,866 carriers of constitutional structural chromosomal rearrangements. Arthritis and Rheumatism, 2007, 56, 2402-2409.	6.7	8
214	A novel subtype of distal symphalangism affecting only the 4th finger. American Journal of Medical Genetics, Part A, 2009, 149A, 1571-1573.	0.7	8
215	Dysregulation of FOXG1 by ring chromosome 14. Molecular Cytogenetics, 2015, 8, 24.	0.4	8
216	Partial USH2A deletions contribute to Usher syndrome in Denmark. European Journal of Human Genetics, 2015, 23, 1646-1651.	1.4	8

#	Article	IF	Citations
217	Breakpoint mapping and haplotype analysis of translocation t(1;12)(q43;q21.1) in two apparently independent families with vascular phenotypes. Molecular Genetics & Genomic Medicine, 2018, 6, 56-68.	0.6	8
218	High resolution chromosomes from first trimester trophoblast cultures. Prenatal Diagnosis, 1985, 5, 291-294.	1.1	7
219	Detection of illegitimate rearrangements within the immunoglobulin light chain loci in B cell malignancies using end sequenced probes. Leukemia, 2002, 16, 2148-2155.	3.3	7
220	Identification of a NovelEYA1Splice-Site Mutation in a Danish Branchio-Oto-Renal Syndrome Family. Genetic Testing and Molecular Biomarkers, 2004, 8, 404-406.	1.7	7
221	Male-to-male transmission in Laurin-Sandrow syndrome and exclusion ofRARBandRARG. American Journal of Medical Genetics, Part A, 2005, 137A, 148-152.	0.7	7
222	Biparental inheritance of chromosomal abnormalities in male twins with non-syndromic mental retardation. European Journal of Medical Genetics, 2011, 54, e383-e388.	0.7	7
223	Automation of a single-DNA molecule stretching device. Review of Scientific Instruments, 2015, 86, 063702.	0.6	7
224	Monozygotic twins discordant for narcolepsy type 1 and multiple sclerosis. Neurology: Neuroimmunology and NeuroInflammation, 2016, 3, e249.	3.1	7
225	Interstitial deletion of 14q24.3â€q32.2 in a male patient with plagiocephaly, BPES features, developmental delay, and congenital heart defects. American Journal of Medical Genetics, Part A, 2011, 155, 203-206.	0.7	6
226	500K SNP array analyses in blood and saliva showed no differences in a pair of monozygotic twins discordant for cleft lip. American Journal of Medical Genetics, Part A, 2011, 155, 652-655.	0.7	6
227	The segregation of different submicroscopic imbalances underlying the clinical variability associated with a familial karyotypically balanced translocation. Molecular Cytogenetics, 2015, 8, 106.	0.4	6
228	Fragile X demonstrated retrospectively in amniotic cells cultured in low folate medium. Prenatal Diagnosis, 1983, 3, 367-369.	1.1	5
229	Identification of triploidy by DA/DAPI staining of trophoblastic interphase nuclei. Placenta, 1985, 6, 363-367.	0.7	5
230	Screening for the fragile X: How many cells should we analyse?. American Journal of Medical Genetics Part A, 1988, 30, 417-422.	2.4	5
231	Induction of the fragile X on BrdU-substituted chromosomes with direct visualization of sister chromatid exchanges on banded chromosomes. Human Genetics, 1989, 81, 377-381.	1.8	5
232	Prolonged extreme thrombocytosis associated with neurofibromatosis type 1. Journal of Pediatrics, 1997, 130, 317-319.	0.9	5
233	Prader-Willi-like phenotype and the proximal long arm of the X chromosome. American Journal of Medical Genetics Part A, 1998, 80, 300-301.	2.4	5
234	Molecular cloning of Xp11 breakpoints in two unrelated mentally retarded females with X;autosome translocations. Cytogenetic and Genome Research, 2000, 90, 126-133.	0.6	5

#	Article	IF	Citations
235	Evaluation of two methods for generating cRNA for microarray experiments from nanogram amounts of total RNA. Analytical Biochemistry, 2006, 358, 111-119.	1.1	5
236	Sequence and expression analysis of gaps in human chromosome 20. Nucleic Acids Research, 2012, 40, 6660-6672.	6.5	5
237	Enrichment of megabase-sized DNA molecules for single-molecule optical mapping and next-generation sequencing. Scientific Reports, 2017, 7, 17893.	1.6	5
238	A GDF5 frameshift mutation segregating with Grebe type chondrodysplasia and brachydactyly type C+ in a 6 generations family: Clinical report and mini review. European Journal of Medical Genetics, 2021, 64, 104226.	0.7	5
239	Assignment<footref rid="foot01">¹</footref> of the human zinc finger gene, ZNF288, to chromosome 3 band q13.2 by radiation hybrid mapping and fluorescence in situ hybridisation. Cytogenetic and Genome Research, 2000, 89, 156-157.	0.6	4
240	Mutation analysis of the Sonic hedgehog promoter and putative enhancer elements in Parkinson's disease patients. Molecular Brain Research, 2004, 126, 207-211.	2.5	4
241	Eponymous Jacobsen syndrome: Mapping the breakpoints of the original family suggests an association between the distal 1.1 Mb of chromosome 21 and osteoporosis in Down syndrome. American Journal of Medical Genetics, Part A, 2005, 135A, 339-341.	0.7	4
242	Localization in man of fifteen DNA sequences within the chromosome segment 13q12-q22. Hereditas, 1989, 110, 253-265.	0.5	4
243	Molecular characterization of two patients with de novo interstitial deletions in 4q22â€“q24. American Journal of Medical Genetics, Part A, 2009, 149A, 1830-1833.	0.7	4
244	Genomeâ€wide Gene Expression Profiling of SCID Mice with Tâ€cellâ€mediated Colitis. Scandinavian Journal of Immunology, 2009, 69, 437-446.	1.3	4
245	The lrrk2 p.Gly2019Ser mutation is uncommon in a Danish cohort with various neurodegenerative disorders. Parkinsonism and Related Disorders, 2011, 17, 398-399.	1.1	4
246	Autozygosity mapping of a large consanguineous Pakistani family reveals a novel non-syndromic autosomal recessive mental retardation locus on 11p15-tel. Neurogenetics, 2011, 12, 247-251.	0.7	4
247	An association study between the norepinephrine transporter gene and depression. Psychiatric Genetics, 2013, 23, 217-221.	0.6	4
248	Homozygous mutation in the NPHP3 gene causing foetal nephronophthisis. Nephrology, 2017, 22, 818-820.	0.7	4
249	Multigenic truncation of the semaphorinâ€“plexin pathway by a germline chromothriptic rearrangement associated with Moebius syndrome. Human Mutation, 2019, 40, 1057-1062.	1.1	4
250	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies syndrome due to disruption of BPTF in a 35â€yearâ€old man initially diagnosed with Silverâ€Russell syndrome. Clinical Genetics, 2019, 95, 534-536.	1.0	4
251	Specfic staining of 9h in human somatic interphase cells by D 287/170. Human Genetics, 1982, 62, 301-304.	1.8	3
252	DNA-aneuploidy in 46,XX hydatidiform moles. Cancer Genetics and Cytogenetics, 1987, 27, 225-228.	1.0	3

#	Article	IF	Citations
253	Assignment of the Human Gene for Oct-Binding Factor-1 (OBF1), a B-Cell-Specific Coactivator of Octamer-Binding Transcription Factors 1 and 2, to 11q23.1 by Somatic Cell Hybridization andin SituHybridization. Genomics, 1996, 33, 143-145.	1.3	3
254	Isolation of the Human Beaded-Filament Structural Protein 1 Gene (BFSP1) and Assignment to Chromosome 20p11.23â€“p12.1. Genomics, 1998, 53, 114-116.	1.3	3
255	A shared somatic translocation involving CUX1 in monozygotic twins as an early driver of AMKL in Down syndrome. Blood Cancer Journal, 2020, 10, 27.	2.8	3
256	Does rapid sequence divergence preclude RNA structure conservation in vertebrates?. Nucleic Acids Research, 2022, 50, 2452-2463.	6.5	3
257	No mutations found by RET mutation scanning in sporadic and hereditary neuroblastoma. Human Genetics, 1996, 97, 362-364.	1.8	3
258	Blood Group Substances, T6 Antigen and Heterochromatin Pattern as Species Markers in the Nude Mouse/Human Skin Model. Pathobiology, 1984, 52, 251-259.	1.9	2
259	Induction of the fra(X) in amniotic fluid cells by excess thymidine. American Journal of Medical Genetics Part A, 1988, 30, 451-453.	2.4	2
260	Assignment‹footref rid="foot01"›‹sup›1‹/sup›‹/footref› of the NR2E3 gene to mouse chromosome 9 and to human chromosome 15q22.33â†’q23. Cytogenetic and Genome Research, 2000, 89, 279-280.	0.6	2
261	A cryptic unbalanced translocation resulting in del 13q and dup 15q. American Journal of Medical Genetics, Part A, 2008, 146A, 2570-2573.	0.7	2
262	Paroxysmal Cranial Dyskinesia and Nailâ€Patella Syndrome Caused by a Novel Variant in the LMX1B Gene. Movement Disorders, 2020, 35, 2343-2347.	2.2	2
263	Rare Pathogenic Variants in Genes Implicated in Glutamatergic Neurotransmission Pathway Segregate with Schizophrenia in Pakistani Families. Genes, 2021, 12, 1899.	1.0	2
264	Fragile X: Carrier detection in pregnancy. American Journal of Medical Genetics Part A, 1986, 23, 527-530.	2.4	1
265	A holder for critical point drying of large numbers of EM-grids. Microscopy Research and Technique, 1992, 23, 353-354.	1.2	1
266	Structural organization, tissue expression, and chromosomal localization of Ciao 1, a functional modulator of the Wilms' tumor suppressor, WT1. Immunogenetics, 1999, 49, 900-905.	1.2	1
267	A splice-site variant in the lncRNA gene cosegregates in the large Volkmann cataract family. Molecular Vision, 2019, 25, 1-11.	1.1	1
268	Hydatidiform mole: a chromosomal search for a recessive mutation. Human Reproduction, 1986, 1, 337-340.	0.4	0
269	PCR-based screening of YAC clones without DNA extraction. Technical Tips Online, 1999, 4, 1-3.	0.2	0
270	Sequencing and mapping of the porcineCCSgene. Animal Genetics, 2004, 35, 353-354.	0.6	0

#	Article	IF	Citations
271	9q subtelomeric deletion syndrome with diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2009, 149A, 1086-1088.	0.7	0
272	Regulatory Mutations of FOXG1 in the Context of Topological Domains. Cancer Genetics, 2016, 209, 245.	0.2	0
273	A novel in-frame mutation in CLN3 leads to Juvenile neuronal ceroid lipofuscinosis in a large Pakistani family. International Journal of Neuroscience, 2019, 129, 890-895.	0.8	0
274	Title is missing!. , 2020, 16, e1009231.		0
275	Title is missing!. , 2020, 16, e1009231.		0
276	Title is missing!. , 2020, 16, e1009231.		0
277	Title is missing!. , 2020, 16, e1009231.		0