Niels Tommerup

List of Publications by Year
in descending order

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277	19,707	17440 63	12597 132
papers	citations	h-index	g-index

283	283	283	26467
all docs	docs citations	times ranked	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.	28.9	1,417
2	Rubinstein-Taybi syndrome caused by mutations in the transcriptional co-activator CBP. Nature, 1995, 376, 348-351.	27.8	1,140
3	Chromosome instability and immunodeficiency syndrome caused by mutations in a DNA methyltransferase gene. Nature, 1999, 402, 187-191.	27.8	1,056
4	A human phenome-interactome network of protein complexes implicated in genetic disorders. Nature Biotechnology, 2007, 25, 309-316.	17.5	871
5	Ancient human genome sequence of an extinct Palaeo-Eskimo. Nature, 2010, 463, 757-762.	27.8	750
6	Isolation of a candidate gene for Menkes disease that encodes a potential heavy metal binding protein. Nature Genetics, 1993, 3, 14-19.	21.4	708
7	Direct Diagnosis by DNA Analysis of the Fragile X Syndrome of Mental Retardation. New England Journal of Medicine, 1991, 325, 1673-1681.	27.0	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.	5.9	636
9	JARID2 regulates binding of the Polycomb repressive complexâ€‰2 to target genes in ES cells. Nature, 2010, 464, 306-310.	27.8	499
10	MicroRNA expression in the adult mouse central nervous system. Rna, 2008, 14, 432-444.	3.5	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.	21.4	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.	3.4	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.	6.2	265
14	The phenotypic spectrum of SCN8A encephalopathy. Neurology, 2015, 84, 480-489.	1.1	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.	3.4	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.	6.2	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.	12.0	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

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19	Assignment of an autosomal sex reversaâ€“ locus (SRA1) and campomelic dysplasia (CMPD1) to 17q24.3â€“q25.1. Nature Genetics, 1993, 4, 170-174.	21.4	196
20	Mutations inSYNGAP1Cause Intellectual Disability, Autism, and a Specific Form of Epilepsy by Inducing Haploinsufficiency. Human Mutation, 2013, 34, 385-394.	2.5	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.	9.7	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.	7.1	183
23	Jarid1b targets genes regulating development and is involved in neural differentiation. EMBO Journal, 2011, 30, 4586-4600.	7.8	183
24	Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. Annals of Neurology, 2016, 79, 428-436.	5.3	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.	1.4	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.	3.3	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.	6.2	145
29	RESTâ€“Mediated Recruitment of Polycomb Repressor Complexes in Mammalian Cells. PLoS Genetics, 2012, 8, e1002494.	3.5	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.	6.2	139
31	A balanced chromosomal translocation disruptingARHGEF9is associated with epilepsy, anxiety, aggression, and mental retardation. Human Mutation, 2009, 30, 61-68.	2.5	131
32	Corpus callosum abnormalities, intellectual disability, speech impairment, and autism in patients with haploinsufficiency of ARID1B. Clinical Genetics, 2012, 82, 248-255.	2.0	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.	2.9	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.	2.9	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.	7.1	117
36	Mutations in autism susceptibility candidate 2 (AUTS2) in patients with mental retardation. Human Genetics, 2007, 121, 501-509.	3.8	116

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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.	3.2	110
38	Distinct CDH3 mutations cause ectodermal dysplasia, ectrodactyly, macular dystrophy (EEM) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 702	3.2	108
39	Gene Panel Testing in Epileptic Encephalopathies and Familial Epilepsies. Molecular Syndromology, 2016, 7, 210-219.	0.8	103
40	Haploinsufficiency of TAB2 Causes Congenital Heart Defects in Humans. American Journal of Human Genetics, 2010, 86, 839-849.	6.2	97
41	Haploinsufficiency of novel FOXG1B variants in a patient with severe mental retardation, brain malformations and microcephaly. Human Genetics, 2005, 117, 536-544.	3.8	96
42	The Genetic Basis of the Pierre Robin Sequence. Cleft Palate-Craniofacial Journal, 2006, 43, 155-159.	0.9	95
43	Pierre Robin sequence may be caused by dysregulation of SOX9 and KCNJ2. Journal of Medical Genetics, 2007, 44, 381-386.	3.2	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.	2.8	90
45	Genetic Association Studies in Lumbar Disc Degeneration: A Systematic Review. PLoS ONE, 2012, 7, e49995.	2.5	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.	6.2	87
47	Mutations in GABRB3. Neurology, 2017, 88, 483-492.	1.1	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.	3.5	83
50	Assignment of Human Elongation Factor 1Î± Genes:EEF1AMaps to Chromosome 6q14 andEEF1A2to 20q13.3. Genomics, 1996, 36, 359-361.	2.9	82
51	Comparative genomics beyond sequence-based alignments: RNA structures in the ENCODE regions. Genome Research, 2008, 18, 242-251.	5.5	82
52	Hedgehog signaling in small-cell lung cancer: Frequent in vivo but a rare event in vitro. Lung Cancer, 2006, 52, 281-290.	2.0	80
53	Dissecting spatioâ€temporal protein networks driving human heart development and related disorders. Molecular Systems Biology, 2010, 6, 381.	7.2	80
54	Genome-wide detection of chromosomal rearrangements, indels, and mutations in circular chromosomes by short read sequencing. Genome Research, 2011, 21, 1388-1393.	5.5	79

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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.	21.4	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.	3.8	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.	5.1	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.	5.1	71
60	The identification and functional annotation of RNA structures conserved in vertebrates. Genome Research, 2017, 27, 1371-1383.	5.5	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.	2.8	68
63	Genome-wide Analysis of CDX2 Binding in Intestinal Epithelial Cells (Caco-2). Journal of Biological Chemistry, 2010, 285, 25115-25125.	3.4	68
64	Isolated and syndromic forms of congenital anosmia. Clinical Genetics, 2012, 81, 210-215.	2.0	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.	3.5	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.	1.5	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.	1.7	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.	2.8	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.	3.4	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.	6.2	59
71	Exonâ€disrupting deletions of <scp>NRXN1</scp> in idiopathic generalized epilepsy. Epilepsia, 2013, 54, 256-264.	5.1	59
72	FISHing with locked nucleic acids (LNA): evaluation of different LNA/DNA mixmers. Molecular and Cellular Probes, 2003, 17, 165-169.	2.1	56

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73	An excess of chromosome 1 breakpoints in male infertility. European Journal of Human Genetics, 2004, 12, 993-1000.	2.8	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.	3.8	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.	1.2	54
77	Profiling microRNAs in lung tissue from pigs infected with Actinobacillus pleuropneumoniae. BMC Genomics, 2012, 13, 459.	2.8	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.	2.8	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.	0.7	51
81	LNA-modified oligonucleotides are highly efficient as FISH probes. Cytogenetic and Genome Research, 2004, 107, 32-37.	1.1	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.	1.6	51
83	Reduced ceramide synthase 2 activity causes progressive myoclonic epilepsy. Annals of Clinical and Translational Neurology, 2014, 1, 88-98.	3.7	50
84	Germline Chromothripsis Driven by L1-Mediated Retrotransposition and Alu/Alu Homologous Recombination. Human Mutation, 2016, 37, 385-395.	2.5	50
85	Macroorchidism and fragile X in mentally retarded males. Human Genetics, 1982, 61, 113-117.	3.8	49
86	A neocentromere on human chromosome 3 without detectable Î±-satellite DNA forms morphologically normal kinetochores. Chromosoma, 1998, 107, 359-365.	2.2	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.	2.9	48
88	Mapping of the Human PAWR (par-4) Gene to Chromosome 12q21. Genomics, 1998, 53, 241-243.	2.9	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.	0.7	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.	2.8	46

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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.	2.8	45
92	Dysfunction of the Heteromeric KV7.3/KV7.5 Potassium Channel is Associated with Autism Spectrum Disorders. Frontiers in Genetics, 2013, 4, 54.	2.3	45
93	CHEMOATTRACTION INTETRAHYMENA: ON THE ROLE OF CHEMOKINESIS. Biological Bulletin, 1986, 170, 357-367.	1.8	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.	3.2	44
96	Mutational analysis of the human FATE gene in 144 infertile men. Human Genetics, 2003, 113, 195-201.	3.8	41
97	Genetic heterogeneity in Pakistani microcephaly families. Clinical Genetics, 2013, 83, 446-451.	2.0	41
98	Sequence assembly. Computational Biology and Chemistry, 2009, 33, 121-136.	2.3	39
99	Identification of human candidate genes for male infertility by digital differential display. Molecular Human Reproduction, 2001, 7, 11-20.	2.8	38
100	Interstitial deletion 9q22.32â€q33.2 associated with additional familial translocation t(9;17)(q34.11;p11.2) in a patient with Gorlinâ€“Goltz syndrome and features of Nailâ€Patella syndrome. American Journal of Medical Genetics, Part A, 2004, 124A, 179-191.	1.2	38
101	Non-disjunction of chromosome 13. Human Molecular Genetics, 2007, 16, 2004-2010.	2.9	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.	1.2	37
103	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. European Journal of Human Genetics, 2016, 24, 1761-1770.	2.8	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.	2.8	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.	2.9	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.	2.8	34

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109	RRP7A links primary microcephaly to dysfunction of ribosome biogenesis, resorption of primary cilia, and neurogenesis. Nature Communications, 2020, 11, 5816.	12.8	34
110	Small supernumerary marker chromosomes: A legacy of trisomy rescue?. Human Mutation, 2019, 40, 193-200.	2.5	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.	1.1	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.	2.1	32
114	Compound heterozygous ASPM mutations in Pakistani MCPH families. American Journal of Medical Genetics, Part A, 2009, 149A, 926-930.	1.2	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.	2.8	31
116	Accurate Breakpoint Mapping in Apparently Balanced Translocation Families with Discordant Phenotypes Using Whole Genome Mate-Pair Sequencing. PLoS ONE, 2017, 12, e0169935.	2.5	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.	3.2	30
119	Metaphase FISH on a Chip: Miniaturized Microfluidic Device for Fluorescence in situ Hybridization. Sensors, 2010, 10, 9831-9846.	3.8	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.	2.8	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.	1.2	29
122	Identification of the BRD1 interaction network and its impact on mental disorder risk. Genome Medicine, 2016, 8, 53.	8.2	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.	6.2	29
124	Structural genomic variation in childhood epilepsies with complex phenotypes. European Journal of Human Genetics, 2014, 22, 896-901.	2.8	28
125	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. PLoS Genetics, 2018, 14, e1007780.	3.5	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.	1.2	27

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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.	2.9	26
129	Human FATE is a novel X-linked gene expressed in fetal and adult testis. Molecular and Cellular Endocrinology, 2001, 184, 25-32.	3.2	26
130	The Hedgehog signaling pathway â€“ implications for drug targets in cancer and neurodegenerative disorders. Pharmacogenomics, 2003, 4, 411-429.	1.3	26
131	A balanced translocation disruptsSYNGAP1in a patient with intellectual disability, speech impairment, and epilepsy with myoclonic absences (EMA). Epilepsia, 2011, 52, e190-e193.	5.1	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.	3.2	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.	3.8	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.	7.7	25
136	The first mutation in <scp>CNGA2</scp> in two brothers with anosmia. Clinical Genetics, 2015, 88, 293-296.	2.0	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.	2.5	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.	6.2	23
141	Regional differences in expression of specific markers for human embryonic stem cells. Reproductive BioMedicine Online, 2007, 15, 89-98.	2.4	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.	2.9	23
143	De novo unbalanced translocations have a complex history/aetiology. Human Genetics, 2018, 137, 817-829.	3.8	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.	2.9	22

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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.	2.9	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.	2.9	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.	1.2	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.	2.8	20
150	Stones in the lacrimal gland: a rare condition. Acta Ophthalmologica, 2009, 87, 672-675.	1.1	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.	7.2	20
152	Neural correlates of taste perception in congenital olfactory impairment. Neuropsychologia, 2014, 62, 297-305.	1.6	20
153	Regulatory variants of FOXG1 in the context of its topological domain organisation. European Journal of Human Genetics, 2018, 26, 186-196.	2.8	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.	13.7	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.	1.3	19
157	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. Human Molecular Genetics, 2014, 23, 6163-6176.	2.9	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.	2.5	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.	1.2	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.	2.1	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.	2.3	18
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