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	Does rapid sequence divergence preclude RNA structure conservation in vertebrates?. Nucleic Acids Research, 2022, 50, 2452-2463.	14.5	3
2	Acute and persistent symptoms in non-hospitalized PCR-confirmed COVID-19 patients. Scientific Reports, 2021, 11, 13153.	3.3	147
3	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.	1.3	5
4	Rare Pathogenic Variants in Genes Implicated in Glutamatergic Neurotransmission Pathway Segregate with Schizophrenia in Pakistani Families. Genes, 2021, 12, 1899.	2.4	2
5	RRP7A links primary microcephaly to dysfunction of ribosome biogenesis, resorption of primary cilia, and neurogenesis. Nature Communications, 2020, 11, 5816.	12.8	34
6	Paroxysmal Cranial Dyskinesia and Nailâ€Patella Syndrome Caused by a Novel Variant in the LMX1B Gene. Movement Disorders, 2020, 35, 2343-2347.	3.9	2
7	A shared somatic translocation involving CUX1 in monozygotic twins as an early driver of AMKL in Down syndrome. Blood Cancer Journal, 2020, 10, 27.	6.2	3
8	Chromothripsis and DNA Repair Disorders. Journal of Clinical Medicine, 2020, 9, 613.	2.4	18
9	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
10	Title is missing!. , 2020, 16, e1009231.		0
11	Title is missing!. , 2020, 16, e1009231.		0
12	Title is missing!. , 2020, 16, e1009231.		0
13	Title is missing!. , 2020, 16, e1009231.		0
14	Multigenic truncation of the semaphorinâ€“plexin pathway by a germline chromothriptic rearrangement associated with Moebius syndrome. Human Mutation, 2019, 40, 1057-1062.	2.5	4
15	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.	1.6	0
16	Haploinsufficiency of ARHGAP42 is associated with hypertension. European Journal of Human Genetics, 2019, 27, 1296-1303.	2.8	12
17	Small supernumerary marker chromosomes: A legacy of trisomy rescue?. Human Mutation, 2019, 40, 193-200.	2.5	33
18	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.	2.0	4

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19	A splice-site variant in the lncRNA gene cosegregates in the large Volkmann cataract family. Molecular Vision, 2019, 25, 1-11.	1.1	1
20	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
21	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
22	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.	1.2	8
23	Regulatory variants of FOXG1 in the context of its topological domain organisation. European Journal of Human Genetics, 2018, 26, 186-196.	2.8	20
24	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
25	De novo unbalanced translocations have a complex history/aetiology. Human Genetics, 2018, 137, 817-829.	3.8	23
26	Position effect, cryptic complexity, and direct gene disruption as disease mechanisms in de novo apparently balanced translocation cases. PLoS ONE, 2018, 13, e0205298.	2.5	14
27	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
28	Cryptic breakpoint identified by whole-genome mate-pair sequencing in a rare paternally inherited complex chromosomal rearrangement. Molecular Cytogenetics, 2018, 11, 34.	0.9	11
29	Challenges for the Sustainability of University-Run Biobanks. Biopreservation and Biobanking, 2018, 16, 312-321.	1.0	12
30	The identification and functional annotation of RNA structures conserved in vertebrates. Genome Research, 2017, 27, 1371-1383.	5.5	71
31	Abdominal Wall Defects in Greenland 1989â€“2015. Birth Defects Research, 2017, 109, 836-842.	1.5	13
32	Mutations in GABRB3. Neurology, 2017, 88, 483-492.	1.1	87
33	Homozygous mutation in the NPHP3 gene causing foetal nephronophthisis. Nephrology, 2017, 22, 818-820.	1.6	4
34	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.	1.3	10
35	Enrichment of megabase-sized DNA molecules for single-molecule optical mapping and next-generation sequencing. Scientific Reports, 2017, 7, 17893.	3.3	5
36	Genome-Wide Supported Risk Variants inMIR137,CACNA1C,CSMD1,DRD2, andGRM3Contribute to Schizophrenia Susceptibility in Pakistani Population. Psychiatry Investigation, 2017, 14, 687.	1.6	13

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37	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
38	Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. Annals of Neurology, 2016, 79, 428-436.	5.3	159
39	Germline Chromothripsis Driven by L1-Mediated Retrotransposition and Alu/Alu Homologous Recombination. Human Mutation, 2016, 37, 385-395.	2.5	50
40	Regulatory Mutations of FOXG1 in the Context of Topological Domains. Cancer Genetics, 2016, 209, 245.	0.4	0
41	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
42	Gene Panel Testing in Epileptic Encephalopathies and Familial Epilepsies. Molecular Syndromology, 2016, 7, 210-219.	0.8	103
43	Monozygotic twins discordant for narcolepsy type 1 and multiple sclerosis. Neurology: Neuroimmunology and NeuroInflammation, 2016, 3, e249.	6.0	7
44	Phenotypic subregions within the split-hand/foot malformation 1 locus. Human Genetics, 2016, 135, 345-357.	3.8	15
45	Identification of the BRD1 interaction network and its impact on mental disorder risk. Genome Medicine, 2016, 8, 53.	8.2	29
46	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
47	A novel splice site mutation in CEP135 is associated with primary microcephaly in a Pakistani family. Journal of Human Genetics, 2016, 61, 271-273.	2.3	16
48	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.	1.3	12
49	The segregation of different submicroscopic imbalances underlying the clinical variability associated with a familial karyotypically balanced translocation. Molecular Cytogenetics, 2015, 8, 106.	0.9	6
50	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.	8.2	16
51	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
52	A Novel Locus Harbouring a Functional CD164 Nonsense Mutation Identified in a Large Danish Family with Nonsyndromic Hearing Impairment. PLoS Genetics, 2015, 11, e1005386.	3.5	18
53	Dysregulation of FOXG1 by ring chromosome 14. Molecular Cytogenetics, 2015, 8, 24.	0.9	8
54	The phenotypic spectrum of SCN8A encephalopathy. Neurology, 2015, 84, 480-489.	1.1	246

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55	Automation of a single-DNA molecule stretching device. Review of Scientific Instruments, 2015, 86, 063702.	1.3	7
56	The small RNA content of human sperm reveals pseudogene-derived piRNAs complementary to protein-coding genes. Rna, 2015, 21, 1085-1095.	3.5	83
57	Partial USH2A deletions contribute to Usher syndrome in Denmark. European Journal of Human Genetics, 2015, 23, 1646-1651.	2.8	8
58	Optical mapping of single-molecule human DNA in disposable, mass-produced all-polymer devices. Journal of Micromechanics and Microengineering, 2015, 25, 105002.	2.6	18
59	The first mutation in <scp>CNGA2</scp> in two brothers with anosmia. Clinical Genetics, 2015, 88, 293-296.	2.0	25
60	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
61	Reduced ceramide synthase 2 activity causes progressive myoclonic epilepsy. Annals of Clinical and Translational Neurology, 2014, 1, 88-98.	3.7	50
62	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. Human Molecular Genetics, 2014, 23, 6163-6176.	2.9	19
63	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.	3.8	17
64	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
65	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
66	Neural correlates of taste perception in congenital olfactory impairment. Neuropsychologia, 2014, 62, 297-305.	1.6	20
67	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
68	Sequence analysis of 17 NRXN1 deletions. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 52-61.	1.7	11
69	Structural genomic variation in childhood epilepsies with complex phenotypes. European Journal of Human Genetics, 2014, 22, 896-901.	2.8	28
70	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
71	Mutations inSYNGAP1Cause Intellectual Disability, Autism, and a Specific Form of Epilepsy by Inducing Haploinsufficiency. Human Mutation, 2013, 34, 385-394.	2.5	196
72	The role of SLC2A1 in early onset and childhood absence epilepsies. Epilepsy Research, 2013, 105, 229-233.	1.6	13

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73	Genetic heterogeneity in Pakistani microcephaly families. Clinical Genetics, 2013, 83, 446-451.	2.0	41
74	An association study between the norepinephrine transporter gene and depression. Psychiatric Genetics, 2013, 23, 217-221.	1.1	4
75	Exonâ€disrupting deletions of <scp>NRXN1</scp> in idiopathic generalized epilepsy. Epilepsia, 2013, 54, 256-264.	5.1	59
76	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
77	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
78	RESTâ€“Mediated Recruitment of Polycomb Repressor Complexes in Mammalian Cells. PLoS Genetics, 2012, 8, e1002494.	3.5	140
79	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
80	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
81	Sequence and expression analysis of gaps in human chromosome 20. Nucleic Acids Research, 2012, 40, 6660-6672.	14.5	5
82	Corpus callosum abnormalities, intellectual disability, speech impairment, and autism in patients with haploinsufficiency of ARID1B. Clinical Genetics, 2012, 82, 248-255.	2.0	126
83	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.	2.8	16
84	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
85	Profiling microRNAs in lung tissue from pigs infected with Actinobacillus pleuropneumoniae. BMC Genomics, 2012, 13, 459.	2.8	54
86	Genetic Association Studies in Lumbar Disc Degeneration: A Systematic Review. PLoS ONE, 2012, 7, e49995.	2.5	90
87	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
88	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
89	Genetic studies in congenital anterior midline cervical cleft. American Journal of Medical Genetics, Part A, 2012, 158A, 2021-2026.	1.2	12
90	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

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91	Isolated and syndromic forms of congenital anosmia. Clinical Genetics, 2012, 81, 210-215.	2.0	66
92	Biparental inheritance of chromosomal abnormalities in male twins with non-syndromic mental retardation. European Journal of Medical Genetics, 2011, 54, e383-e388.	1.3	7
93	The lrrk2 p.Gly2019Ser mutation is uncommon in a Danish cohort with various neurodegenerative disorders. Parkinsonism and Related Disorders, 2011, 17, 398-399.	2.2	4
94	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
95	Duplication of MAOA, MAOB, and NDP in a patient with mental retardation and epilepsy. European Journal of Human Genetics, 2011, 19, 1-2.	2.8	9
96	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
97	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.	1.4	4
98	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
99	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.	1.2	6
100	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.	1.2	6
101	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
102	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
103	Jarid1b targets genes regulating development and is involved in neural differentiation. EMBO Journal, 2011, 30, 4586-4600.	7.8	183
104	Dissecting spatioâ€temporal protein networks driving human heart development and related disorders. Molecular Systems Biology, 2010, 6, 381.	7.2	80
105	Haploinsufficiency of TAB2 Causes Congenital Heart Defects in Humans. American Journal of Human Genetics, 2010, 86, 839-849.	6.2	97
106	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
107	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
108	Autoimmune diseases in women with Turner's Syndrome. Arthritis and Rheumatism, 2010, 62, 658-666.	6.7	147

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109	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
110	JARID2 regulates binding of the Polycomb repressive complexâ€‰2 to target genes in ES cells. Nature, 2010, 464, 306-310.	27.8	499
111	Ancient human genome sequence of an extinct Palaeo-Eskimo. Nature, 2010, 463, 757-762.	27.8	750
112	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
113	Genome-wide Analysis of CDX2 Binding in Intestinal Epithelial Cells (Caco-2). Journal of Biological Chemistry, 2010, 285, 25115-25125.	3.4	68
114	Metaphase FISH on a Chip: Miniaturized Microfluidic Device for Fluorescence in situ Hybridization. Sensors, 2010, 10, 9831-9846.	3.8	30
115	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
116	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
117	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
118	Compound heterozygous ASPM mutations in Pakistani MCPH families. American Journal of Medical Genetics, Part A, 2009, 149A, 926-930.	1.2	31
119	9q subtelomeric deletion syndrome with diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2009, 149A, 1086-1088.	1.2	0
120	A novel subtype of distal symphalangism affecting only the 4th finger. American Journal of Medical Genetics, Part A, 2009, 149A, 1571-1573.	1.2	8
121	Molecular characterization of two patients with de novo interstitial deletions in 4q22â€“q24. American Journal of Medical Genetics, Part A, 2009, 149A, 1830-1833.	1.2	4
122	A balanced chromosomal translocation disruptingARHGEF9is associated with epilepsy, anxiety, aggression, and mental retardation. Human Mutation, 2009, 30, 61-68.	2.5	131
123	Genomeâ€wide Gene Expression Profiling of SCID Mice with Tâ€cellâ€mediated Colitis. Scandinavian Journal of Immunology, 2009, 69, 437-446.	2.7	4
124	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
125	Sequence assembly. Computational Biology and Chemistry, 2009, 33, 121-136.	2.3	39
126	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

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127	Stones in the lacrimal gland: a rare condition. Acta Ophthalmologica, 2009, 87, 672-675.	1.1	20
128	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.	1.2	12
129	A cryptic unbalanced translocation resulting in del 13q and dup 15q. American Journal of Medical Genetics, Part A, 2008, 146A, 2570-2573.	1.2	2
130	Investigation of 4qâ€deletion in two unrelated patients using array CGH. American Journal of Medical Genetics, Part A, 2008, 146A, 2431-2434.	1.2	17
131	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.	2.8	15
132	Balanced translocation in a patient with severe myoclonic epilepsy of infancy disrupts the sodium channel gene SCN1A. Epilepsia, 2008, 49, 1091-1094.	5.1	10
133	Mowatâ€“Wilson syndrome: an underdiagnosed syndrome?. Clinical Genetics, 2008, 73, 579-584.	2.0	15
134	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
135	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
136	GLI1 Is Involved in Cell Cycle Regulation and Proliferation of NT2 Embryonal Carcinoma Stem Cells. DNA and Cell Biology, 2008, 27, 251-256.	1.9	11
137	MicroRNA expression in the adult mouse central nervous system. Rna, 2008, 14, 432-444.	3.5	427
138	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
139	Comparative genomics beyond sequence-based alignments: RNA structures in the ENCODE regions. Genome Research, 2008, 18, 242-251.	5.5	82
140	Non-disjunction of chromosome 13. Human Molecular Genetics, 2007, 16, 2004-2010.	2.9	38
141	Pierre Robin sequence may be caused by dysregulation of SOX9 and KCNJ2. Journal of Medical Genetics, 2007, 44, 381-386.	3.2	91
142	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.	2.4	11
143	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
144	Regional differences in expression of specific markers for human embryonic stem cells. Reproductive BioMedicine Online, 2007, 15, 89-98.	2.4	23

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145	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
146	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.	1.2	9
147	A human phenome-interactome network of protein complexes implicated in genetic disorders. Nature Biotechnology, 2007, 25, 309-316.	17.5	871
148	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
149	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
150	Cytogenetically invisible microdeletions involving PITX2 in Rieger syndrome. Clinical Genetics, 2007, 72, 464-470.	2.0	15
151	Interstitial deletion of chromosome 4p associated with mild mental retardation, epilepsy and polymicrogyria of the left temporal lobe. Clinical Genetics, 2007, 72, 593-598.	2.0	10
152	Mutations in autism susceptibility candidate 2 (AUTS2) in patients with mental retardation. Human Genetics, 2007, 121, 501-509.	3.8	116
153	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
154	Screening of 99 Danish Patients with Congenital Heart Disease for GATA4 Mutations. Genetic Testing and Molecular Biomarkers, 2006, 10, 277-280.	1.7	24
155	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
156	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
157	A novel primate specific gene, CEI, is located in the homeobox gene IRXA2 promoter in Homo sapiens. Gene, 2006, 371, 167-173.	2.2	8
158	The Genetic Basis of the Pierre Robin Sequence. Cleft Palate-Craniofacial Journal, 2006, 43, 155-159.	0.9	95
159	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.	2.8	17
160	Evaluation of two methods for generating cRNA for microarray experiments from nanogram amounts of total RNA. Analytical Biochemistry, 2006, 358, 111-119.	2.4	5
161	Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. Human Genetics, 2006, 118, 578-590.	3.8	55
162	Population-based study of cancer among carriers of a constitutional structural chromosomal rearrangement. Genes Chromosomes and Cancer, 2006, 45, 231-246.	2.8	13

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163	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
164	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
165	4q35 deletion and 10p15 duplication associated with immunodeficiency. American Journal of Medical Genetics, Part A, 2006, 140A, 2231-2235.	1.2	10
166	Molecular characterization of a balanced chromosome translocation in psoriasis vulgaris. Clinical Genetics, 2005, 69, 189-193.	2.0	8
167	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
168	Delineation of an interstitial 9q22 deletion in basal cell nevus syndrome. , 2005, 132A, 324-328.		32
169	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.	1.2	4
170	Male-to-male transmission in Laurin-Sandrow syndrome and exclusion ofRARBandRARG. American Journal of Medical Genetics, Part A, 2005, 137A, 148-152.	1.2	7
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