

Niels Tommerup

List of Publications by Year in descending order

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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	Does rapid sequence divergence preclude RNA structure conservation in vertebrates?. <i>Nucleic Acids Research</i> , 2022, 50, 2452-2463.	6.5	3
2	Acute and persistent symptoms in non-hospitalized PCR-confirmed COVID-19 patients. <i>Scientific Reports</i> , 2021, 11, 13153.	1.6	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. <i>European Journal of Medical Genetics</i> , 2021, 64, 104226.	0.7	5
4	Rare Pathogenic Variants in Genes Implicated in Glutamatergic Neurotransmission Pathway Segregate with Schizophrenia in Pakistani Families. <i>Genes</i> , 2021, 12, 1899.	1.0	2
5	RRP7A links primary microcephaly to dysfunction of ribosome biogenesis, resorption of primary cilia, and neurogenesis. <i>Nature Communications</i> , 2020, 11, 5816.	5.8	34
6	Paroxysmal Cranial Dyskinesia and Nailâ€‘Patella Syndrome Caused by a Novel Variant in the LMX1B Gene. <i>Movement Disorders</i> , 2020, 35, 2343-2347.	2.2	2
7	A shared somatic translocation involving CUX1 in monozygotic twins as an early driver of AMKL in Down syndrome. <i>Blood Cancer Journal</i> , 2020, 10, 27.	2.8	3
8	Chromothripsis and DNA Repair Disorders. <i>Journal of Clinical Medicine</i> , 2020, 9, 613.	1.0	18
9	Nationwide germline whole genome sequencing of 198 consecutive pediatric cancer patients reveals a high incidence of cancer prone syndromes. <i>PLoS Genetics</i> , 2020, 16, e1009231.	1.5	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. <i>Human Mutation</i> , 2019, 40, 1057-1062.	1.1	4
15	A novel in-frame mutation in CLN3 leads to Juvenile neuronal ceroid lipofuscinosis in a large Pakistani family. <i>International Journal of Neuroscience</i> , 2019, 129, 890-895.	0.8	0
16	Haploinsufficiency of ARHGAP42 is associated with hypertension. <i>European Journal of Human Genetics</i> , 2019, 27, 1296-1303.	1.4	12
17	Small supernumerary marker chromosomes: A legacy of trisomy rescue?. <i>Human Mutation</i> , 2019, 40, 193-200.	1.1	33
18	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies syndrome due to disruption of <i>BPTF</i> in a 35-year-old man initially diagnosed with Silverâ€‘Russell syndrome. <i>Clinical Genetics</i> , 2019, 95, 534-536.	1.0	4

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19	A splice-site variant in the lncRNA gene cosegregates in the large Volkmann cataract family. <i>Molecular Vision</i> , 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. <i>Human Mutation</i> , 2018, 39, 709-716.	1.1	19
21	Congenital olfactory impairment is linked to cortical changes in prefrontal and limbic brain regions. <i>Brain Imaging and Behavior</i> , 2018, 12, 1569-1582.	1.1	32
22	Breakpoint mapping and haplotype analysis of translocation t(1;12)(q43;q21.1) in two apparently independent families with vascular phenotypes. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 56-68.	0.6	8
23	Regulatory variants of FOXP1 in the context of its topological domain organisation. <i>European Journal of Human Genetics</i> , 2018, 26, 186-196.	1.4	20
24	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. <i>PLoS Genetics</i> , 2018, 14, e1007780.	1.5	28
25	De novo unbalanced translocations have a complex history/aetiology. <i>Human Genetics</i> , 2018, 137, 817-829.	1.8	23
26	Position effect, cryptic complexity, and direct gene disruption as disease mechanisms in de novo apparently balanced translocation cases. <i>PLoS ONE</i> , 2018, 13, e0205298.	1.1	14
27	Risks and Recommendations in Prenatally Detected De Novo Balanced Chromosomal Rearrangements from Assessment of Long-Term Outcomes. <i>American Journal of Human Genetics</i> , 2018, 102, 1090-1103.	2.6	29
28	Cryptic breakpoint identified by whole-genome mate-pair sequencing in a rare paternally inherited complex chromosomal rearrangement. <i>Molecular Cytogenetics</i> , 2018, 11, 34.	0.4	11
29	Challenges for the Sustainability of University-Run Biobanks. <i>Biopreservation and Biobanking</i> , 2018, 16, 312-321.	0.5	12
30	The identification and functional annotation of RNA structures conserved in vertebrates. <i>Genome Research</i> , 2017, 27, 1371-1383.	2.4	71
31	Abdominal Wall Defects in Greenland 1989â€“2015. <i>Birth Defects Research</i> , 2017, 109, 836-842.	0.8	13
32	Mutations in <i>GABRB3</i> . <i>Neurology</i> , 2017, 88, 483-492.	1.5	87
33	Homozygous mutation in the <i>NPHP3</i> gene causing foetal nephronophthisis. <i>Nephrology</i> , 2017, 22, 818-820.	0.7	4
34	A novel mutation in CDK5RAP2 gene causes primary microcephaly with speech impairment and sparse eyebrows in a consanguineous Pakistani family. <i>European Journal of Medical Genetics</i> , 2017, 60, 627-630.	0.7	10
35	Enrichment of megabase-sized DNA molecules for single-molecule optical mapping and next-generation sequencing. <i>Scientific Reports</i> , 2017, 7, 17893.	1.6	5
36	Genome-Wide Supported Risk Variants in <i>MIR137</i> , <i>CACNA1C</i> , <i>CSMD1</i> , <i>DRD2</i> , and <i>GRM3</i> Contribute to Schizophrenia Susceptibility in Pakistani Population. <i>Psychiatry Investigation</i> , 2017, 14, 687.	0.7	13

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37	Accurate Breakpoint Mapping in Apparently Balanced Translocation Families with Discordant Phenotypes Using Whole Genome Mate-Pair Sequencing. <i>PLoS ONE</i> , 2017, 12, e0169935.	1.1	31
38	Benign infantile seizures and paroxysmal dyskinesia caused by an <i>SCN8A</i> mutation. <i>Annals of Neurology</i> , 2016, 79, 428-436.	2.8	159
39	Germline Chromothripsis Driven by L1-Mediated Retrotransposition and Alu/Alu Homologous Recombination. <i>Human Mutation</i> , 2016, 37, 385-395.	1.1	50
40	Regulatory Mutations of <i>FOXP1</i> in the Context of Topological Domains. <i>Cancer Genetics</i> , 2016, 209, 245.	0.2	0
41	Genome-wide DNA methylation analysis of transient neonatal diabetes type 1 patients with mutations in <i>ZFP57</i> . <i>BMC Medical Genetics</i> , 2016, 17, 29.	2.1	27
42	Gene Panel Testing in Epileptic Encephalopathies and Familial Epilepsies. <i>Molecular Syndromology</i> , 2016, 7, 210-219.	0.3	103
43	Monozygotic twins discordant for narcolepsy type 1 and multiple sclerosis. <i>Neurology: Neuroimmunology and Neuroinflammation</i> , 2016, 3, e249.	3.1	7
44	Phenotypic subregions within the split-hand/foot malformation 1 locus. <i>Human Genetics</i> , 2016, 135, 345-357.	1.8	15
45	Identification of the <i>BRD1</i> interaction network and its impact on mental disorder risk. <i>Genome Medicine</i> , 2016, 8, 53.	3.6	29
46	Loss of function of the retinoid-related nuclear receptor (<i>RORB</i>) gene and epilepsy. <i>European Journal of Human Genetics</i> , 2016, 24, 1761-1770.	1.4	36
47	A novel splice site mutation in <i>CEP135</i> is associated with primary microcephaly in a Pakistani family. <i>Journal of Human Genetics</i> , 2016, 61, 271-273.	1.1	16
48	Two rare deletions upstream of the <i>NRXN1</i> gene (2p16.3) affecting the non-coding mRNA AK127244 segregate with diverse psychopathological phenotypes in a family. <i>European Journal of Medical Genetics</i> , 2015, 58, 650-653.	0.7	12
49	The segregation of different submicroscopic imbalances underlying the clinical variability associated with a familial karyotypically balanced translocation. <i>Molecular Cytogenetics</i> , 2015, 8, 106.	0.4	6
50	Complete re-sequencing of a 2Mb topological domain encompassing the <i>FTO/IRXB</i> genes identifies a novel obesity-associated region upstream of <i>IRX5</i> . <i>Genome Medicine</i> , 2015, 7, 126.	3.6	16
51	The role of <i>SLC2A1</i> mutations in myoclonic astatic epilepsy and absence epilepsy, and the estimated frequency of <i>GLUT1</i> deficiency syndrome. <i>Epilepsia</i> , 2015, 56, e203-8.	2.6	71
52	A Novel Locus Harboring a Functional <i>CD164</i> Nonsense Mutation Identified in a Large Danish Family with Nonsyndromic Hearing Impairment. <i>PLoS Genetics</i> , 2015, 11, e1005386.	1.5	18
53	Dysregulation of <i>FOXP1</i> by ring chromosome 14. <i>Molecular Cytogenetics</i> , 2015, 8, 24.	0.4	8
54	The phenotypic spectrum of <i>SCN8A</i> encephalopathy. <i>Neurology</i> , 2015, 84, 480-489.	1.5	246

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55	Automation of a single-DNA molecule stretching device. Review of Scientific Instruments, 2015, 86, 063702.	0.6	7
56	The small RNA content of human sperm reveals pseudogene-derived piRNAs complementary to protein-coding genes. Rna, 2015, 21, 1085-1095.	1.6	83
57	Partial USH2A deletions contribute to Usher syndrome in Denmark. European Journal of Human Genetics, 2015, 23, 1646-1651.	1.4	8
58	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
59	The first mutation in <i>CNGA2</i> in two brothers with anosmia. Clinical Genetics, 2015, 88, 293-296.	1.0	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.	1.4	31
61	Reduced ceramide synthase 2 activity causes progressive myoclonic epilepsy. Annals of Clinical and Translational Neurology, 2014, 1, 88-98.	1.7	50
62	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. Human Molecular Genetics, 2014, 23, 6163-6176.	1.4	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.	1.8	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.	1.4	46
65	Aberrant expression of miR-18 and miR-204 in human mesial temporal lobe epilepsy and hippocampal sclerosis—Convergence on axonal guidance. Epilepsia, 2014, 55, 2017-2027.	2.6	71
66	Neural correlates of taste perception in congenital olfactory impairment. Neuropsychologia, 2014, 62, 297-305.	0.7	20
67	Neurodevelopmental disorders associated with dosage imbalance of <i>ZBTB20</i> correlate with the morbidity spectrum of ZBTB20 candidate target genes. Journal of Medical Genetics, 2014, 51, 605-613.	1.5	26
68	Sequence analysis of 17 <i>NRXN1</i> deletions. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 52-61.	1.1	11
69	Structural genomic variation in childhood epilepsies with complex phenotypes. European Journal of Human Genetics, 2014, 22, 896-901.	1.4	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.	2.6	87
71	Mutations in <i>SYNGAP1</i> Cause Intellectual Disability, Autism, and a Specific Form of Epilepsy by Inducing Haploinsufficiency. Human Mutation, 2013, 34, 385-394.	1.1	196
72	The role of SLC2A1 in early onset and childhood absence epilepsies. Epilepsy Research, 2013, 105, 229-233.	0.8	13

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73	Genetic heterogeneity in Pakistani microcephaly families. <i>Clinical Genetics</i> , 2013, 83, 446-451.	1.0	41
74	An association study between the norepinephrine transporter gene and depression. <i>Psychiatric Genetics</i> , 2013, 23, 217-221.	0.6	4
75	Exonâ€disrupting deletions of <sc><i>NRXN1</i></sc> in idiopathic generalized epilepsy. <i>Epilepsia</i> , 2013, 54, 256-264.	2.6	59
76	Nextâ€generation sequencing: proof of concept for antenatal prediction of the fetal <sc>K</sc>ell blood group phenotype from cellâ€free fetal <sc>DNA</sc> in maternal plasma. <i>Transfusion</i> , 2013, 53, 2892-2898.	0.8	51
77	Dysfunction of the Heteromeric KV7.3/KV7.5 Potassium Channel is Associated with Autism Spectrum Disorders. <i>Frontiers in Genetics</i> , 2013, 4, 54.	1.1	45
78	RESTâ€Mediated Recruitment of Polycomb Repressor Complexes in Mammalian Cells. <i>PLoS Genetics</i> , 2012, 8, e1002494.	1.5	140
79	Genetic and environmental risk factors in congenital heart disease functionally converge in protein networks driving heart development. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 14035-14040.	3.3	117
80	Haploinsufficiency of CELF4 at 18q12.2 is associated with developmental and behavioral disorders, seizures, eye manifestations, and obesity. <i>European Journal of Human Genetics</i> , 2012, 20, 1315-1319.	1.4	34
81	Sequence and expression analysis of gaps in human chromosome 20. <i>Nucleic Acids Research</i> , 2012, 40, 6660-6672.	6.5	5
82	Corpus callosum abnormalities, intellectual disability, speech impairment, and autism in patients with haploinsufficiency of <i>ARID1B</i>. <i>Clinical Genetics</i> , 2012, 82, 248-255.	1.0	126
83	No evidence for pathogenic variants or maternal effect of ZFP57 as the cause of Beckwithâ€Wiedemann Syndrome. <i>European Journal of Human Genetics</i> , 2012, 20, 119-121.	1.4	16
84	Translocations Disrupting PHF21A in the Potocki-Shaffer-Syndrome Region Are Associated with Intellectual Disability and Craniofacial Anomalies. <i>American Journal of Human Genetics</i> , 2012, 91, 56-72.	2.6	59
85	Profiling microRNAs in lung tissue from pigs infected with <i>Actinobacillus pleuropneumoniae</i> . <i>BMC Genomics</i> , 2012, 13, 459.	1.2	54
86	Genetic Association Studies in Lumbar Disc Degeneration: A Systematic Review. <i>PLoS ONE</i> , 2012, 7, e49995.	1.1	90
87	Validation of Genome-Wide Intervertebral Disk Calcification Associations in Dachshund and Further Investigation of the Chromosome 12 Susceptibility Locus. <i>Frontiers in Genetics</i> , 2012, 3, 225.	1.1	18
88	Screening of congenital heart disease patients using multiplex ligationâ€dependent probe amplification: Early diagnosis of syndromic patients. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 720-725.	0.7	27
89	Genetic studies in congenital anterior midline cervical cleft. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2021-2026.	0.7	12
90	Mutations in <i>NRXN1</i> in a family multiply affected with brain disorders: <i>NRXN1</i> mutations and brain disorders. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012, 159B, 354-358.	1.1	63

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91	Isolated and syndromic forms of congenital anosmia. <i>Clinical Genetics</i> , 2012, 81, 210-215.	1.0	66
92	Biparental inheritance of chromosomal abnormalities in male twins with non-syndromic mental retardation. <i>European Journal of Medical Genetics</i> , 2011, 54, e383-e388.	0.7	7
93	The <i>Irrk2</i> p.Gly2019Ser mutation is uncommon in a Danish cohort with various neurodegenerative disorders. <i>Parkinsonism and Related Disorders</i> , 2011, 17, 398-399.	1.1	4
94	A balanced translocation disrupts <i>SYNGAP1</i> in a patient with intellectual disability, speech impairment, and epilepsy with myoclonic absences (EMA). <i>Epilepsia</i> , 2011, 52, e190-e193.	2.6	26
95	Duplication of MAOA, MAOB, and NDP in a patient with mental retardation and epilepsy. <i>European Journal of Human Genetics</i> , 2011, 19, 1-2.	1.4	9
96	Cost-effective multiplexing before capture allows screening of 25,000 clinically relevant SNPs in childhood acute lymphoblastic leukemia. <i>Leukemia</i> , 2011, 25, 1001-1006.	3.3	20
97	Autozygosity mapping of a large consanguineous Pakistani family reveals a novel non-syndromic autosomal recessive mental retardation locus on 11p15-tel. <i>Neurogenetics</i> , 2011, 12, 247-251.	0.7	4
98	A Cohort of Balanced Reciprocal Translocations Associated with Dyslexia: Identification of Two Putative Candidate Genes at <i>DYX1</i> . <i>Behavior Genetics</i> , 2011, 41, 125-133.	1.4	18
99	Interstitial deletion of 14q24.3q32.2 in a male patient with plagiocephaly, BPES features, developmental delay, and congenital heart defects. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 203-206.	0.7	6
100	500K SNP array analyses in blood and saliva showed no differences in a pair of monozygotic twins discordant for cleft lip. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 652-655.	0.7	6
101	High frequency of rare copy number variants affecting functionally related genes in patients with structural brain malformations. <i>Human Mutation</i> , 2011, 32, 1427-1435.	1.1	24
102	Genome-wide detection of chromosomal rearrangements, indels, and mutations in circular chromosomes by short read sequencing. <i>Genome Research</i> , 2011, 21, 1388-1393.	2.4	79
103	<i>Jarid1b</i> targets genes regulating development and is involved in neural differentiation. <i>EMBO Journal</i> , 2011, 30, 4586-4600.	3.5	183
104	Dissecting spatio-temporal protein networks driving human heart development and related disorders. <i>Molecular Systems Biology</i> , 2010, 6, 381.	3.2	80
105	Haploinsufficiency of <i>TAB2</i> Causes Congenital Heart Defects in Humans. <i>American Journal of Human Genetics</i> , 2010, 86, 839-849.	2.6	97
106	Craniosynostosis-microcephaly with chromosomal breakage and other abnormalities is caused by a truncating <i>MCPH1</i> mutation and is allelic to premature chromosomal condensation syndrome and primary autosomal recessive microcephaly type 1. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 495-497.	0.7	18
107	Deletion of 7q34-q36.2 in two siblings with mental retardation, language delay, primary amenorrhea, and dysmorphic features. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 3115-3119.	0.7	37
108	Autoimmune diseases in women with Turner's Syndrome. <i>Arthritis and Rheumatism</i> , 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. <i>European Journal of Human Genetics</i> , 2010, 18, 733-736.	1.4	53
110	JARID2 regulates binding of the Polycomb repressive complex to target genes in ES cells. <i>Nature</i> , 2010, 464, 306-310.	13.7	499
111	Ancient human genome sequence of an extinct Palaeo-Eskimo. <i>Nature</i> , 2010, 463, 757-762.	13.7	750
112	Single-molecule denaturation mapping of DNA in nanofluidic channels. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 13294-13299.	3.3	183
113	Genome-wide Analysis of CDX2 Binding in Intestinal Epithelial Cells (Caco-2). <i>Journal of Biological Chemistry</i> , 2010, 285, 25115-25125.	1.6	68
114	Metaphase FISH on a Chip: Miniaturized Microfluidic Device for Fluorescence in situ Hybridization. <i>Sensors</i> , 2010, 10, 9831-9846.	2.1	30
115	A Functional Link between the Histone Demethylase PHF8 and the Transcription Factor ZNF711 in X-Linked Mental Retardation. <i>Molecular Cell</i> , 2010, 38, 165-178.	4.5	186
116	A mutation in the FOXE3 gene causes congenital primary aphakia in an autosomal recessive consanguineous Pakistani family. <i>Molecular Vision</i> , 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. <i>Experimental and Molecular Medicine</i> , 2009, 41, 77.	3.2	25
118	Compound heterozygous ASPM mutations in Pakistani MCPH families. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 926-930.	0.7	31
119	9q subtelomeric deletion syndrome with diaphragmatic hernia. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1086-1088.	0.7	0
120	A novel subtype of distal symphalangism affecting only the 4th finger. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1571-1573.	0.7	8
121	Molecular characterization of two patients with de novo interstitial deletions in 4q22-q24. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1830-1833.	0.7	4
122	A balanced chromosomal translocation disrupting <i>ARHGEF9</i> is associated with epilepsy, anxiety, aggression, and mental retardation. <i>Human Mutation</i> , 2009, 30, 61-68.	1.1	131
123	Genome-wide Gene Expression Profiling of SCID Mice with T cell-mediated Colitis. <i>Scandinavian Journal of Immunology</i> , 2009, 69, 437-446.	1.3	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. <i>European Journal of Human Genetics</i> , 2009, 17, 1010-1018.	1.4	20
125	Sequence assembly. <i>Computational Biology and Chemistry</i> , 2009, 33, 121-136.	1.1	39
126	Duplications Involving a Conserved Regulatory Element Downstream of BMP2 Are Associated with Brachydactyly Type A2. <i>American Journal of Human Genetics</i> , 2009, 84, 483-492.	2.6	139

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127	Stones in the lacrimal gland: a rare condition. <i>Acta Ophthalmologica</i> , 2009, 87, 672-675.	0.6	20
128	A novel mutation in <i>IRF6</i> resulting in VWSâ€‘PPS spectrum disorder with renal aplasia. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1605-1608.	0.7	12
129	A cryptic unbalanced translocation resulting in del 13q and dup 15q. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2570-2573.	0.7	2
130	Investigation of 4qâ€‘deletion in two unrelated patients using array CGH. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2431-2434.	0.7	17
131	A 3.2â€‘Mb deletion on 18q12 in a patient with childhood autism and high-grade myopia. <i>European Journal of Human Genetics</i> , 2008, 16, 312-319.	1.4	15
132	Balanced translocation in a patient with severe myoclonic epilepsy of infancy disrupts the sodium channel gene <i>SCN1A</i> . <i>Epilepsia</i> , 2008, 49, 1091-1094.	2.6	10
133	Mowatâ€‘Wilson syndrome: an underdiagnosed syndrome?. <i>Clinical Genetics</i> , 2008, 73, 579-584.	1.0	15
134	Truncation of the Down Syndrome Candidate Gene <i>DYRK1A</i> in Two Unrelated Patients with Microcephaly. <i>American Journal of Human Genetics</i> , 2008, 82, 1165-1170.	2.6	145
135	Fine mapping of a de novo interstitial 10q22â€‘q23 duplication in a patient with congenital heart disease and microcephaly. <i>European Journal of Medical Genetics</i> , 2008, 51, 81-86.	0.7	19
136	<i>GLI1</i> Is Involved in Cell Cycle Regulation and Proliferation of NT2 Embryonal Carcinoma Stem Cells. <i>DNA and Cell Biology</i> , 2008, 27, 251-256.	0.9	11
137	MicroRNA expression in the adult mouse central nervous system. <i>Rna</i> , 2008, 14, 432-444.	1.6	427
138	High frequency of submicroscopic genomic aberrations detected by tiling path array comparative genome hybridisation in patients with isolated congenital heart disease. <i>Journal of Medical Genetics</i> , 2008, 45, 704-709.	1.5	110
139	Comparative genomics beyond sequence-based alignments: RNA structures in the ENCODE regions. <i>Genome Research</i> , 2008, 18, 242-251.	2.4	82
140	Non-disjunction of chromosome 13. <i>Human Molecular Genetics</i> , 2007, 16, 2004-2010.	1.4	38
141	Pierre Robin sequence may be caused by dysregulation of <i>SOX9</i> and <i>KCNJ2</i> . <i>Journal of Medical Genetics</i> , 2007, 44, 381-386.	1.5	91
142	Genetic counseling in adult carriers of a balanced chromosomal rearrangement ascertained in childhood: Experiences from a nationwide reexamination of translocation carriers. <i>Genetics in Medicine</i> , 2007, 9, 185-187.	1.1	11
143	Recurrent Reciprocal Genomic Rearrangements of 17q12 Are Associated with Renal Disease, Diabetes, and Epilepsy. <i>American Journal of Human Genetics</i> , 2007, 81, 1057-1069.	2.6	222
144	Regional differences in expression of specific markers for human embryonic stem cells. <i>Reproductive BioMedicine Online</i> , 2007, 15, 89-98.	1.1	23

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145	Autoimmune diseases in a Danish cohort of 4,866 carriers of constitutional structural chromosomal rearrangements. <i>Arthritis and Rheumatism</i> , 2007, 56, 2402-2409.	6.7	8
146	Suggestive linkage to a neighboring region of <i>IRF6</i> in a cleft lip and palate multiplex family. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 2716-2721.	0.7	9
147	A human phenome-interactome network of protein complexes implicated in genetic disorders. <i>Nature Biotechnology</i> , 2007, 25, 309-316.	9.4	871
148	Detection of microRNAs in frozen tissue sections by fluorescence in situ hybridization using locked nucleic acid probes and tyramide signal amplification. <i>Nature Protocols</i> , 2007, 2, 2520-2528.	5.5	221
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