Niels Tommerup

List of Publications by Year in Descending Order

276 papers	16,618 citations	57 h-index	123 g-index
283 ext. papers	18,345 ext. citations	7.1 avg, IF	5.65 L-index

#	Paper	IF	Citations
276	Acute and persistent symptoms in non-hospitalized PCR-confirmed COVID-19 patients. *Scientific Reports*, **2021**, 11, 13153	4.9	27
275	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	2.6	1
274	A shared somatic translocation involving CUX1 in monozygotic twins as an early driver of AMKL in Down syndrome. *Blood Cancer Journal*, **2020**, 10, 27	7	3
273	Chromothripsis and DNA Repair Disorders. *Journal of Clinical Medicine*, **2020**, 9,	5.1	9
272	Nationwide germline whole genome sequencing of 198 consecutive pediatric cancer patients reveals a high incidence of cancer prone syndromes. *PLoS Genetics*, **2020**, 16, e1009231	6	16
271	RRP7A links primary microcephaly to dysfunction of ribosome biogenesis, resorption of primary cilia, and neurogenesis. *Nature Communications*, **2020**, 11, 5816	17.4	15
270	Paroxysmal Cranial Dyskinesia and Nail-Patella Syndrome Caused by a Novel Variant in the LMX1B Gene. *Movement Disorders*, **2020**, 35, 2343-2347	7	2
269	Nationwide germline whole genome sequencing of 198 consecutive pediatric cancer patients reveals a high incidence of cancer prone syndromes **2020**, 16, e1009231		
268	Nationwide germline whole genome sequencing of 198 consecutive pediatric cancer patients reveals a high incidence of cancer prone syndromes **2020**, 16, e1009231		
267	Nationwide germline whole genome sequencing of 198 consecutive pediatric cancer patients reveals a high incidence of cancer prone syndromes **2020**, 16, e1009231		
266	Nationwide germline whole genome sequencing of 198 consecutive pediatric cancer patients reveals a high incidence of cancer prone syndromes **2020**, 16, e1009231		
265	Multigenic truncation of the semaphorin-plexin pathway by a germline chromothriptic rearrangement associated with Moebius syndrome. *Human Mutation*, **2019**, 40, 1057-1062	4.7	3
264	A novel in-frame mutation in leads to Juvenile neuronal ceroid lipofuscinosis in a large Pakistani family. *International Journal of Neuroscience*, **2019**, 129, 890-895	2	
263	Haploinsufficiency of ARHGAP42 is associated with hypertension. *European Journal of Human Genetics*, **2019**, 27, 1296-1303	5.3	6
262	A splice-site variant in the lncRNA gene cosegregates in the large Volkmann cataract family. *Molecular Vision*, **2019**, 25, 1-11	2.3	1
261	Small supernumerary marker chromosomes: A legacy of trisomy rescue?. *Human Mutation*, **2019**, 40, 193-200	4.7	15
260	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies syndrome due to disruption of BPTF in a 35-year-old man initially diagnosed with Silver-Russell syndrome. *Clinical Genetics*, **2019**, 95, 534-536	4	3

259	Very short DNA segments can be detected and handled by the repair machinery during germline chromothriptic chromosome reassembly. *Human Mutation*, **2018**, 39, 709-716	4.7	15
258	Congenital olfactory impairment is linked to cortical changes in prefrontal and limbic brain regions. *Brain Imaging and Behavior*, **2018**, 12, 1569-1582	4.1	16
257	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	2.3	5
256	Regulatory variants of FOXG1 in the context of its topological domain organisation. *European Journal of Human Genetics*, **2018**, 26, 186-196	5.3	9
255	Cryptic breakpoint identified by whole-genome mate-pair sequencing in a rare paternally inherited complex chromosomal rearrangement. *Molecular Cytogenetics*, **2018**, 11, 34	2	6
254	Challenges for the Sustainability of University-Run Biobanks. *Biopreservation and Biobanking*, **2018**, 16, 312-321	2.1	10
253	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. *PLoS Genetics*, **2018**, 14, e1007780	6	22
252	De novo unbalanced translocations have a complex history/aetiology. *Human Genetics*, **2018**, 137, 817-829	4.3	13
251	Position effect, cryptic complexity, and direct gene disruption as disease mechanisms in de novo apparently balanced translocation cases. *PLoS ONE*, **2018**, 13, e0205298	3.7	9
250	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	11	19
249	The identification and functional annotation of RNA structures conserved in vertebrates. *Genome Research*, **2017**, 27, 1371-1383	9.7	46
248	Abdominal Wall Defects in Greenland 1989-2015. *Birth Defects Research*, **2017**, 109, 836-842	2.9	10
247	Mutations in GABRB3: From febrile seizures to epileptic encephalopathies. *Neurology*, **2017**, 88, 483-492	6.5	68
246	Genome-Wide Supported Risk Variants in , , , , and Contribute to Schizophrenia Susceptibility in Pakistani Population. *Psychiatry Investigation*, **2017**, 14, 687-692	3.1	10
245	Homozygous mutation in the NPHP3 gene causing foetal nephronophthisis. *Nephrology*, **2017**, 22, 818-820	2.2	2
244	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	2.6	6
243	Enrichment of megabase-sized DNA molecules for single-molecule optical mapping and next-generation sequencing. *Scientific Reports*, **2017**, 7, 17893	4.9	2
242	Accurate Breakpoint Mapping in Apparently Balanced Translocation Families with Discordant Phenotypes Using Whole Genome Mate-Pair Sequencing. *PLoS ONE*, **2017**, 12, e0169935	3.7	17

241	Gene Panel Testing in Epileptic Encephalopathies and Familial Epilepsies. *Molecular Syndromology*, **2016**, 7, 210-219	1.5	65
240	Monozygotic twins discordant for narcolepsy type 1 and multiple sclerosis. *Neurology: Neuroimmunology and NeuroInflammation*, **2016**, 3, e249	9.1	3
239	Phenotypic subregions within the split-hand/foot malformation 1 locus. *Human Genetics*, **2016**, 135, 345-57	4.3	11
238	Identification of the BRD1 interaction network and its impact on mental disorder risk. *Genome Medicine*, **2016**, 8, 53	14.4	23
237	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. *European Journal of Human Genetics*, **2016**, 24, 1761-1770	5.3	23
236	A novel splice site mutation in CEP135 is associated with primary microcephaly in a Pakistani family. *Journal of Human Genetics*, **2016**, 61, 271-3	4.3	14
235	Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. *Annals of Neurology*, **2016**, 79, 428-36	9.4	124
234	Germline Chromothripsis Driven by L1-Mediated Retrotransposition and Alu/Alu Homologous Recombination. *Human Mutation*, **2016**, 37, 385-95	4.7	40
233	Regulatory Mutations of FOXG1 in the Context of Topological Domains. *Cancer Genetics*, **2016**, 209, 245	2.3	
232	Genome-wide DNA methylation analysis of transient neonatal diabetes type 1 patients with mutations in ZFP57. *BMC Medical Genetics*, **2016**, 17, 29	2.1	21
231	Automation of a single-DNA molecule stretching device. *Review of Scientific Instruments*, **2015**, 86, 063702	1.27	7
230	The small RNA content of human sperm reveals pseudogene-derived piRNAs complementary to protein-coding genes. *Rna*, **2015**, 21, 1085-95	5.8	59
229	Partial USH2A deletions contribute to Usher syndrome in Denmark. *European Journal of Human Genetics*, **2015**, 23, 1646-51	5.3	5
228	Optical mapping of single-molecule human DNA in disposable, mass-produced all-polymer devices. *Journal of Micromechanics and Microengineering*, **2015**, 25, 105002	2	15
227	The first mutation in CNGA2 in two brothers with anosmia. *Clinical Genetics*, **2015**, 88, 293-6	4	17
226	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, 650-3	2.6	10
225	The segregation of different submicroscopic imbalances underlying the clinical variability associated with a familial karyotypically balanced translocation. *Molecular Cytogenetics*, **2015**, 8, 106	2	5
224	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	14.4	12

223	The role of SLC2A1 mutations in myoclonic astatic epilepsy and absence epilepsy, and the estimated frequency of GLUT1 deficiency syndrome. *Epilepsia*, **2015**, 56, e203-8	6.4	52
222	A Novel Locus Harbouring a Functional CD164 Nonsense Mutation Identified in a Large Danish Family with Nonsyndromic Hearing Impairment. *PLoS Genetics*, **2015**, 11, e1005386	6	13
221	Dysregulation of FOXG1 by ring chromosome 14. *Molecular Cytogenetics*, **2015**, 8, 24	2	6
220	The phenotypic spectrum of SCN8A encephalopathy. *Neurology*, **2015**, 84, 480-9	6.5	199
219	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, 338-43	5.3	38
218	Aberrant expression of miR-218 and miR-204 in human mesial temporal lobe epilepsy and hippocampal sclerosis-convergence on axonal guidance. *Epilepsia*, **2014**, 55, 2017-27	6.4	51
217	Neural correlates of taste perception in congenital olfactory impairment. *Neuropsychologia*, **2014**, 62, 297-305	3.2	16
216	Neurodevelopmental disorders associated with dosage imbalance of ZBTB20 correlate with the morbidity spectrum of ZBTB20 candidate target genes. *Journal of Medical Genetics*, **2014**, 51, 605-13	5.8	23
215	Sequence analysis of 17 NRXN1 deletions. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2014**, 165B, 52-61	3.5	10
214	Structural genomic variation in childhood epilepsies with complex phenotypes. *European Journal of Human Genetics*, **2014**, 22, 896-901	5.3	24
213	The myosin chaperone UNC45B is involved in lens development and autosomal dominant juvenile cataract. *European Journal of Human Genetics*, **2014**, 22, 1290-7	5.3	23
212	Reduced ceramide synthase 2 activity causes progressive myoclonic epilepsy. *Annals of Clinical and Translational Neurology*, **2014**, 1, 88-98	5.3	42
211	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. *Human Molecular Genetics*, **2014**, 23, 6163-76	5.6	16
210	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, 625-38	6.3	14
209	Hypomorphic mutations in PGAP2, encoding a GPI-anchor-remodeling protein, cause autosomal-recessive intellectual disability. *American Journal of Human Genetics*, **2013**, 92, 575-83	11	79
208	Mutations in SYNGAP1 cause intellectual disability, autism, and a specific form of epilepsy by inducing haploinsufficiency. *Human Mutation*, **2013**, 34, 385-94	4.7	126
207	The role of SLC2A1 in early onset and childhood absence epilepsies. *Epilepsy Research*, **2013**, 105, 229-33	3	12
206	Genetic heterogeneity in Pakistani microcephaly families. *Clinical Genetics*, **2013**, 83, 446-51	4	37

205	An association study between the norepinephrine transporter gene and depression. *Psychiatric Genetics*, **2013**, 23, 217-21	2.9	4
204	Exon-disrupting deletions of NRXN1 in idiopathic generalized epilepsy. *Epilepsia*, **2013**, 54, 256-64	6.4	48
203	Next-generation sequencing: proof of concept for antenatal prediction of the fetal Kell blood group phenotype from cell-free fetal DNA in maternal plasma. *Transfusion*, **2013**, 53, 2892-8	2.9	24
202	Dysfunction of the Heteromeric KV7.3/KV7.5 Potassium Channel is Associated with Autism Spectrum Disorders. *Frontiers in Genetics*, **2013**, 4, 54	4.5	34
201	Corpus callosum abnormalities, intellectual disability, speech impairment, and autism in patients with haploinsufficiency of ARID1B. *Clinical Genetics*, **2012**, 82, 248-55	4	104
200	No evidence for pathogenic variants or maternal effect of ZFP57 as the cause of Beckwith-Wiedemann Syndrome. *European Journal of Human Genetics*, **2012**, 20, 119-21	5.3	13
199	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	11	42
198	Profiling microRNAs in lung tissue from pigs infected with Actinobacillus pleuropneumoniae. *BMC Genomics*, **2012**, 13, 459	4.5	47
197	Genetic association studies in lumbar disc degeneration: a systematic review. *PLoS ONE*, **2012**, 7, e49995	3.7	67
196	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	4.5	15
195	Screening of congenital heart disease patients using multiplex ligation-dependent probe amplification: early diagnosis of syndromic patients. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 720-5	2.5	24
194	Genetic studies in congenital anterior midline cervical cleft. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2021-6	2.5	12
193	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, 354-8	3.5	51
192	Isolated and syndromic forms of congenital anosmia. *Clinical Genetics*, **2012**, 81, 210-5	4	47
191	REST-mediated recruitment of polycomb repressor complexes in mammalian cells. *PLoS Genetics*, **2012**, 8, e1002494	6	116
190	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, 14035-40	11.5	90
189	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, 1315-9	5.3	23
188	Sequence and expression analysis of gaps in human chromosome 20. *Nucleic Acids Research*, **2012**, 40, 6660-72	20.1	4

187	Biparental inheritance of chromosomal abnormalities in male twins with non-syndromic mental retardation. *European Journal of Medical Genetics*, **2011**, 54, e383-8	2.6	7
186	The lrrk2 p.Gly2019Ser mutation is uncommon in a Danish cohort with various neurodegenerative disorders. *Parkinsonism and Related Disorders*, **2011**, 17, 398-9	3.6	4
185	A balanced translocation disrupts SYNGAP1 in a patient with intellectual disability, speech impairment, and epilepsy with myoclonic absences (EMA). *Epilepsia*, **2011**, 52, e190-3	6.4	20
184	Duplication of MAOA, MAOB, and NDP in a patient with mental retardation and epilepsy. *European Journal of Human Genetics*, **2011**, 19, 1-2	5.3	9
183	Cost-effective multiplexing before capture allows screening of 25 000 clinically relevant SNPs in childhood acute lymphoblastic leukemia. *Leukemia*, **2011**, 25, 1001-6	10.7	17
182	Autozygosity mapping of a large consanguineous Pakistani family reveals a novel non-syndromic autosomal recessive mental retardation locus on 11p15-tel. *Neurogenetics*, **2011**, 12, 247-51	3	2
181	A cohort of balanced reciprocal translocations associated with dyslexia: identification of two putative candidate genes at DYX1. *Behavior Genetics*, **2011**, 41, 125-33	3.2	12
180	Interstitial deletion of 14q24.3-q32.2 in a male patient with plagiocephaly, BPES features, developmental delay, and congenital heart defects. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 203-6	2.5	6
179	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**, 155A, 652-5	2.5	6
178	High frequency of rare copy number variants affecting functionally related genes in patients with structural brain malformations. *Human Mutation*, **2011**, 32, 1427-35	4.7	20
177	Genome-wide detection of chromosomal rearrangements, indels, and mutations in circular chromosomes by short read sequencing. *Genome Research*, **2011**, 21, 1388-93	9.7	60
176	Jarid1b targets genes regulating development and is involved in neural differentiation. *EMBO Journal*, **2011**, 30, 4586-600	13	135
175	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, 733-6	5.3	40
174	JARID2 regulates binding of the Polycomb repressive complex 2 to target genes in ES cells. *Nature*, **2010**, 464, 306-10	50.4	427
173	Ancient human genome sequence of an extinct Palaeo-Eskimo. *Nature*, **2010**, 463, 757-62	50.4	567
172	Single-molecule denaturation mapping of DNA in nanofluidic channels. *Proceedings of the National Academy of Sciences of the United States of America*, **2010**, 107, 13294-9	11.5	167
171	Genome-wide analysis of CDX2 binding in intestinal epithelial cells (Caco-2). *Journal of Biological Chemistry*, **2010**, 285, 25115-25	5.4	57
170	Metaphase FISH on a chip: miniaturized microfluidic device for fluorescence in situ hybridization. *Sensors*, **2010**, 10, 9831-46	3.8	27

169	A functional link between the histone demethylase PHF8 and the transcription factor ZNF711 in X-linked mental retardation. *Molecular Cell*, **2010**, 38, 165-78	17.6	162
168	Dissecting spatio-temporal protein networks driving human heart development and related disorders. *Molecular Systems Biology*, **2010**, 6, 381	12.2	72
167	Haploinsufficiency of TAB2 causes congenital heart defects in humans. *American Journal of Human Genetics*, **2010**, 86, 839-49	11	75
166	Craniosynostosis-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, 495-7	2.5	17
165	Deletion of 7q34-q36.2 in two siblings with mental retardation, language delay, primary amenorrhea, and dysmorphic features. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 3115-9	2.5	31
164	Autoimmune diseases in women with Turner's syndrome. *Arthritis and Rheumatism*, **2010**, 62, 658-66		111
163	A mutation in the FOXE3 gene causes congenital primary aphakia in an autosomal recessive consanguineous Pakistani family. *Molecular Vision*, **2010**, 16, 549-55	2.3	20
162	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, 77-85	12.8	15
161	Compound heterozygous ASPM mutations in Pakistani MCPH families. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 926-30	2.5	24
160	9q Subtelomeric deletion syndrome with diaphragmatic hernia. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1086-8	2.5	
159	A novel subtype of distal symphalangism affecting only the 4th finger. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1571-3	2.5	4
158	Molecular characterization of two patients with de novo interstitial deletions in 4q22-q24. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1830-3	2.5	2
157	A balanced chromosomal translocation disrupting ARHGEF9 is associated with epilepsy, anxiety, aggression, and mental retardation. *Human Mutation*, **2009**, 30, 61-8	4.7	108
156	Genome-wide gene expression profiling of SCID mice with T-cell-mediated Colitis. *Scandinavian Journal of Immunology*, **2009**, 69, 437-46	3.4	4
155	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, 1010-8	5.3	15
154	Sequence assembly. *Computational Biology and Chemistry*, **2009**, 33, 121-36	3.6	37
153	Duplications involving a conserved regulatory element downstream of BMP2 are associated with brachydactyly type A2. *American Journal of Human Genetics*, **2009**, 84, 483-92	11	122
152	Stones in the lacrimal gland: a rare condition. *Acta Ophthalmologica*, **2009**, 87, 672-5	3.7	13

151	A 3.2 Mb deletion on 18q12 in a patient with childhood autism and high-grade myopia. *European Journal of Human Genetics*, **2008**, 16, 312-9	5.3	14
150	Balanced translocation in a patient with severe myoclonic epilepsy of infancy disrupts the sodium channel gene SCN1A. *Epilepsia*, **2008**, 49, 1091-4	6.4	10
149	Mowat-Wilson syndrome: an underdiagnosed syndrome?. *Clinical Genetics*, **2008**, 73, 579-84	4	13
148	Truncation of the Down syndrome candidate gene DYRK1A in two unrelated patients with microcephaly. *American Journal of Human Genetics*, **2008**, 82, 1165-70	11	118
147	Fine mapping of a de novo interstitial 10q22-q23 duplication in a patient with congenital heart disease and microcephaly. *European Journal of Medical Genetics*, **2008**, 51, 81-6	2.6	15
146	GLI1 is involved in cell cycle regulation and proliferation of NT2 embryonal carcinoma stem cells. *DNA and Cell Biology*, **2008**, 27, 251-6	3.6	9
145	MicroRNA expression in the adult mouse central nervous system. *Rna*, **2008**, 14, 432-44	5.8	360
144	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, 704-9	5.8	102
143	Comparative genomics beyond sequence-based alignments: RNA structures in the ENCODE regions. *Genome Research*, **2008**, 18, 242-51	9.7	76
142	A novel mutation in IRF6 resulting in VWS-PPS spectrum disorder with renal aplasia. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1605-8	2.5	11
141	A cryptic unbalanced translocation resulting in del 13q and dup 15q. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2570-3	2.5	2
140	Investigation of 4q-deletion in two unrelated patients using array CGH. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2431-4	2.5	16
139	Regional differences in expression of specific markers for human embryonic stem cells. *Reproductive BioMedicine Online*, **2007**, 15, 89-98	4	18
138	Autoimmune diseases in a Danish cohort of 4,866 carriers of constitutional structural chromosomal rearrangements. *Arthritis and Rheumatism*, **2007**, 56, 2402-9		8
137	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, 2716-21	2.5	6
136	A human phenome-interactome network of protein complexes implicated in genetic disorders. *Nature Biotechnology*, **2007**, 25, 309-16	44.5	761
135	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, 2520-8	18.8	204
134	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, 711-3	5.3	63

133	Cytogenetically invisible microdeletions involving PITX2 in Rieger syndrome. *Clinical Genetics*, **2007**, 72, 464-70	4	15
132	Interstitial deletion of chromosome 4p associated with mild mental retardation, epilepsy and polymicrogyria of the left temporal lobe. *Clinical Genetics*, **2007**, 72, 593-8	4	9
131	Mutations in autism susceptibility candidate 2 (AUTS2) in patients with mental retardation. *Human Genetics*, **2007**, 121, 501-9	6.3	97
130	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, 207-21	4.2	21
129	Non-disjunction of chromosome 13. *Human Molecular Genetics*, **2007**, 16, 2004-10	5.6	32
128	Pierre Robin sequence may be caused by dysregulation of SOX9 and KCNJ2. *Journal of Medical Genetics*, **2007**, 44, 381-6	5.8	75
127	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, 185-7	8.1	9
126	Recurrent reciprocal genomic rearrangements of 17q12 are associated with renal disease, diabetes, and epilepsy. *American Journal of Human Genetics*, **2007**, 81, 1057-69	11	193
125	Population-based study of cancer among carriers of a constitutional structural chromosomal rearrangement. *Genes Chromosomes and Cancer*, **2006**, 45, 231-46	5	11
124	Delineation of a 2.2 Mb microdeletion at 5q35 associated with microcephaly and congenital heart disease. *American Journal of Medical Genetics, Part A*, **2006**, 140, 427-33	2.5	26
123	Additional chromosomal abnormalities in patients with a previously detected abnormal karyotype, mental retardation, and dysmorphic features. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2180-7	2.5	45
122	4q35 deletion and 10p15 duplication associated with immunodeficiency. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2231-5	2.5	9
121	Screening of 99 Danish patients with congenital heart disease for GATA4 mutations. *Genetic Testing and Molecular Biomarkers*, **2006**, 10, 277-80		16
120	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	11	19
119	Hedgehog signaling in small-cell lung cancer: frequent in vivo but a rare event in vitro. *Lung Cancer*, **2006**, 52, 281-90	5.9	71
118	A novel primate specific gene, CEI, is located in the homeobox gene IRXA2 promoter in Homo sapiens. *Gene*, **2006**, 371, 167-73	3.8	7
117	The genetic basis of the Pierre Robin Sequence. *Cleft Palate-Craniofacial Journal*, **2006**, 43, 155-9	1.9	82
116	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, 410-7	5.3	15

115	Evaluation of two methods for generating cRNA for microarray experiments from nanogram amounts of total RNA. *Analytical Biochemistry*, **2006**, 358, 111-9	3.1	5
114	Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. *Human Genetics*, **2006**, 118, 578-90	6.3	44
113	Molecular characterization of a balanced chromosome translocation in psoriasis vulgaris. *Clinical Genetics*, **2006**, 69, 189-93	4	7
112	Breakpoints around the HOXD cluster result in various limb malformations. *Journal of Medical Genetics*, **2006**, 43, 111-8	5.8	39
111	A mutation in the receptor binding site of GDF5 causes Mohr-Wriedt brachydactyly type A2. *Journal of Medical Genetics*, **2006**, 43, 225-31	5.8	22
110	Disruption of Netrin G1 by a balanced chromosome translocation in a girl with Rett syndrome. *European Journal of Human Genetics*, **2005**, 13, 921-7	5.3	78
109	Delineation of an interstitial 9q22 deletion in basal cell nevus syndrome **2005**, 132A, 324-8		27
108	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**, 135, 339-41	2.5	4
107	Male-to-male transmission in Laurin-Sandrow syndrome and exclusion of RARB and RARG. *American Journal of Medical Genetics, Part A*, **2005**, 137, 148-52	2.5	5
106	A 72-year-old Danish puzzle resolved--comparative analysis of phenotypes in families with different-sized HOXD13 polyalanine expansions. *American Journal of Medical Genetics, Part A*, **2005**, 138, 328-39	2.5	17
105	Haploinsufficiency of novel FOXG1B variants in a patient with severe mental retardation, brain malformations and microcephaly. *Human Genetics*, **2005**, 117, 536-44	6.3	83
104	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	20
103	Distinct CDH3 mutations cause ectodermal dysplasia, ectrodactyly, macular dystrophy (EEM syndrome). *Journal of Medical Genetics*, **2005**, 42, 292-8	5.8	86
102	Identification of a novel EYA1 splice-site mutation in a Danish branchio-oto-renal syndrome family. *Genetic Testing and Molecular Biomarkers*, **2004**, 8, 404-6		6
101	Sequencing and mapping of the porcine CCS gene. *Animal Genetics*, **2004**, 35, 353-4	2.5	
100	An excess of chromosome 1 breakpoints in male infertility. *European Journal of Human Genetics*, **2004**, 12, 993-1000	5.3	48
99	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, 179-91		29
98	Novel Connexin 43 (GJA1) mutation causes oculo-dento-digital dysplasia with curly hair. *American Journal of Medical Genetics Part A*, **2004**, 127A, 152-7		77

97	Molecular cytogenetic characterization of ring chromosome 15 in three unrelated patients **2004**, 130A, 340-4		49
96	LNA-modified oligonucleotides are highly efficient as FISH probes. *Cytogenetic and Genome Research*, **2004**, 107, 32-7	1.9	47
95	Mutation analysis of the Sonic hedgehog promoter and putative enhancer elements in Parkinson's disease patients. *Molecular Brain Research*, **2004**, 126, 207-11		4
94	The Hedgehog signaling pathway--implications for drug targets in cancer and neurodegenerative disorders. *Pharmacogenomics*, **2003**, 4, 411-29	2.6	25
93	Mutational analysis of the human FATE gene in 144 infertile men. *Human Genetics*, **2003**, 113, 195-201	6.3	35
92	Disruption of the serine/threonine kinase 9 gene causes severe X-linked infantile spasms and mental retardation. *American Journal of Human Genetics*, **2003**, 72, 1401-11	11	227
91	FISHing with locked nucleic acids (LNA): evaluation of different LNA/DNA mixmers. *Molecular and Cellular Probes*, **2003**, 17, 165-9	3.3	55
90	Human CCS gene: genomic organization and exclusion as a candidate for amyotrophic lateral sclerosis (ALS). *BMC Genetics*, **2002**, 3, 5	2.6	9
89	HOXD13 polyalanine tract expansion in classical synpolydactyly type Vordingborg. *American Journal of Medical Genetics Part A*, **2002**, 110, 116-21		22
88	Expression and post-translational modification of human 4-hydroxy-phenylpyruvate dioxygenase. *Cell Biology International*, **2002**, 26, 615-25	4.5	5
87	Detection of illegitimate rearrangements within the immunoglobulin light chain loci in B cell malignancies using end sequenced probes. *Leukemia*, **2002**, 16, 2148-55	10.7	7
86	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 12q13-14 (IBD2). *Gene*, **2002**, 288, 179-85	3.8	16
85	The human hedgehog-interacting protein gene: structure and chromosome mapping to 4q31.21-->q31.3. *Cytogenetic and Genome Research*, **2001**, 92, 300-3	1.9	25
84	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, 265-74	5	11
83	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, 178-84	5.3	57
82	Genetic linkage of autosomal dominant primary open angle glaucoma to chromosome 3q in a Greek pedigree. *European Journal of Human Genetics*, **2001**, 9, 452-7	5.3	25
81	Identification of human candidate genes for male infertility by digital differential display. *Molecular Human Reproduction*, **2001**, 7, 11-20	4.4	27
80	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	4.3	20

79	Human FATE is a novel X-linked gene expressed in fetal and adult testis. *Molecular and Cellular Endocrinology*, **2001**, 184, 25-32	4.4	25
78	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, 661-8	5.3	41
77	Molecular cloning of Xp11 breakpoints in two unrelated mentally retarded females with X;autosome translocations. *Cytogenetic and Genome Research*, **2000**, 90, 126-33	1.9	5
76	Assignment 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, 156-7	1.9	4
75	Assignment of the NR2E3 gene to mouse chromosome 9 and to human chromosome 15q22.33-->q23. *Cytogenetic and Genome Research*, **2000**, 89, 279-80	1.9	
74	Filter-grown TR146 cells as an in vitro model of human buccal epithelial permeability. *European Journal of Oral Sciences*, **1999**, 107, 138-46	2.3	53
73	Psoriasis upregulated phorbolin-1 shares structural but not functional similarity to the mRNA-editing protein apobec-1. *Journal of Investigative Dermatology*, **1999**, 113, 162-9	4.3	45
72	Chromosome instability and immunodeficiency syndrome caused by mutations in a DNA methyltransferase gene. *Nature*, **1999**, 402, 187-91	50.4	928
71	Molecular cytogenetic detection of 9q34 breakpoints associated with nail patella syndrome. *European Journal of Human Genetics*, **1999**, 7, 68-76	5.3	16
70	Structural organization, tissue expression, and chromosomal localization of Ciao 1, a functional modulator of the WilmsPtumor suppressor, WT1. *Immunogenetics*, **1999**, 49, 900-905	3.2	1
69	PCR-based screening of YAC clones without DNA extraction. *Technical Tips Online*, **1999**, 4, 1-3		
68	Sequence variants in the human cocaine and amphetamine-regulated transcript (CART) gene in subjects with early onset obesity. *Obesity*, **1999**, 7, 532-6		31
67	BAP1: a novel ubiquitin hydrolase which binds to the BRCA1 RING finger and enhances BRCA1-mediated cell growth suppression. *Oncogene*, **1998**, 16, 1097-112	9.2	545
66	Occurrence of cancer in a cohort of 183 persons with constitutional chromosome 7 abnormalities. *Cancer Genetics and Cytogenetics*, **1998**, 105, 39-42		21
65	Prader-Willi□ke phenotype and the proximal long arm of the X chromosome. *American Journal of Medical Genetics Part A*, **1998**, 80, 300-301		3
64	A neocentromere on human chromosome 3 without detectable alpha-satellite DNA forms morphologically normal kinetochores. *Chromosoma*, **1998**, 107, 359-65	2.8	42
63	Human rab11a: transcription, chromosome mapping and effect on the expression levels of host GTP-binding proteins. *FEBS Letters*, **1998**, 429, 359-64	3.8	10
62	Isolation of the human beaded-filament structural protein 1 gene (BFSP1) and assignment to chromosome 20p11.23-p12.1. *Genomics*, **1998**, 53, 114-6	4.3	3

61	Mapping of the human PAWR (par-4) gene to chromosome 12q21. *Genomics*, **1998**, 53, 241-3	4.3	44
60	Ciao 1 is a novel WD40 protein that interacts with the tumor suppressor protein WT1. *Journal of Biological Chemistry*, **1998**, 273, 10880-7	5.4	54
59	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	2.2	137
58	Molecular identification of a novel candidate sorting receptor purified from human brain by receptor-associated protein affinity chromatography. *Journal of Biological Chemistry*, **1997**, 272, 3599-605	5.4	322
57	Linkage mapping in 29 Bardet-Biedl syndrome families confirms loci in chromosomal regions 11q13, 15q22.3-q23, and 16q21. *Genomics*, **1997**, 41, 93-9	4.3	115
56	Prolonged extreme thrombocytosis associated with neurofibromatosis type 1. *Journal of Pediatrics*, **1997**, 130, 317-9	3.6	4
55	Investigation of deletions at 7q11.23 in 44 patients referred for Williams-Beuren syndrome, using FISH and four DNA polymorphisms. *Human Genetics*, **1997**, 99, 56-61	6.3	16
54	Assignment of human KH-box-containing genes by in situ hybridization: HNRNPK maps to 9q21.32-q21.33, PCBP1 to 2p12-p13, and PCBP2 to 12q13.12-q13.13, distal to FRA12A. *Genomics*, **1996**, 32, 297-8	4.3	22
53	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 and in situ hybridization. *Genomics*, **1996**, 33, 143-5	4.3	3
52	Assignment of the human genes encoding 14,3-3 Eta (YWHAH) to 22q12, 14-3-3 zeta (YWHAZ) to 2p25.1-p25.2, and 14-3-3 beta (YWHAB) to 20q13.1 by in situ hybridization. *Genomics*, **1996**, 33, 149-50	4.3	21
51	Assignment of human elongation factor 1alpha genes: EEF1A maps to chromosome 6q14 and EEF1A2 to 20q13.3. *Genomics*, **1996**, 36, 359-61	4.3	71
50	Exclusion of SNRPN as a major determinant of Prader-Willi syndrome by a translocation breakpoint. *Nature Genetics*, **1996**, 12, 452-4	36.3	64
49	Molecular characterization of a novel human hybrid-type receptor that binds the alpha2-macroglobulin receptor-associated protein. *Journal of Biological Chemistry*, **1996**, 271, 31379-83	5.4	194
48	Tetrasomy 18p de novo: parental origin and different mechanisms of formation. *European Journal of Human Genetics*, **1996**, 4, 160-7	5.3	29
47	No mutations found by RET mutation scanning in sporadic and hereditary neuroblastoma. *Human Genetics*, **1996**, 97, 362-364	6.3	
46	Partial deletion 11q: report of a case with a large terminal deletion 11q21-qter without loss of telomeric sequences, and review of the literature. *Clinical Genetics*, **1995**, 47, 231-5	4	13
45	Rubinstein-Taybi syndrome caused by mutations in the transcriptional co-activator CBP. *Nature*, **1995**, 376, 348-51	50.4	1018
44	Isolation and fine mapping of 16 novel human zinc finger-encoding cDNAs identify putative candidate genes for developmental and malignant disorders. *Genomics*, **1995**, 27, 259-64	4.3	117

43	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, 210-4		52
42	Obstetrical and gynecological complications in fragile X carriers: a multicenter study. *American Journal of Medical Genetics Part A*, **1994**, 51, 400-2		178
41	Deletions of a differentially methylated CpG island at the SNRPN gene define a putative imprinting control region. *Nature Genetics*, **1994**, 8, 52-8	36.3	388
40	Autosomal sex reversal and campomelic dysplasia are caused by mutations in and around the SRY-related gene SOX9. *Cell*, **1994**, 79, 1111-20	56.2	1293
39	Localization of the human gene for advanced glycosylation end product-specific receptor (AGER) to chromosome 6p21.3. *Genomics*, **1994**, 24, 606-8	4.3	25
38	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, 1571-5	5.6	44
37	Isolation of a candidate gene for Menkes disease that encodes a potential heavy metal binding protein. *Nature Genetics*, **1993**, 3, 14-9	36.3	645
36	Assignment of an autosomal sex reversal locus (SRA1) and campomelic dysplasia (CMPD1) to 17q24.3-q25.1. *Nature Genetics*, **1993**, 4, 170-4	36.3	180
35	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, 339-46	6.3	11
34	Characterization of a 1.0 Mb YAC contig spanning two chromosome breakpoints related to Menkes disease. *Human Molecular Genetics*, **1992**, 1, 483-9	5.6	30
33	A holder for critical point drying of large numbers of EM-grids. *Microscopy Research and Technique*, **1992**, 23, 353-4	2.8	1
32	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, 237-41		34
31	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, 534-8		20
30	Direct diagnosis by DNA analysis of the fragile X syndrome of mental retardation. *New England Journal of Medicine*, **1991**, 325, 1673-81	59.2	583
29	Chemotaxis in tetrahymena. *European Journal of Protistology*, **1990**, 25, 229-33	3.6	6
28	Localization in man of fifteen DNA sequences within the chromosome segment 13q12-q22. *Hereditas*, **1989**, 110, 253-65	2.4	4
27	Induction of the fragile X on BrdU-substituted chromosomes with direct visualization of sister chromatid exchanges on banded chromosomes. *Human Genetics*, **1989**, 81, 377-81	6.3	4
26	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, 739-53		32

25	Screening for the fragile X: how many cells should we analyse?. *American Journal of Medical Genetics Part A*, **1988**, 30, 417-22		5
24	Induction of the fra(X) in amniotic fluid cells by excess thymidine. *American Journal of Medical Genetics Part A*, **1988**, 30, 451-3		2
23	DNA-aneuploidy in 46,XX hydatidiform moles. *Cancer Genetics and Cytogenetics*, **1987**, 27, 225-8		3
22	Screening test for ataxia telangiectasia. *Lancet, The*, **1987**, 2, 1398-9	40	18
21	Second trimester prenatal diagnosis of the fragile X. *American Journal of Medical Genetics Part A*, **1986**, 23, 313-24		15
20	Fragile X: carrier detection in pregnancy. *American Journal of Medical Genetics Part A*, **1986**, 23, 527-30		1
19	CHEMOATTRACTION INTETRAHYMENA: ON THE ROLE OF CHEMOKINESIS. *Biological Bulletin*, **1986** , 170, 357-367	1.5	38
18	Hydatidiform moles: methods for culture and cytogenetic analyses. *Cancer Genetics and Cytogenetics*, **1986**, 22, 19-27		10
17	Hydatidiform mole: a chromosomal search for a recessive mutation. *Human Reproduction*, **1986**, 1, 337-40	3.7	
16	A folate sensitive heritable fragile site at 19p13. *Clinical Genetics*, **1985**, 27, 510-4	4	11
15	High resolution chromosomes from first trimester trophoblast cultures. *Prenatal Diagnosis*, **1985**, 5, 291-4	3.2	7
14	Genetic instability of cell lines derived from a single human small cell carcinoma of the lung. *European Journal of Cancer & Clinical Oncology*, **1985**, 21, 815-24		46
13	Identification of triploidy by DA/DAPI staining of trophoblastic interphase nuclei. *Placenta*, **1985**, 6, 363-7	3.4	5
12	Chromosomal studies in familial polyposis coli. *Cancer Genetics and Cytogenetics*, **1985**, 17, 355-7		8
11	Blood group substances, T6 antigen and heterochromatin pattern as species markers in the nude mouse/human skin model. *Pathobiology*, **1984**, 52, 251-9	3.6	1
10	Folic acid metabolism in a patient with fragile X. *Clinical Genetics*, **1983**, 24, 153-5	4	11
9	Fragile X demonstrated retrospectively in amniotic cells cultured in low folate medium. *Prenatal Diagnosis*, **1983**, 3, 367-9	3.2	5
8	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, 313-21		59

7	Specific staining of 9h in human somatic interphase cells by D 287/170. *Human Genetics*, **1982**, 62, 301-4	6.3	3
6	Macroorchidism and fragile X in mentally retarded males. Clinical, cytogenetic, and some hormonal investigations in mentally retarded males, including two with the fragile site at Xq28, fra(X)(q28). *Human Genetics*, **1982**, 61, 113-7	6.3	46
5	Apparent homozygosity for the fragile site at Xq28 in a normal female. *Human Genetics*, **1982**, 61, 60-2	6.3	14
4	Marker X chromosome induction in fibroblasts by FUdR. *American Journal of Medical Genetics Part A* , **1981**, 9, 263-4		51
3	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, 23-5	6.3	68
2	Macroorchidism, mental retardation, and the fragile X. *New England Journal of Medicine*, **1981**, 305, 1348	59.2	13
1	Chromothripsis and Human Genetic Disease1-10		5