# Niels Tommerup

## List of Publications by Citations



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| 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 | 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 |
| 275 | Rubinstein-Taybi syndrome caused by mutations in the transcriptional co-activator CBP. *Nature*, **1995**, 376, 348-51 | 50.4 | 1018 |
| 274 | Chromosome instability and immunodeficiency syndrome caused by mutations in a DNA methyltransferase gene. *Nature*, **1999**, 402, 187-91 | 50.4 | 928 |
| 273 | A human phenome-interactome network of protein complexes implicated in genetic disorders. *Nature Biotechnology*, **2007**, 25, 309-16 | 44.5 | 761 |
| 272 | 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 |
| 271 | 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 |
| 270 | Ancient human genome sequence of an extinct Palaeo-Eskimo. *Nature*, **2010**, 463, 757-62 | 50.4 | 567 |
| 269 | 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 |
| 268 | JARID2 regulates binding of the Polycomb repressive complex 2 to target genes in ES cells. *Nature*, **2010**, 464, 306-10 | 50.4 | 427 |
| 267 | 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 |
| 266 | MicroRNA expression in the adult mouse central nervous system. *Rna*, **2008**, 14, 432-44 | 5.8 | 360 |
| 265 | 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 |
| 264 | 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 |
| 263 | 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 |
| 262 | The phenotypic spectrum of SCN8A encephalopathy. *Neurology*, **2015**, 84, 480-9 | 6.5 | 199 |
| 261 | 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 |
| 260 | 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 |

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|---|---|---|---|
| 259 | 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 |
| 258 | Obstetrical and gynecological complications in fragile X carriers: a multicenter study. *American Journal of Medical Genetics Part A*, **1994**, 51, 400-2 | | 178 |
| 257 | 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 |
| 256 | 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 |
| 255 | 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 |
| 254 | Jarid1b targets genes regulating development and is involved in neural differentiation. *EMBO Journal*, **2011**, 30, 4586-600 | 13 | 135 |
| 253 | 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 |
| 252 | Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. *Annals of Neurology*, **2016**, 79, 428-36 | 9.4 | 124 |
| 251 | 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 |
| 250 | 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 |
| 249 | 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 |
| 248 | REST-mediated recruitment of polycomb repressor complexes in mammalian cells. *PLoS Genetics*, **2012**, 8, e1002494 | 6 | 116 |
| 247 | 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 |
| 246 | Autoimmune diseases in women with Turner's syndrome. *Arthritis and Rheumatism*, **2010**, 62, 658-66 | | 111 |
| 245 | A balanced chromosomal translocation disrupting ARHGEF9 is associated with epilepsy, anxiety, aggression, and mental retardation. *Human Mutation*, **2009**, 30, 61-8 | 4.7 | 108 |
| 244 | Corpus callosum abnormalities, intellectual disability, speech impairment, and autism in patients with haploinsufficiency of ARID1B. *Clinical Genetics*, **2012**, 82, 248-55 | 4 | 104 |
| 243 | 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 |
| 242 | Mutations in autism susceptibility candidate 2 (AUTS2) in patients with mental retardation. *Human Genetics*, **2007**, 121, 501-9 | 6.3 | 97 |

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|---|---|---|---|
| 241 | 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 |
| 240 | Distinct CDH3 mutations cause ectodermal dysplasia, ectrodactyly, macular dystrophy (EEM syndrome). *Journal of Medical Genetics*, **2005**, 42, 292-8 | 5.8 | 86 |
| 239 | 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 |
| 238 | The genetic basis of the Pierre Robin Sequence. *Cleft Palate-Craniofacial Journal*, **2006**, 43, 155-9 | 1.9 | 82 |
| 237 | 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 |
| 236 | 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 |
| 235 | 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 |
| 234 | Comparative genomics beyond sequence-based alignments: RNA structures in the ENCODE regions. *Genome Research*, **2008**, 18, 242-51 | 9.7 | 76 |
| 233 | Haploinsufficiency of TAB2 causes congenital heart defects in humans. *American Journal of Human Genetics*, **2010**, 86, 839-49 | 11 | 75 |
| 232 | Pierre Robin sequence may be caused by dysregulation of SOX9 and KCNJ2. *Journal of Medical Genetics*, **2007**, 44, 381-6 | 5.8 | 75 |
| 231 | Dissecting spatio-temporal protein networks driving human heart development and related disorders. *Molecular Systems Biology*, **2010**, 6, 381 | 12.2 | 72 |
| 230 | 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 |
| 229 | 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 |
| 228 | Mutations in GABRB3: From febrile seizures to epileptic encephalopathies. *Neurology*, **2017**, 88, 483-492 | 6.5 | 68 |
| 227 | 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 |
| 226 | Genetic association studies in lumbar disc degeneration: a systematic review. *PLoS ONE*, **2012**, 7, e49995 | 3.7 | 67 |
| 225 | Gene Panel Testing in Epileptic Encephalopathies and Familial Epilepsies. *Molecular Syndromology*, **2016**, 7, 210-219 | 1.5 | 65 |
| 224 | Exclusion of SNRPN as a major determinant of Prader-Willi syndrome by a translocation breakpoint. *Nature Genetics*, **1996**, 12, 452-4 | 36.3 | 64 |

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| 223 | 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 |
| 222 | 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 |
| 221 | The small RNA content of human sperm reveals pseudogene-derived piRNAs complementary to protein-coding genes. *Rna*, **2015**, 21, 1085-95 | 5.8 | 59 |
| 220 | 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 |
| 219 | Genome-wide analysis of CDX2 binding in intestinal epithelial cells (Caco-2). *Journal of Biological Chemistry*, **2010**, 285, 25115-25 | 5.4 | 57 |
| 218 | 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 |
| 217 | FISHing with locked nucleic acids (LNA): evaluation of different LNA/DNA mixmers. *Molecular and Cellular Probes*, **2003**, 17, 165-9 | 3.3 | 55 |
| 216 | 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 |
| 215 | 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 |
| 214 | 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 |
| 213 | 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 |
| 212 | 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 |
| 211 | 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 |
| 210 | Marker X chromosome induction in fibroblasts by FUdR. *American Journal of Medical Genetics Part A* , **1981**, 9, 263-4 | | 51 |
| 209 | Molecular cytogenetic characterization of ring chromosome 15 in three unrelated patients **2004**, 130A, 340-4 | | 49 |
| 208 | Exon-disrupting deletions of NRXN1 in idiopathic generalized epilepsy. *Epilepsia*, **2013**, 54, 256-64 | 6.4 | 48 |
| 207 | An excess of chromosome 1 breakpoints in male infertility. *European Journal of Human Genetics*, **2004**, 12, 993-1000 | 5.3 | 48 |
| 206 | Profiling microRNAs in lung tissue from pigs infected with Actinobacillus pleuropneumoniae. *BMC Genomics*, **2012**, 13, 459 | 4.5 | 47 |

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| 205 | Isolated and syndromic forms of congenital anosmia. *Clinical Genetics*, **2012**, 81, 210-5 | 4 | 47 |
| 204 | LNA-modified oligonucleotides are highly efficient as FISH probes. *Cytogenetic and Genome Research*, **2004**, 107, 32-7 | 1.9 | 47 |
| 203 | The identification and functional annotation of RNA structures conserved in vertebrates. *Genome Research*, **2017**, 27, 1371-1383 | 9.7 | 46 |
| 202 | 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 |
| 201 | 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 |
| 200 | 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 |
| 199 | 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 |
| 198 | Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. *Human Genetics*, **2006**, 118, 578-90 | 6.3 | 44 |
| 197 | Mapping of the human PAWR (par-4) gene to chromosome 12q21. *Genomics*, **1998**, 53, 241-3 | 4.3 | 44 |
| 196 | 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 |
| 195 | Reduced ceramide synthase 2 activity causes progressive myoclonic epilepsy. *Annals of Clinical and Translational Neurology*, **2014**, 1, 88-98 | 5.3 | 42 |
| 194 | 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 |
| 193 | A neocentromere on human chromosome 3 without detectable alpha-satellite DNA forms morphologically normal kinetochores. *Chromosoma*, **1998**, 107, 359-65 | 2.8 | 42 |
| 192 | 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 |
| 191 | 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 |
| 190 | Germline Chromothripsis Driven by L1-Mediated Retrotransposition and Alu/Alu Homologous Recombination. *Human Mutation*, **2016**, 37, 385-95 | 4.7 | 40 |
| 189 | Breakpoints around the HOXD cluster result in various limb malformations. *Journal of Medical Genetics*, **2006**, 43, 111-8 | 5.8 | 39 |
| 188 | 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 |

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| 187 | CHEMOATTRACTION INTETRAHYMENA: ON THE ROLE OF CHEMOKINESIS. *Biological Bulletin*, **1986**, 170, 357-367 | 1.5 | 38 |
| 186 | Genetic heterogeneity in Pakistani microcephaly families. *Clinical Genetics*, **2013**, 83, 446-51 | 4 | 37 |
| 185 | Sequence assembly. *Computational Biology and Chemistry*, **2009**, 33, 121-36 | 3.6 | 37 |
| 184 | Mutational analysis of the human FATE gene in 144 infertile men. *Human Genetics*, **2003**, 113, 195-201 | 6.3 | 35 |
| 183 | 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 |
| 182 | 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 |
| 181 | Non-disjunction of chromosome 13. *Human Molecular Genetics*, **2007**, 16, 2004-10 | 5.6 | 32 |
| 180 | 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 |
| 179 | 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 |
| 178 | Sequence variants in the human cocaine and amphetamine-regulated transcript (CART) gene in subjects with early onset obesity. *Obesity*, **1999**, 7, 532-6 | | 31 |
| 177 | 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 |
| 176 | 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 |
| 175 | Tetrasomy 18p de novo: parental origin and different mechanisms of formation. *European Journal of Human Genetics*, **1996**, 4, 160-7 | 5.3 | 29 |
| 174 | Metaphase FISH on a chip: miniaturized microfluidic device for fluorescence in situ hybridization. *Sensors*, **2010**, 10, 9831-46 | 3.8 | 27 |
| 173 | Delineation of an interstitial 9q22 deletion in basal cell nevus syndrome **2005**, 132A, 324-8 | | 27 |
| 172 | Identification of human candidate genes for male infertility by digital differential display. *Molecular Human Reproduction*, **2001**, 7, 11-20 | 4.4 | 27 |
| 171 | Acute and persistent symptoms in non-hospitalized PCR-confirmed COVID-19 patients. *Scientific Reports*, **2021**, 11, 13153 | 4.9 | 27 |
| 170 | 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 |

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| 169 | The Hedgehog signaling pathway--implications for drug targets in cancer and neurodegenerative disorders. *Pharmacogenomics*, **2003**, 4, 411-29 | 2.6 | 25 |
| 168 | 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 |
| 167 | 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 |
| 166 | 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 |
| 165 | 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 |
| 164 | Structural genomic variation in childhood epilepsies with complex phenotypes. *European Journal of Human Genetics*, **2014**, 22, 896-901 | 5.3 | 24 |
| 163 | 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 |
| 162 | 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 |
| 161 | Compound heterozygous ASPM mutations in Pakistani MCPH families. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 926-30 | 2.5 | 24 |
| 160 | Identification of the BRD1 interaction network and its impact on mental disorder risk. *Genome Medicine*, **2016**, 8, 53 | 14.4 | 23 |
| 159 | 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 |
| 158 | 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 |
| 157 | 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 |
| 156 | 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 |
| 155 | HOXD13 polyalanine tract expansion in classical synpolydactyly type Vordingborg. *American Journal of Medical Genetics Part A*, **2002**, 110, 116-21 | | 22 |
| 154 | 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 |
| 153 | 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 |
| 152 | Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. *PLoS Genetics*, **2018**, 14, e1007780 | 6 | 22 |

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| 151 | Occurrence of cancer in a cohort of 183 persons with constitutional chromosome 7 abnormalities. *Cancer Genetics and Cytogenetics*, **1998**, 105, 39-42 | | 21 |
| 150 | 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 |
| 149 | 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 |
| 148 | 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 |
| 147 | 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 |
| 146 | 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 |
| 145 | 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 |
| 144 | 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 |
| 143 | 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 |
| 142 | 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 |
| 141 | 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 |
| 140 | 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 |
| 139 | Regional differences in expression of specific markers for human embryonic stem cells. *Reproductive BioMedicine Online*, **2007**, 15, 89-98 | 4 | 18 |
| 138 | Screening test for ataxia telangiectasia. *Lancet, The*, **1987**, 2, 1398-9 | 40 | 18 |
| 137 | The first mutation in CNGA2 in two brothers with anosmia. *Clinical Genetics*, **2015**, 88, 293-6 | 4 | 17 |
| 136 | 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 |
| 135 | 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 |
| 134 | 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 |

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| 133 | 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 |
| 132 | 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 |
| 131 | Neural correlates of taste perception in congenital olfactory impairment. *Neuropsychologia*, **2014**, 62, 297-305 | 3.2 | 16 |
| 130 | Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. *Human Molecular Genetics*, **2014**, 23, 6163-76 | 5.6 | 16 |
| 129 | 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 |
| 128 | Screening of 99 Danish patients with congenital heart disease for GATA4 mutations. *Genetic Testing and Molecular Biomarkers*, **2006**, 10, 277-80 | | 16 |
| 127 | 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 |
| 126 | Molecular cytogenetic detection of 9q34 breakpoints associated with nail patella syndrome. *European Journal of Human Genetics*, **1999**, 7, 68-76 | 5.3 | 16 |
| 125 | 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 |
| 124 | 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 |
| 123 | Optical mapping of single-molecule human DNA in disposable, mass-produced all-polymer devices. *Journal of Micromechanics and Microengineering*, **2015**, 25, 105002 | 2 | 15 |
| 122 | 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 |
| 121 | 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 |
| 120 | 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 |
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