Ann Nordgren

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

137 papers	3,868 citations	36 h-index	57 g-index
143 ext. papers	4,870 ext. citations	6.7 avg, IF	4.56 L-index

#	Paper	IF	Citations
137	Detection of germline mosaicism in fathers of children with intellectual disability syndromes caused by de novo variants.. *Molecular Genetics & Genomic Medicine*, **2022**, e1880	2.3	1
136	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders.. *Npj Genomic Medicine*, **2022**, 7, 11	6.2	1
135	Williams syndrome: reduced orienting to others' eyes in a hypersocial phenotype.. *Journal of Autism and Developmental Disorders*, **2022**, 1	4.6	0
134	Pupillary response in reward processing in adults with major depressive disorder in remission.. *Journal of the International Neuropsychological Society*, **2022**, 1-10	3.1	
133	Williams syndrome: on the role of intellectual abilities in anxiety. *Orphanet Journal of Rare Diseases*, **2021**, 16, 472	4.2	0
132	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. *Genome Medicine*, **2021**, 13, 40	14.4	22
131	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. *Genome Medicine*, **2021**, 13, 63	14.4	9
130	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. *Journal of Human Genetics*, **2021**, 66, 995-1008	4.3	6
129	Birth Characteristics Among Children Diagnosed with Neurofibromatosis Type 1 and Tuberous Sclerosis. *Journal of Pediatrics*, **2021**, 239, 200-205.e2	3.6	
128	Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for non-oncologic disorders. *American Journal of Medical Genetics, Part A*, **2021**, 185, 517-527	2.5	1
127	Clinical versus automated assessments of morphological variants in twins with and without neurodevelopmental disorders. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1177-1189	2.5	3
126	p53 controls genomic stability and temporal differentiation of human neural stem cells and affects neural organization in human brain organoids. *Cell Death and Disease*, **2020**, 11, 52	9.8	15
125	Pathogenic copy number variants are detected in a subset of patients with gastrointestinal malformations. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1084	2.3	2
124	Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders. *Orphanet Journal of Rare Diseases*, **2020**, 15, 103	4.2	14
123	Heterozygous variants in : Beyond congenital mirror movements. *Neurology: Genetics*, **2020**, 6, e526	3.8	0
122	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. *Nature Communications*, **2020**, 11, 4932	17.4	25
121	Variable neurodevelopmental and morphological phenotypes of carriers with 12q12 duplications. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1013	2.3	2

120	A call for global action for rare diseases in Africa. *Nature Genetics*, **2020**, 52, 21-26	36.3	11
119	A complex DICER1 syndrome phenotype associated with a germline pathogenic variant affecting the RNase IIIa domain of DICER1. *Journal of Medical Genetics*, **2020**,	5.8	2
118	Modeling SHH-driven medulloblastoma with patient iPS cell-derived neural stem cells. *Proceedings of the National Academy of Sciences of the United States of America*, **2020**, 117, 20127-20138	11.5	9
117	Overexpression of chromatin remodeling and tyrosine kinase genes in iAMP21-positive acute lymphoblastic leukemia. *Leukemia and Lymphoma*, **2020**, 61, 604-613	1.9	2
116	Biallelic mutations in WRAP53 result in dysfunctional telomeres, Cajal bodies and DNA repair, thereby causing Hoyeraal-Hreidarsson syndrome. *Cell Death and Disease*, **2020**, 11, 238	9.8	8
115	Mutations in COL1A1/A2 and CREB3L1 are associated with oligodontia in osteogenesis imperfecta. *Orphanet Journal of Rare Diseases*, **2020**, 15, 80	4.2	4
114	Maternal smoking during pregnancy and risk of phacomatoses: results from a Swedish register-based study. *Clinical Epidemiology*, **2019**, 11, 793-800	5.9	2
113	Disruptive variants of associate with autism and interfere with neuronal development and synaptic transmission. *Science Advances*, **2019**, 5, eaax2166	14.3	16
112	Diagnosis and management in Pitt-Hopkins syndrome: First international consensus statement. *Clinical Genetics*, **2019**, 95, 462-478	4	34
111	Early activating somatic PIK3CA mutations promote ectopic muscle development and upper limb overgrowth. *Clinical Genetics*, **2019**, 96, 118-125	4	9
110	Gain-of-function mutation of microRNA-140 in human skeletal dysplasia. *Nature Medicine*, **2019**, 25, 583-590	36.5	38
109	Microdeletion of 7p12.1p13, including IKZF1, causes intellectual impairment, overgrowth, and susceptibility to leukaemia. *British Journal of Haematology*, **2019**, 185, 354-357	4.5	3
108	Mowat-Wilson syndrome: Generation of two human iPS cell lines (UUIGPi004A and UUIGPi005A) from siblings with a truncating ZEB2 gene variant. *Stem Cell Research*, **2019**, 39, 101518	1.6	3
107	Ataxia in Patients With Bi-Allelic Mutations and Absence of Full-Length NF186. *Frontiers in Genetics*, **2019**, 10, 896	4.5	4
106	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. *Genome Medicine*, **2019**, 11, 68	14.4	44
105	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. *American Journal of Human Genetics*, **2019**, 104, 530-541	11	17
104	Predisposition to childhood acute lymphoblastic leukemia caused by a constitutional translocation disrupting. *Blood Advances*, **2019**, 3, 2722-2731	7.8	4
103	Rare copy number variants contribute pathogenic alleles in patients with intestinal malrotation. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e549	2.3	7

102	Somatic Structural Alterations in Childhood Leukemia Can Be Backtracked in Neonatal Dried Blood Spots by Use of Whole-Genome Sequencing and Digital PCR. *Clinical Chemistry*, **2019**, 65, 345-347	5.5	1
101	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. *Biological Psychiatry*, **2019**, 85, 287-297	7.9	55
100	The clinical utility of PGD with HLA matching: a collaborative multi-centre ESHRE study. *Human Reproduction*, **2018**, 33, 520-530	5.7	15
99	Copy Number Variation Analysis of 100 Twin Pairs Enriched for Neurodevelopmental Disorders. *Twin Research and Human Genetics*, **2018**, 21, 1-11	2.2	17
98	Alu-Alu mediated intragenic duplications in IFT81 and MATN3 are associated with skeletal dysplasias. *Human Mutation*, **2018**, 39, 1456-1467	4.7	9
97	Reversed gender ratio of autism spectrum disorder in Smith-Magenis syndrome. *Molecular Autism*, **2018**, 9, 1	6.5	40
96	High-resolution detection of chromosomal rearrangements in leukemias through mate pair whole genome sequencing. *PLoS ONE*, **2018**, 13, e0193928	3.7	9
95	Goltz syndrome in males: A clinical report of a male patient carrying a novel variant and a review of the literature. *Clinical Case Reports (discontinued)*, **2018**, 6, 2103-2110	0.7	7
94	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. *PLoS Genetics*, **2018**, 14, e1007780	6	22
93	A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation. *American Journal of Human Genetics*, **2018**, 103, 553-567	11	45
92	Genomic screening in rare disorders: New mutations and phenotypes, highlighting ALG14 as a novel cause of severe intellectual disability. *Clinical Genetics*, **2018**, 94, 528-537	4	14
91	Axial spondylometaphyseal dysplasia is also caused by NEK1 mutations. *Journal of Human Genetics*, **2017**, 62, 503-506	4.3	18
90	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. *Genetics in Medicine*, **2017**, 19, 900-908	8.1	30
89	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. *Nature Genetics*, **2017**, 49, 515-526	36.3	283
88	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. *Nature Neuroscience*, **2017**, 20, 1043-1051	25.5	94
87	Further evidence for specific IFIH1 mutation as a cause of Singleton-Merten syndrome with phenotypic heterogeneity. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1396-1399	2.5	21
86	Minor physical anomalies in neurodevelopmental disorders: a twin study. *Child and Adolescent Psychiatry and Mental Health*, **2017**, 11, 57	6.8	16
85	Intellectual Disability & Rare Disorders: A Diagnostic Challenge. *Advances in Experimental Medicine and Biology*, **2017**, 1031, 39-54	3.6	13

84	Whole-Genome Sequencing of Cytogenetically Balanced Chromosome Translocations Identifies Potentially Pathological Gene Disruptions and Highlights the Importance of Microhomology in the Mechanism of Formation. *Human Mutation*, **2017**, 38, 180-192	4.7	40
83	Phenotype and genotype in 52 patients with Rubinstein-Taybi syndrome caused by EP300 mutations. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3069-3082	2.5	60
82	A novel phenotype in N-glycosylation disorders: Gillessen-Kaesbach-Nishimura skeletal dysplasia due to pathogenic variants in ALG9. *European Journal of Human Genetics*, **2016**, 24, 198-207	5.3	19
81	Small 6q16.1 Deletions Encompassing POU3F2 Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability. *American Journal of Human Genetics*, **2016**, 98, 363-72	11	26
80	Increased risk of colorectal cancer in patients diagnosed with breast cancer in women. *Cancer Epidemiology*, **2016**, 41, 57-62	2.8	12
79	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. *American Journal of Human Genetics*, **2016**, 98, 373-81	11	55
78	PAX5-ESRRB is a recurrent fusion gene in B-cell precursor pediatric acute lymphoblastic leukemia. *Haematologica*, **2016**, 101, e20-3	6.6	7
77	Periodontal Ehlers-Danlos Syndrome Is Caused by Mutations in C1R and C1S, which Encode Subcomponents C1r and C1s of Complement. *American Journal of Human Genetics*, **2016**, 99, 1005-1014	11	70
76	Expanding the ataxia with oculomotor apraxia type 4 phenotype. *Neurology: Genetics*, **2016**, 2, e49	3.8	26
75	Detailed gene dose analysis reveals recurrent focal gene deletions in pediatric B-cell precursor acute lymphoblastic leukemia. *Leukemia and Lymphoma*, **2016**, 57, 2161-70	1.9	9
74	Mutations in FLVCR2 associated with Fowler syndrome and survival beyond infancy. *Clinical Genetics*, **2016**, 89, 99-103	4	12
73	Pathogenenic variant in the COL2A1 gene is associated with Spondyloepiphyseal dysplasia type Stanescu. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 266-9	2.5	2
72	CREBBP and EP300 mutational spectrum and clinical presentations in a cohort of Swedish patients with Rubinstein-Taybi syndrome. *Molecular Genetics & Genomic Medicine*, **2016**, 4, 39-45	2.3	17
71	DNA methylation-based subtype prediction for pediatric acute lymphoblastic leukemia. *Clinical Epigenetics*, **2015**, 7, 11	7.7	40
70	No evidence for mosaic pathogenic copy number variations in cardiac tissue from patients with congenital heart malformations. *European Journal of Medical Genetics*, **2015**, 58, 129-33	2.6	4
69	The genomic landscape of high hyperdiploid childhood acute lymphoblastic leukemia. *Nature Genetics*, **2015**, 47, 672-6	36.3	113
68	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. *American Journal of Human Genetics*, **2015**, 96, 784-96	11	35
67	Further delineation of the KBG syndrome phenotype caused by ANKRD11 aberrations. *European Journal of Human Genetics*, **2015**, 23, 1176-85	5.3	49

66	The clinical impact of IKZF1 deletions in paediatric B-cell precursor acute lymphoblastic leukaemia is independent of minimal residual disease stratification in Nordic Society for Paediatric Haematology and Oncology treatment protocols used between 1992 and 2013. *British Journal of Haematology*, **2015**, 170, 847-58	4.5	26
65	SLC26A2 disease spectrum in Sweden - high frequency of recessive multiple epiphyseal dysplasia (rMED). *Clinical Genetics*, **2015**, 87, 273-8	4	8
64	Autosomal recessive mutations in the COL2A1 gene cause severe spondyloepiphyseal dysplasia. *Clinical Genetics*, **2015**, 87, 496-8	4	7
63	Dominant mutations in KAT6A cause intellectual disability with recognizable syndromic features. *American Journal of Human Genetics*, **2015**, 96, 507-13	11	70
62	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. *Nature Genetics*, **2014**, 46, 380-4	36.3	197
61	Different mutations in PDE4D associated with developmental disorders with mirror phenotypes. *Journal of Medical Genetics*, **2014**, 51, 45-54	5.8	51
60	Neu-Laxova syndrome is a heterogeneous metabolic disorder caused by defects in enzymes of the L-serine biosynthesis pathway. *American Journal of Human Genetics*, **2014**, 95, 285-93	11	82
59	Clinical and genetic features of pediatric acute lymphoblastic leukemia in Down syndrome in the Nordic countries. *Journal of Hematology and Oncology*, **2014**, 7, 32	22.4	9
58	Autosomal dominant brachyolmia in a large Swedish family: phenotypic spectrum and natural course. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1635-41	2.5	6
57	Mutation screening and array comparative genomic hybridization using a 180K oligonucleotide array in VACTERL association. *PLoS ONE*, **2014**, 9, e85313	3.7	21
56	The Roots of Autism and ADHD Twin Study in Sweden (RATSS). *Twin Research and Human Genetics*, **2014**, 17, 164-76	2.2	46
55	Detecting dic(9;20)(p13.2;p11.2)-positive B-cell precursor acute lymphoblastic leukemia in a clinical setting using fluorescence in situ hybridization. *Leukemia*, **2014**, 28, 196-8	10.7	5
54	Small mosaic deletion encompassing the snoRNAs and SNURF-SNRPN results in an atypical Prader-Willi syndrome phenotype. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 425-31	2.5	20
53	The transcriptional regulator ADNP links the BAF (SWI/SNF) complexes with autism. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 315-26	3.1	53
52	Identification of three novel FGF16 mutations in X-linked recessive fusion of the fourth and fifth metacarpals and possible correlation with heart disease. *Molecular Genetics & Genomic Medicine*, **2014**, 2, 402-11	2.3	15
51	Novel Focal Gene Deletions in Pediatric B-Cell Precursor Acute Lymphoblastic Leukemia Detected By Array Comparative Genomic Hybridization. *Blood*, **2014**, 124, 1085-1085	2.2	
50	DNA Methylation-Based Subtype Prediction for Pediatric Acute Lymphoblastic Leukemia (ALL). *Blood*, **2014**, 124, 490-490	2.2	
49	The ETV6/RUNX1 fusion transcript is not detected in RNA isolated from neonatal dried blood spots from children later diagnosed with the corresponding leukemia. *Leukemia and Lymphoma*, **2013**, 54, 2742-4	1.9	3

48	Partial tetrasomy 14 associated with multiple malformations. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1284-90	2.5	4
47	Molecular and clinical delineation of the 17q22 microdeletion phenotype. *European Journal of Human Genetics*, **2013**, 21, 1085-92	5.3	8
46	High modal number and triple trisomies are highly correlated favorable factors in childhood B-cell precursor high hyperdiploid acute lymphoblastic leukemia treated according to the NOPHO ALL 1992/2000 protocols. *Haematologica*, **2013**, 98, 1424-32	6.6	36
45	The phenotype range of achondrogenesis 1A. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2554-8	2.5	4
44	A novel intellectual disability syndrome caused by GPI anchor deficiency due to homozygous mutations in PIGT. *Journal of Medical Genetics*, **2013**, 50, 521-8	5.8	92
43	Additional aberrations of the ETV6 and RUNX1 genes have no prognostic impact in 229 t(12;21)(p13;q22)-positive B-cell precursor acute lymphoblastic leukaemias treated according to the NOPHO-ALL-2000 protocol. *Leukemia Research*, **2012**, 36, 936-8	2.7	4
42	High frequency of BTG1 deletions in acute lymphoblastic leukemia in children with down syndrome. *Genes Chromosomes and Cancer*, **2012**, 51, 196-206	5	19
41	Homozygous deletions of CDKN2A are present in all dic(9;20)(p13□2;q11□2)-positive B-cell precursor acute lymphoblastic leukaemias and may be important for leukaemic transformation. *British Journal of Haematology*, **2012**, 159, 488-91	4.5	5
40	A novel 13 base pair insertion in the sonic hedgehog ZRS limb enhancer (ZRS/LMBR1) causes preaxial polydactyly with triphalangeal thumb. *Human Mutation*, **2012**, 33, 1063-6	4.7	26
39	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. *Nature*, **2011**, 478, 97-102	50.4	322
38	Interphase fluorescent in situ hybridization deletion analysis of the 9p21 region and prognosis in childhood acute lymphoblastic leukaemia (ALL): results from a prospective analysis of 519 Nordic patients treated according to the NOPHO-ALL 2000 protocol. *British Journal of Haematology*, **2011**, 152, 615-22	4.5	14
37	Paediatric B-cell precursor acute lymphoblastic leukaemia with t(1;19)(q23;p13): clinical and cytogenetic characteristics of 47 cases from the Nordic countries treated according to NOPHO protocols. *British Journal of Haematology*, **2011**, 155, 235-43	4.5	26
36	Copy number variation characteristics in subpopulations of patients with autism spectrum disorders. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2011**, 156, 115-24	3.5	56
35	Detailed molecular and clinical characterization of three patients with 21q deletions. *Clinical Genetics*, **2010**, 77, 145-54	4	47
34	Improved structural characterization of chromosomal breakpoints using high resolution custom array-CGH. *Clinical Genetics*, **2010**, 77, 552-62	4	10
33	Characterisation of hairy cell leukaemia by tiling resolution array-based comparative genome hybridisation: a series of 13 cases and review of the literature. *European Journal of Haematology*, **2010**, 84, 17-25	3.8	13
32	Molecular and clinical characterization of patients with overlapping 10p deletions. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1233-43	2.5	48
31	Chimerism resulting from parthenogenetic activation and dispermic fertilization. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2277-86	2.5	21

30	Germinal and Somatic Trisomy 21 Mosaicism: How Common is it, What are the Implications for Individual Carriers and How Does it Come About?. *Current Genomics*, **2010**, 11, 409-19	2.6	34
29	Jumping translocation in a phenotypically normal male: A study of mosaicism in spermatozoa, lymphocytes, and fibroblasts. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1706-11	2.5	7
28	Clinical and cytogenetic features of a population-based consecutive series of 285 pediatric T-cell acute lymphoblastic leukemias: rare T-cell receptor gene rearrangements are associated with poor outcome. *Genes Chromosomes and Cancer*, **2009**, 48, 795-805	5	31
27	Quantitation of RNA decay in dried blood spots during 20 years of storage. *Clinical Chemistry and Laboratory Medicine*, **2009**, 47, 1467-9	5.9	23
26	Expression of PTEN and SHP1, investigated from tissue microarrays in pediatric acute lymphoblastic, leukemia. *Pediatric Hematology and Oncology*, **2009**, 26, 48-56	1.7	14
25	Expression of VEGF and VEGF receptors in childhood precursor B-cell acute lymphoblastic leukemia evaluated by immunohistochemistry. *Journal of Pediatric Hematology/Oncology*, **2009**, 31, 696-701	1.2	9
24	Single nucleotide polymorphism genomic arrays analysis of t(8;21) acute myeloid leukemia cells. *Haematologica*, **2009**, 94, 1301-6	6.6	15
23	Relapsed Childhood High Hyperdiploid Acute Lymphoblastic Leukemia: Genome-Wide Screening Reveals the Presence of Preleukemic Ancestral Clones and the Secondary Nature of Microdeletions and RTK-RAS Mutations.. *Blood*, **2009**, 114, 2591-2591	2.2	
22	Array-CGH reveals hidden gene dose changes in children with acute lymphoblastic leukaemia and a normal or failed karyotype by G-banding. *British Journal of Haematology*, **2008**, 140, 572-7	4.5	20
21	Outcome of ETV6/RUNX1-positive childhood acute lymphoblastic leukaemia in the NOPHO-ALL-1992 protocol: frequent late relapses but good overall survival. *British Journal of Haematology*, **2008**, 140, 665-72	4.5	64
20	U-2973, a novel B-cell line established from a patient with a mature B-cell leukemia displaying concurrent t(14;18) and MYC translocation to a non-IG gene partner. *European Journal of Haematology*, **2008**, 81, 218-25	3.8	4
19	Characterization of 6q deletions in mature B cell lymphomas and childhood acute lymphoblastic leukemia. *Leukemia and Lymphoma*, **2008**, 49, 477-87	1.9	46
18	Clinical and cytogenetic features of pediatric dic(9;20)(p13.2;q11.2)-positive B-cell precursor acute lymphoblastic leukemias: a Nordic series of 24 cases and review of the literature. *Genes Chromosomes and Cancer*, **2008**, 47, 149-58	5	27
17	Cytogenetic patterns in ETV6/RUNX1-positive pediatric B-cell precursor acute lymphoblastic leukemia: A Nordic series of 245 cases and review of the literature. *Genes Chromosomes and Cancer*, **2007**, 46, 440-50	5	33
16	Comprehensive mutational analysis of a cohort of Swedish Cornelia de Lange syndrome patients. *European Journal of Human Genetics*, **2007**, 15, 143-9	5.3	36
15	Left-sided embryonic expression of the BCL-6 corepressor, BCOR, is required for vertebrate laterality determination. *Human Molecular Genetics*, **2007**, 16, 1773-82	5.6	42
14	PGD for dystrophin gene deletions using fluorescence in situ hybridization. *Molecular Human Reproduction*, **2006**, 12, 353-6	4.4	10
13	Characterisation of dic(9;20)(p11-13;q11) in childhood B-cell precursor acute lymphoblastic leukaemia by tiling resolution array-based comparative genomic hybridisation reveals clustered breakpoints at 9p13.2 and 20q11.2. *British Journal of Haematology*, **2006**, 135, 492-9	4.5	48

12	Genome-wide screening using array-CGH does not reveal microdeletions/microduplications in children with Kabuki syndrome. *European Journal of Human Genetics*, **2005**, 13, 260-3	5.3	20
11	Molecular cytogenetic characterization of an insertional translocation, ins(6;7)(p25;q33q34): deletion/duplication of 7q33-34 and clinical correlations. *American Journal of Medical Genetics, Part A*, **2005**, 139, 25-31	2.5	5
10	Hidden aberrations diagnosed by interphase fluorescence in situ hybridisation and spectral karyotyping in childhood acute lymphoblastic leukaemia. *Leukemia and Lymphoma*, **2003**, 44, 2039-53	1.9	17
9	Cytogenetic abnormalities in childhood acute myeloid leukaemia: a Nordic series comprising all children enrolled in the NOPHO-93-AML trial between 1993 and 2001. *British Journal of Haematology*, **2003**, 121, 566-77	4.5	54
8	Spectral karyotyping and interphase FISH reveal abnormalities not detected by conventional G-banding. Implications for treatment stratification of childhood acute lymphoblastic leukaemia: detailed analysis of 70 cases. *European Journal of Haematology*, **2002**, 68, 31-41	3.8	50
7	Single cell CGH analysis reveals a high degree of mosaicism in human embryos from patients with balanced structural chromosome aberrations. *Molecular Human Reproduction*, **2002**, 8, 502-10	4.4	49
6	Interphase fluorescence in situ hybridization and spectral karyotyping reveals hidden genetic aberrations in children with acute lymphoblastic leukaemia and a normal banded karyotype. *British Journal of Haematology*, **2001**, 114, 786-93	4.5	14
5	Identification of numerical and structural chromosome aberrations in 15 high hyperdiploid childhood acute lymphoblastic leukemias using spectral karyotyping. *European Journal of Haematology*, **2001**, 66, 297-304	3.8	13
4	Limitations of chromosome classification by multicolor karyotyping. *American Journal of Human Genetics*, **2001**, 68, 1043-7	11	66
3	Chromosomal alterations in 15 breast cancer cell lines by comparative genomic hybridization and spectral karyotyping. *Genes Chromosomes and Cancer*, **2000**, 28, 308-17	5	99
2	A case of acute lymphoblastic leukemia, near-triploidy, and poor outcome: characterization by fluorescence in situ hybridization using chromosome-specific libraries from all human chromosomes. *Cancer Genetics and Cytogenetics*, **1997**, 99, 93-6		6
1	Trisomy 5q12-->q13.3 in a patient with add(13q): characterization of an interchromosomal insertion by forward and reverse chromosome painting. *American Journal of Medical Genetics Part A*, **1997**, 73, 351-5		3