

# Ann Nordgren

## List of Publications by Citations

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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	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. <i>Nature</i> , <b>2011</b> , 478, 97-102	50.4	322
136	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. <i>Nature Genetics</i> , <b>2017</b> , 49, 515-526	36.3	283
135	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. <i>Nature Genetics</i> , <b>2014</b> , 46, 380-4	36.3	197
134	The genomic landscape of high hyperdiploid childhood acute lymphoblastic leukemia. <i>Nature Genetics</i> , <b>2015</b> , 47, 672-6	36.3	113
133	Chromosomal alterations in 15 breast cancer cell lines by comparative genomic hybridization and spectral karyotyping. <i>Genes Chromosomes and Cancer</i> , <b>2000</b> , 28, 308-17	5	99
132	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. <i>Nature Neuroscience</i> , <b>2017</b> , 20, 1043-1051	25.5	94
131	A novel intellectual disability syndrome caused by GPI anchor deficiency due to homozygous mutations in PIGT. <i>Journal of Medical Genetics</i> , <b>2013</b> , 50, 521-8	5.8	92
130	Neu-Laxova syndrome is a heterogeneous metabolic disorder caused by defects in enzymes of the L-serine biosynthesis pathway. <i>American Journal of Human Genetics</i> , <b>2014</b> , 95, 285-93	11	82
129	Dominant mutations in KAT6A cause intellectual disability with recognizable syndromic features. <i>American Journal of Human Genetics</i> , <b>2015</b> , 96, 507-13	11	70
128	Periodontal Ehlers-Danlos Syndrome Is Caused by Mutations in C1R and C1S, which Encode Subcomponents C1r and C1s of Complement. <i>American Journal of Human Genetics</i> , <b>2016</b> , 99, 1005-1014	11	70
127	Limitations of chromosome classification by multicolor karyotyping. <i>American Journal of Human Genetics</i> , <b>2001</b> , 68, 1043-7	11	66
126	Outcome of ETV6/RUNX1-positive childhood acute lymphoblastic leukaemia in the NOPHO-ALL-1992 protocol: frequent late relapses but good overall survival. <i>British Journal of Haematology</i> , <b>2008</b> , 140, 665-72	4.5	64
125	Phenotype and genotype in 52 patients with Rubinstein-Taybi syndrome caused by EP300 mutations. <i>American Journal of Medical Genetics, Part A</i> , <b>2016</b> , 170, 3069-3082	2.5	60
124	Copy number variation characteristics in subpopulations of patients with autism spectrum disorders. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , <b>2011</b> , 156, 115-24	3.5	56
123	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. <i>American Journal of Human Genetics</i> , <b>2016</b> , 98, 373-81	11	55
122	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. <i>Biological Psychiatry</i> , <b>2019</b> , 85, 287-297	7.9	55
121	Cytogenetic abnormalities in childhood acute myeloid leukaemia: a Nordic series comprising all children enrolled in the NOPHO-93-AML trial between 1993 and 2001. <i>British Journal of Haematology</i> , <b>2003</b> , 121, 566-77	4.5	54

120	The transcriptional regulator ADNP links the BAF (SWI/SNF) complexes with autism. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2014</b> , 166C, 315-26	3.1	53
119	Different mutations in PDE4D associated with developmental disorders with mirror phenotypes. <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 45-54	5.8	51
118	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. <i>European Journal of Haematology</i> , <b>2002</b> , 68, 31-41	3.8	50
117	Further delineation of the KBG syndrome phenotype caused by ANKRD11 aberrations. <i>European Journal of Human Genetics</i> , <b>2015</b> , 23, 1176-85	5.3	49
116	Single cell CGH analysis reveals a high degree of mosaicism in human embryos from patients with balanced structural chromosome aberrations. <i>Molecular Human Reproduction</i> , <b>2002</b> , 8, 502-10	4.4	49
115	Molecular and clinical characterization of patients with overlapping 10p deletions. <i>American Journal of Medical Genetics, Part A</i> , <b>2010</b> , 152A, 1233-43	2.5	48
114	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. <i>British Journal of Haematology</i> , <b>2006</b> , 135, 492-9	4.5	48
113	Detailed molecular and clinical characterization of three patients with 21q deletions. <i>Clinical Genetics</i> , <b>2010</b> , 77, 145-54	4	47
112	The Roots of Autism and ADHD Twin Study in Sweden (RATSS). <i>Twin Research and Human Genetics</i> , <b>2014</b> , 17, 164-76	2.2	46
111	Characterization of 6q deletions in mature B cell lymphomas and childhood acute lymphoblastic leukemia. <i>Leukemia and Lymphoma</i> , <b>2008</b> , 49, 477-87	1.9	46
110	A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation. <i>American Journal of Human Genetics</i> , <b>2018</b> , 103, 553-567	11	45
109	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. <i>Genome Medicine</i> , <b>2019</b> , 11, 68	14.4	44
108	Left-sided embryonic expression of the BCL-6 corepressor, BCOR, is required for vertebrate laterality determination. <i>Human Molecular Genetics</i> , <b>2007</b> , 16, 1773-82	5.6	42
107	DNA methylation-based subtype prediction for pediatric acute lymphoblastic leukemia. <i>Clinical Epigenetics</i> , <b>2015</b> , 7, 11	7.7	40
106	Reversed gender ratio of autism spectrum disorder in Smith-Magenis syndrome. <i>Molecular Autism</i> , <b>2018</b> , 9, 1	6.5	40
105	Whole-Genome Sequencing of Cytogenetically Balanced Chromosome Translocations Identifies Potentially Pathological Gene Disruptions and Highlights the Importance of Microhomology in the Mechanism of Formation. <i>Human Mutation</i> , <b>2017</b> , 38, 180-192	4.7	40
104	Gain-of-function mutation of microRNA-140 in human skeletal dysplasia. <i>Nature Medicine</i> , <b>2019</b> , 25, 583-590	5.9	38
103	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. <i>Haematologica</i> , <b>2013</b> , 98, 1424-32	6.6	36

102	Comprehensive mutational analysis of a cohort of Swedish Cornelia de Lange syndrome patients. <i>European Journal of Human Genetics</i> , <b>2007</b> , 15, 143-9	5.3	36
101	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. <i>American Journal of Human Genetics</i> , <b>2015</b> , 96, 784-96	11	35
100	Diagnosis and management in Pitt-Hopkins syndrome: First international consensus statement. <i>Clinical Genetics</i> , <b>2019</b> , 95, 462-478	4	34
99	Germinal and Somatic Trisomy 21 Mosaicism: How Common is it, What are the Implications for Individual Carriers and How Does it Come About?. <i>Current Genomics</i> , <b>2010</b> , 11, 409-19	2.6	34
98	Cytogenetic patterns in ETV6/RUNX1-positive pediatric B-cell precursor acute lymphoblastic leukemia: A Nordic series of 245 cases and review of the literature. <i>Genes Chromosomes and Cancer</i> , <b>2007</b> , 46, 440-50	5	33
97	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. <i>Genes Chromosomes and Cancer</i> , <b>2009</b> , 48, 795-805	5	31
96	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 900-908	8.1	30
95	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. <i>Genes Chromosomes and Cancer</i> , <b>2008</b> , 47, 149-58	5	27
94	Small 6q16.1 Deletions Encompassing POU3F2 Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability. <i>American Journal of Human Genetics</i> , <b>2016</b> , 98, 363-72	11	26
93	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. <i>British Journal of Haematology</i> , <b>2015</b> , 170, 847-58	4.5	26
92	A novel 13 base pair insertion in the sonic hedgehog ZRS limb enhancer (ZRS/LMBR1) causes preaxial polydactyly with triphalangeal thumb. <i>Human Mutation</i> , <b>2012</b> , 33, 1063-6	4.7	26
91	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. <i>British Journal of Haematology</i> , <b>2011</b> , 155, 235-43	4.5	26
90	Expanding the ataxia with oculomotor apraxia type 4 phenotype. <i>Neurology: Genetics</i> , <b>2016</b> , 2, e49	3.8	26
89	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. <i>Nature Communications</i> , <b>2020</b> , 11, 4932	17.4	25
88	Quantitation of RNA decay in dried blood spots during 20 years of storage. <i>Clinical Chemistry and Laboratory Medicine</i> , <b>2009</b> , 47, 1467-9	5.9	23
87	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. <i>Genome Medicine</i> , <b>2021</b> , 13, 40	14.4	22
86	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. <i>PLoS Genetics</i> , <b>2018</b> , 14, e1007780	6	22
85	Further evidence for specific IFIH1 mutation as a cause of Singleton-Merten syndrome with phenotypic heterogeneity. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 1396-1399	2.5	21

84	Mutation screening and array comparative genomic hybridization using a 180K oligonucleotide array in VACTERL association. <i>PLoS ONE</i> , <b>2014</b> , 9, e85313	3.7	21
83	Chimerism resulting from parthenogenetic activation and dispermic fertilization. <i>American Journal of Medical Genetics, Part A</i> , <b>2010</b> , 152A, 2277-86	2.5	21
82	Small mosaic deletion encompassing the snoRNAs and SNURF-SNRPN results in an atypical Prader-Willi syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , <b>2014</b> , 164A, 425-31	2.5	20
81	Array-CGH reveals hidden gene dose changes in children with acute lymphoblastic leukaemia and a normal or failed karyotype by G-banding. <i>British Journal of Haematology</i> , <b>2008</b> , 140, 572-7	4.5	20
80	Genome-wide screening using array-CGH does not reveal microdeletions/microduplications in children with Kabuki syndrome. <i>European Journal of Human Genetics</i> , <b>2005</b> , 13, 260-3	5.3	20
79	A novel phenotype in N-glycosylation disorders: Gillesen-Kaesbach-Nishimura skeletal dysplasia due to pathogenic variants in ALG9. <i>European Journal of Human Genetics</i> , <b>2016</b> , 24, 198-207	5.3	19
78	High frequency of BTG1 deletions in acute lymphoblastic leukemia in children with down syndrome. <i>Genes Chromosomes and Cancer</i> , <b>2012</b> , 51, 196-206	5	19
77	Axial spondylometaphyseal dysplasia is also caused by NEK1 mutations. <i>Journal of Human Genetics</i> , <b>2017</b> , 62, 503-506	4.3	18
76	Copy Number Variation Analysis of 100 Twin Pairs Enriched for Neurodevelopmental Disorders. <i>Twin Research and Human Genetics</i> , <b>2018</b> , 21, 1-11	2.2	17
75	Hidden aberrations diagnosed by interphase fluorescence in situ hybridisation and spectral karyotyping in childhood acute lymphoblastic leukaemia. <i>Leukemia and Lymphoma</i> , <b>2003</b> , 44, 2039-53	1.9	17
74	CREBBP and EP300 mutational spectrum and clinical presentations in a cohort of Swedish patients with Rubinstein-Taybi syndrome. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2016</b> , 4, 39-45	2.3	17
73	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 530-541	11	17
72	Disruptive variants of associate with autism and interfere with neuronal development and synaptic transmission. <i>Science Advances</i> , <b>2019</b> , 5, eaax2166	14.3	16
71	Minor physical anomalies in neurodevelopmental disorders: a twin study. <i>Child and Adolescent Psychiatry and Mental Health</i> , <b>2017</b> , 11, 57	6.8	16
70	p53 controls genomic stability and temporal differentiation of human neural stem cells and affects neural organization in human brain organoids. <i>Cell Death and Disease</i> , <b>2020</b> , 11, 52	9.8	15
69	The clinical utility of PGD with HLA matching: a collaborative multi-centre ESHRE study. <i>Human Reproduction</i> , <b>2018</b> , 33, 520-530	5.7	15
68	Identification of three novel FGF16 mutations in X-linked recessive fusion of the fourth and fifth metacarpals and possible correlation with heart disease. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2014</b> , 2, 402-11	2.3	15
67	Single nucleotide polymorphism genomic arrays analysis of t(8;21) acute myeloid leukemia cells. <i>Haematologica</i> , <b>2009</b> , 94, 1301-6	6.6	15

66	Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders. <i>Orphanet Journal of Rare Diseases</i> , <b>2020</b> , 15, 103	4.2	14
65	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. <i>British Journal of Haematology</i> , <b>2011</b> , 132, 615-22	4.5	14
64	Expression of PTEN and SHP1, investigated from tissue microarrays in pediatric acute lymphoblastic, leukemia. <i>Pediatric Hematology and Oncology</i> , <b>2009</b> , 26, 48-56	1.7	14
63	Interphase fluorescence in situ hybridization and spectral karyotyping reveals hidden genetic aberrations in children with acute lymphoblastic leukaemia and a normal banded karyotype. <i>British Journal of Haematology</i> , <b>2001</b> , 114, 786-93	4.5	14
62	Genomic screening in rare disorders: New mutations and phenotypes, highlighting ALG14 as a novel cause of severe intellectual disability. <i>Clinical Genetics</i> , <b>2018</b> , 94, 528-537	4	14
61	Intellectual Disability & Rare Disorders: A Diagnostic Challenge. <i>Advances in Experimental Medicine and Biology</i> , <b>2017</b> , 1031, 39-54	3.6	13
60	Characterisation of hairy cell leukaemia by tiling resolution array-based comparative genome hybridisation: a series of 13 cases and review of the literature. <i>European Journal of Haematology</i> , <b>2010</b> , 84, 17-25	3.8	13
59	Identification of numerical and structural chromosome aberrations in 15 high hyperdiploid childhood acute lymphoblastic leukemias using spectral karyotyping. <i>European Journal of Haematology</i> , <b>2001</b> , 66, 297-304	3.8	13
58	Increased risk of colorectal cancer in patients diagnosed with breast cancer in women. <i>Cancer Epidemiology</i> , <b>2016</b> , 41, 57-62	2.8	12
57	Mutations in FLVCR2 associated with Fowler syndrome and survival beyond infancy. <i>Clinical Genetics</i> , <b>2016</b> , 89, 99-103	4	12
56	A call for global action for rare diseases in Africa. <i>Nature Genetics</i> , <b>2020</b> , 52, 21-26	36.3	11
55	Improved structural characterization of chromosomal breakpoints using high resolution custom array-CGH. <i>Clinical Genetics</i> , <b>2010</b> , 77, 552-62	4	10
54	PGD for dystrophin gene deletions using fluorescence in situ hybridization. <i>Molecular Human Reproduction</i> , <b>2006</b> , 12, 353-6	4.4	10
53	Early activating somatic PIK3CA mutations promote ectopic muscle development and upper limb overgrowth. <i>Clinical Genetics</i> , <b>2019</b> , 96, 118-125	4	9
52	Alu-Alu mediated intragenic duplications in IFT81 and MATN3 are associated with skeletal dysplasias. <i>Human Mutation</i> , <b>2018</b> , 39, 1456-1467	4.7	9
51	Clinical and genetic features of pediatric acute lymphoblastic leukemia in Down syndrome in the Nordic countries. <i>Journal of Hematology and Oncology</i> , <b>2014</b> , 7, 32	22.4	9
50	Expression of VEGF and VEGF receptors in childhood precursor B-cell acute lymphoblastic leukemia evaluated by immunohistochemistry. <i>Journal of Pediatric Hematology/Oncology</i> , <b>2009</b> , 31, 696-701	1.2	9
49	High-resolution detection of chromosomal rearrangements in leukemias through mate pair whole genome sequencing. <i>PLoS ONE</i> , <b>2018</b> , 13, e0193928	3.7	9

48	Modeling SHH-driven medulloblastoma with patient iPS cell-derived neural stem cells. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2020</b> , 117, 20127-20138	11.5	9
47	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. <i>Genome Medicine</i> , <b>2021</b> , 13, 63	14.4	9
46	Detailed gene dose analysis reveals recurrent focal gene deletions in pediatric B-cell precursor acute lymphoblastic leukemia. <i>Leukemia and Lymphoma</i> , <b>2016</b> , 57, 2161-70	1.9	9
45	SLC26A2 disease spectrum in Sweden - high frequency of recessive multiple epiphyseal dysplasia (rMED). <i>Clinical Genetics</i> , <b>2015</b> , 87, 273-8	4	8
44	Molecular and clinical delineation of the 17q22 microdeletion phenotype. <i>European Journal of Human Genetics</i> , <b>2013</b> , 21, 1085-92	5.3	8
43	Biallelic mutations in WRAP53 result in dysfunctional telomeres, Cajal bodies and DNA repair, thereby causing Hoyeraal-Hreidarsson syndrome. <i>Cell Death and Disease</i> , <b>2020</b> , 11, 238	9.8	8
42	Autosomal recessive mutations in the COL2A1 gene cause severe spondyloepiphyseal dysplasia. <i>Clinical Genetics</i> , <b>2015</b> , 87, 496-8	4	7
41	Jumping translocation in a phenotypically normal male: A study of mosaicism in spermatozoa, lymphocytes, and fibroblasts. <i>American Journal of Medical Genetics, Part A</i> , <b>2009</b> , 149A, 1706-11	2.5	7
40	PAX5-ESRRB is a recurrent fusion gene in B-cell precursor pediatric acute lymphoblastic leukemia. <i>Haematologica</i> , <b>2016</b> , 101, e20-3	6.6	7
39	Rare copy number variants contribute pathogenic alleles in patients with intestinal malrotation. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2019</b> , 7, e549	2.3	7
38	Goltz syndrome in males: A clinical report of a male patient carrying a novel variant and a review of the literature. <i>Clinical Case Reports (discontinued)</i> , <b>2018</b> , 6, 2103-2110	0.7	7
37	Autosomal dominant brachyolmia in a large Swedish family: phenotypic spectrum and natural course. <i>American Journal of Medical Genetics, Part A</i> , <b>2014</b> , 164A, 1635-41	2.5	6
36	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. <i>Cancer Genetics and Cytogenetics</i> , <b>1997</b> , 99, 93-6		6
35	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. <i>Journal of Human Genetics</i> , <b>2021</b> , 66, 995-1008	4.3	6
34	Detecting dic(9;20)(p13.2;p11.2)-positive B-cell precursor acute lymphoblastic leukemia in a clinical setting using fluorescence in situ hybridization. <i>Leukemia</i> , <b>2014</b> , 28, 196-8	10.7	5
33	Homozygous deletions of CDKN2A are present in all dic(9;20)(p13;q11)-positive B-cell precursor acute lymphoblastic leukaemias and may be important for leukaemic transformation. <i>British Journal of Haematology</i> , <b>2012</b> , 159, 488-91	4.5	5
32	Molecular cytogenetic characterization of an insertional translocation, ins(6;7)(p25;q33q34): deletion/duplication of 7q33-34 and clinical correlations. <i>American Journal of Medical Genetics, Part A</i> , <b>2005</b> , 139, 25-31	2.5	5
31	No evidence for mosaic pathogenic copy number variations in cardiac tissue from patients with congenital heart malformations. <i>European Journal of Medical Genetics</i> , <b>2015</b> , 58, 129-33	2.6	4

30	Ataxia in Patients With Bi-Allelic Mutations and Absence of Full-Length NF186. <i>Frontiers in Genetics</i> , <b>2019</b> , 10, 896	4.5	4
29	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. <i>Leukemia Research</i> , <b>2012</b> , 36, 936-8	2.7	4
28	Partial tetrasomy 14 associated with multiple malformations. <i>American Journal of Medical Genetics, Part A</i> , <b>2013</b> , 161A, 1284-90	2.5	4
27	The phenotype range of achondrogenesis 1A. <i>American Journal of Medical Genetics, Part A</i> , <b>2013</b> , 161A, 2554-8	2.5	4
26	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. <i>European Journal of Haematology</i> , <b>2008</b> , 81, 218-25	3.8	4
25	Predisposition to childhood acute lymphoblastic leukemia caused by a constitutional translocation disrupting. <i>Blood Advances</i> , <b>2019</b> , 3, 2722-2731	7.8	4
24	Mutations in COL1A1/A2 and CREB3L1 are associated with oligodontia in osteogenesis imperfecta. <i>Orphanet Journal of Rare Diseases</i> , <b>2020</b> , 15, 80	4.2	4
23	Clinical versus automated assessments of morphological variants in twins with and without neurodevelopmental disorders. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 1177-1189	2.5	3
22	Microdeletion of 7p12.1p13, including IKZF1, causes intellectual impairment, overgrowth, and susceptibility to leukaemia. <i>British Journal of Haematology</i> , <b>2019</b> , 185, 354-357	4.5	3
21	Mowat-Wilson syndrome: Generation of two human iPS cell lines (UIGPi004A and UIGPi005A) from siblings with a truncating ZEB2 gene variant. <i>Stem Cell Research</i> , <b>2019</b> , 39, 101518	1.6	3
20	The ETV6/RUNX1 fusion transcript is not detected in RNA isolated from neonatal dried blood spots from children later diagnosed with the corresponding leukemia. <i>Leukemia and Lymphoma</i> , <b>2013</b> , 54, 2742-9	1.9	3
19	Trisomy 5q12-->q13.3 in a patient with add(13q): characterization of an interchromosomal insertion by forward and reverse chromosome painting. <i>American Journal of Medical Genetics Part A</i> , <b>1997</b> , 73, 351-5		3
18	Maternal smoking during pregnancy and risk of phacomatoses: results from a Swedish register-based study. <i>Clinical Epidemiology</i> , <b>2019</b> , 11, 793-800	5.9	2
17	Pathogenic copy number variants are detected in a subset of patients with gastrointestinal malformations. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2020</b> , 8, e1084	2.3	2
16	Variable neurodevelopmental and morphological phenotypes of carriers with 12q12 duplications. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2020</b> , 8, e1013	2.3	2
15	A complex DICER1 syndrome phenotype associated with a germline pathogenic variant affecting the RNase IIIa domain of DICER1. <i>Journal of Medical Genetics</i> , <b>2020</b> ,	5.8	2
14	Pathogenenic variant in the COL2A1 gene is associated with Spondyloepiphyseal dysplasia type Stanescu. <i>American Journal of Medical Genetics, Part A</i> , <b>2016</b> , 170A, 266-9	2.5	2
13	Overexpression of chromatin remodeling and tyrosine kinase genes in iAMP21-positive acute lymphoblastic leukemia. <i>Leukemia and Lymphoma</i> , <b>2020</b> , 61, 604-613	1.9	2



12	Detection of germline mosaicism in fathers of children with intellectual disability syndromes caused by de novo variants.. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2022</b> , e1880	2.3	1
11	Somatic Structural Alterations in Childhood Leukemia Can Be Backtracked in Neonatal Dried Blood Spots by Use of Whole-Genome Sequencing and Digital PCR. <i>Clinical Chemistry</i> , <b>2019</b> , 65, 345-347	5.5	1
10	Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for non-oncologic disorders. <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 517-527	2.5	1
9	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders.. <i>Npj Genomic Medicine</i> , <b>2022</b> , 7, 11	6.2	1
8	Williams syndrome: on the role of intellectual abilities in anxiety. <i>Orphanet Journal of Rare Diseases</i> , <b>2021</b> , 16, 472	4.2	0
7	Heterozygous variants in : Beyond congenital mirror movements. <i>Neurology: Genetics</i> , <b>2020</b> , 6, e526	3.8	0
6	Williams syndrome: reduced orienting to other's eyes in a hypersocial phenotype.. <i>Journal of Autism and Developmental Disorders</i> , <b>2022</b> , 1	4.6	0
5	Novel Focal Gene Deletions in Pediatric B-Cell Precursor Acute Lymphoblastic Leukemia Detected By Array Comparative Genomic Hybridization. <i>Blood</i> , <b>2014</b> , 124, 1085-1085	2.2	
4	DNA Methylation-Based Subtype Prediction for Pediatric Acute Lymphoblastic Leukemia (ALL). <i>Blood</i> , <b>2014</b> , 124, 490-490	2.2	
3	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.. <i>Blood</i> , <b>2009</b> , 114, 2591-2591	2.2	
2	Birth Characteristics Among Children Diagnosed with Neurofibromatosis Type 1 and Tuberous Sclerosis. <i>Journal of Pediatrics</i> , <b>2021</b> , 239, 200-205.e2	3.6	
1	Pupillary response in reward processing in adults with major depressive disorder in remission.. <i>Journal of the International Neuropsychological Society</i> , <b>2022</b> , 1-10	3.1	