Asbjï¿½rg Stray-Pedersen

List of Publications by Year
in descending order

59	4,743	168829 31	169272 56
papers	citations	h-index	g-index

60	60	60	9939
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Recommendations for uniform definitions used in newborn screening for severe combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2022, 149, 1428-1436.	1.5	19
2	A Nationwide Study of GATA2 Deficiency in Norwayâ€"the Majority of Patients Have Undergone Allo-HSCT. Journal of Clinical Immunology, 2022, 42, 404-420.	2.0	10
3	Delayed Radiation Myelopathy in a Child With Hodgkin Lymphoma and ARTEMIS Mutation. Journal of Pediatric Hematology/Oncology, 2021, 43, e404-e407.	0.3	3
4	Performance of Expanded Newborn Screening in Norway Supported by Post-Analytical Bioinformatics Tools and Rapid Second-Tier DNA Analyses. International Journal of Neonatal Screening, 2020, 6, 51.	1.2	30
5	Second-Tier Next Generation Sequencing Integrated in Nationwide Newborn Screening Provides Rapid Molecular Diagnostics of Severe Combined Immunodeficiency. Frontiers in Immunology, 2020, 11, 1417.	2.2	38
6	Disease-associated CTNNBL1 mutation impairs somatic hypermutation by decreasing nuclear AID. Journal of Clinical Investigation, 2020, 130, 4411-4422.	3.9	11
7	Human NK cell deficiency as a result of biallelic mutations in MCM10. Journal of Clinical Investigation, 2020, 130, 5272-5286.	3.9	44
8	Mutations in PIGU Impair the Function of the GPI Transamidase Complex, Causing Severe Intellectual Disability, Epilepsy, and Brain Anomalies. American Journal of Human Genetics, 2019, 105, 395-402.	2.6	39
9	Infliximab therapy for inflammatory colitis in an infant with NEMO deficiency. Immunologic Research, 2019, 67, 450-453.	1.3	5
10	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. European Journal of Human Genetics, 2019, 27, 738-746.	1.4	32
11	Allogeneic hematopoietic stem cell transplant outcomes for patients with dominant negative IKZF1/IKAROS mutations. Journal of Allergy and Clinical Immunology, 2019, 144, 339-342.	1.5	28
12	A combined immunodeficiency with severe infections, inflammation, and allergy caused by ARPC1B deficiency. Journal of Allergy and Clinical Immunology, 2019, 143, 2296-2299.	1.5	87
13	Nordic Guidelines for Germline Predisposition to Myeloid Neoplasms in Adults: Recommendations for Genetic Diagnosis, Clinical Management and Follow-up. HemaSphere, 2019, 3, e321.	1.2	51
14	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	1.1	52
15	Genomic Characterization of a Pediatric Cohort with Non-Malignant Lymphoproliferative Disorders. Blood, 2019, 134, 83-83.	0.6	0
16	Genetic and mechanistic diversity in pediatric hemophagocytic lymphohistiocytosis. Blood, 2018, 132, 89-100.	0.6	139
17	Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339.	1.4	52
18	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	2.6	59

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19	Dystonia-deafness syndrome caused by ACTB p.Arg183Trp heterozygosity shows striatal dopaminergic dysfunction and response to pallidal stimulation. Journal of Neurodevelopmental Disorders, 2018, 10, 17.	1.5	22
20	Runaway Train: A Leaky Radiosensitive SCID with Skin Lesions and Multiple Lymphomas. Case Reports in Immunology, 2018, 2018, 1-6.	0.2	6
21	A novel NAA10 variant with impaired acetyltransferase activity causes developmental delay, intellectual disability, and hypertrophic cardiomyopathy. European Journal of Human Genetics, 2018, 26, 1294-1305.	1.4	28
22	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. Nucleic Acids Research, 2017, 45, gkw1237.	6.5	98
23	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	2.6	125
24	Chromatin-remodeling factor SMARCD2 regulates transcriptional networks controlling differentiation of neutrophil granulocytes. Nature Genetics, 2017, 49, 742-752.	9.4	87
25	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	3.6	184
26	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245.	1.5	261
27	Novel Combined Immune Deficiency and Radiation Sensitivity Blended Phenotype in an Adult with Biallelic Variations in ZAP70 and RNF168. Frontiers in Immunology, 2017, 8, 576.	2.2	23
28	First Case of CD40LG Deficiency in Ecuador, Diagnosed after Whole Exome Sequencing in a Patient with Severe Cutaneous Histoplasmosis. Frontiers in Pediatrics, 2017, 5, 17.	0.9	13
29	Novel PIGT Variant in Two Brothers: Expansion of the Multiple Congenital Anomalies-Hypotonia Seizures Syndrome 3 Phenotype. Genes, 2016, 7, 108.	1.0	25
30	A potential founder variant inCARMIL2/RLTPRin three Norwegian families with warts, molluscum contagiosum, and T-cell dysfunction. Molecular Genetics & Genomic Medicine, 2016, 4, 604-616.	0.6	59
31	Rapid molecular diagnostics of severe primary immunodeficiency determined by using targeted next-generation sequencing. Journal of Allergy and Clinical Immunology, 2016, 138, 1142-1151.e2.	1.5	85
32	Two male sibs with severe micrognathia and a missense variant in MED12. European Journal of Medical Genetics, 2016, 59, 367-372.	0.7	11
33	Loss of B Cells in Patients with Heterozygous Mutations in IKAROS. New England Journal of Medicine, 2016, 374, 1032-1043.	13.9	217
34	cnvScan: a CNV screening and annotation tool to improve the clinical utility of computational CNV prediction from exome sequencing data. BMC Genomics, 2016, 17, 51.	1.2	24
35	Biallelic Mutations in UNC80 Cause Persistent Hypotonia, Encephalopathy, Growth Retardation, and Severe Intellectual Disability. American Journal of Human Genetics, 2016, 98, 202-209.	2.6	45
36	The Extended Clinical Phenotype of 26 Patients with Chronic Mucocutaneous Candidiasis due to Gain-of-Function Mutations in STAT1. Journal of Clinical Immunology, 2016, 36, 73-84.	2.0	124

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37	Biallelic mutations in IRF8 impair human NK cell maturation and function. Journal of Clinical Investigation, 2016, 127, 306-320.	3.9	76
38	Norsk laboratoriekodeverk â€“ hvor ble det av visjonene?. Tidsskrift for Den Norske Laegeforening, 2016, 136, 1370-1372.	0.2	0
39	Early-onset lymphoproliferation and autoimmunity caused by germline STAT3 gain-of-function mutations. Blood, 2015, 125, 591-599.	0.6	436
40	Syndromic X-linked intellectual disability segregating with a missense variant in RLIM. European Journal of Human Genetics, 2015, 23, 1652-1656.	1.4	30
41	COPA mutations impair ER-Golgi transport and cause hereditary autoimmune-mediated lung disease and arthritis. Nature Genetics, 2015, 47, 654-660.	9.4	302
42	Pure Red Cell Aplasia - a New Manifestation of CTLA4 Mutation. Blood, 2015, 126, 2225-2225.	0.6	0
43	A newly recognized 13q12.3 microdeletion syndrome characterized by intellectual disability, microcephaly, and eczema/atopic dermatitis encompassing the HMGB1 and KATNAL1 genes. American Journal of Medical Genetics, Part A, 2014, 164, 1277-1283.	0.7	25
44	A Dominant STIM1 Mutation Causes Stormorken Syndrome. Human Mutation, 2014, 35, 556-564.	1.1	143
45	Identification of copy number variants from exome sequence data. BMC Genomics, 2014, 15, 661.	1.2	59
46	PGM3 Mutations Cause a Congenital Disorder of Glycosylation with Severe Immunodeficiency and Skeletal Dysplasia. American Journal of Human Genetics, 2014, 95, 96-107.	2.6	148
47	Haploinsufficiency of two histone modifier genes on 6p22.3, ATXN1 and JARID2, is associated with intellectual disability. Orphanet Journal of Rare Diseases, 2013, 8, 3.	1.2	18
48	â€˜North Seaâ€™ progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. Brain, 2013, 136, 1146-1154.	3.7	129
49	Severe ALG8-CDG (CDG-Ih) associated with homozygosity for two novel missense mutations detected by exome sequencing of candidate genes. European Journal of Medical Genetics, 2012, 55, 196-202.	0.7	14
50	Prevalence of hereditary ataxia and spastic paraplegia in southeast Norway: a population-based study. Brain, 2009, 132, 1577-1588.	3.7	175
51	Two brothers with a microduplication including the MECP2 gene: rapid head growth in infancy and resolution of susceptibility to infection. Clinical Dysmorphology, 2009, 18, 78-82.	0.1	29
52	Infections Due to Various Atypical Mycobacteria in a Norwegian Multiplex Family with Dominant Interferon-Â Receptor Deficiency. Clinical Infectious Diseases, 2008, 46, e23-e27.	2.9	27
53	Schimke immunoosseous dysplasia: suggestions of genetic diversity. Human Mutation, 2007, 28, 273-283.	1.1	49
54	Ocular findings in Norwegian patients with ataxia-telangiectasia: a 5â€ƒyear prospective cohort study. Acta Ophthalmologica, 2007, 85, 557-562.	0.4	10

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55	Ocular findings in Norwegian patients with ataxia-telangiectasia: a 5Âyear prospective cohort study. Acta Ophthalmologica, 2007, 85, 557-562.	0.4	12
56	Single-cell analysis of normal and FOXP3-mutant human T cells: FOXP3 expression without regulatory T cell development. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 6659-6664.	3.3	698
57	Coping, quality of life, and hope in adults with primary antibody deficiencies. Health and Quality of Life Outcomes, 2005, 3, 31.	1.0	53
58	Chronic mucocutaneous candidiasis and primary hypothyroidism in two families. European Journal of Pediatrics, 2004, 163, 604-11.	1.3	10
59	Primary immunodeficiency diseases in Norway. Journal of Clinical Immunology, 2000, 20, 477-485.	2.0	163