

Kristina Lagerstedt-Robinson

List of Publications by Year in descending order

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Version: 2024-02-01

44
papers

1,436
citations

516215

16
h-index

344852

36
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46
all docs

46
docs citations

46
times ranked

3134
citing authors

#	ARTICLE	IF	CITATIONS
1	Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. <i>Nature Genetics</i> , 2014, 46, 107-115.	9.4	410
2	Lynch Syndrome (Hereditary Nonpolyposis Colorectal Cancer) Diagnostics. <i>Journal of the National Cancer Institute</i> , 2007, 99, 291-299.	3.0	201
3	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> , 2021, 13, 40.	3.6	116
4	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> , 2019, 11, 68.	3.6	88
5	Leiden open variation database of the MUTYH gene. <i>Human Mutation</i> , 2010, 31, 1205-1215.	1.1	72
6	Incidence of severe congenital neutropenia in Sweden and risk of evolution to myelodysplastic syndrome/leukaemia. <i>British Journal of Haematology</i> , 2012, 158, 363-369.	1.2	53
7	Mutational study of the MAMLD1-gene in hypospadias. <i>European Journal of Medical Genetics</i> , 2010, 53, 122-126.	0.7	51
8	Mismatch repair gene mutation spectrum in the Swedish Lynch syndrome population. <i>Oncology Reports</i> , 2016, 36, 2823-2835.	1.2	43
9	Novel Mutations Including Deletions of the Entire <i>OFD1</i> Gene in 30 Families with Type 1 Orofaciodigital Syndrome: A Study of the Extensive Clinical Variability. <i>Human Mutation</i> , 2013, 34, 237-247.	1.1	41
10	Targeted high-throughput sequencing for genetic diagnostics of hemophagocytic lymphohistiocytosis. <i>Genome Medicine</i> , 2015, 7, 130.	3.6	37
11	Parafibromin immunostainings of parathyroid tumors in clinical routine: a near-decade experience from a tertiary center. <i>Modern Pathology</i> , 2019, 32, 1082-1094.	2.9	35
12	A novel phenotype in N-glycosylation disorders: Gillessen-Kaesbach's Nishimura skeletal dysplasia due to pathogenic variants in <i>ALG9</i> . <i>European Journal of Human Genetics</i> , 2016, 24, 198-207.	1.4	29
13	Genomic screening in rare disorders: New mutations and phenotypes, highlighting <i>ALG14</i> as a novel cause of severe intellectual disability. <i>Clinical Genetics</i> , 2018, 94, 528-537.	1.0	29
14	Chimerism resulting from parthenogenetic activation and dispermic fertilization. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2277-2286.	0.7	28
15	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. <i>Journal of Human Genetics</i> , 2021, 66, 995-1008.	1.1	19
16	Multigeneration Inheritance through Fertile XX Carriers of an <i>NROB1</i> (<i>DAX1</i>) Locus Duplication in a Kindred of Females with Isolated XY Gonadal Dysgenesis. <i>International Journal of Endocrinology</i> , 2012, 2012, 1-7.	0.6	17
17	Benign paroxysmal torticollis of infancy does not lead to neurological sequelae. <i>Developmental Medicine and Child Neurology</i> , 2018, 60, 1251-1255.	1.1	17
18	Genetic anticipation in Swedish Lynch syndrome families. <i>PLoS Genetics</i> , 2017, 13, e1007012.	1.5	14

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19	X-linked Malformation Deafness: Neurodevelopmental Symptoms Are Common in Children With IP3 Malformation and Mutation in POU3F4. <i>Ear and Hearing</i> , 2022, 43, 53-69.	1.0	12
20	A retrospective study of extracolonic, non-endometrial cancer in Swedish Lynch syndrome families. <i>Hereditary Cancer in Clinical Practice</i> , 2018, 16, 16.	0.6	11
21	Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions. <i>Nature Communications</i> , 2019, 10, 1396.	5.8	11
22	Detection of germline mosaicism in fathers of children with intellectual disability syndromes caused by de novo variants. <i>Molecular Genetics & Genomic Medicine</i> , 2022, 10, e1880.	0.6	10
23	A complex DICER1 syndrome phenotype associated with a germline pathogenic variant affecting the RNase IIIa domain of DICER1. <i>Journal of Medical Genetics</i> , 2022, 59, 141-146.	1.5	9
24	<i>SLC1A3</i> variant associated with hemiplegic migraine and acetazolamide-responsive MRS changes. <i>Neurology: Genetics</i> , 2020, 6, e474.	0.9	9
25	Chorea, psychosis, acanthocytosis, and prolonged survival associated with <i>ELAC2</i> mutations. <i>Neurology</i> , 2018, 91, 710-712.	1.5	8
26	No association between a promoter NOS1 polymorphism (rs41279104) and Infantile Hypertrophic Pyloric Stenosis. <i>Journal of Human Genetics</i> , 2009, 54, 706-708.	1.1	7
27	Genetic analyses supporting colorectal, gastric, and prostate cancer syndromes. <i>Genes Chromosomes and Cancer</i> , 2019, 58, 775-782.	1.5	6
28	The Apparent Genetic Anticipation in PMS2-Associated Lynch Syndrome Families Is Explained by Birth-cohort Effect. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2019, 28, 1010-1014.	1.1	6
29	A Case with Bladder Exstrophy and Unbalanced X Chromosome Rearrangement. <i>European Journal of Pediatric Surgery</i> , 2014, 24, 353-359.	0.7	5
30	Novel Mutation of the TGF- β 3 Protein (Loeys-Dietz Type 5) Associated With Aortic and Carotid Dissections. <i>Neurology: Genetics</i> , 2021, 7, e625.	0.9	5
31	A retrospective two centre study of Birt-Hogg-Dubouché syndrome reveals a pathogenic founder mutation in FLCN in the Swedish population. <i>PLoS ONE</i> , 2022, 17, e0264056.	1.1	5
32	PatientMatcher: A customizable Python-based open-source tool for matching undiagnosed rare disease patients via the Matchmaker Exchange network. <i>Human Mutation</i> , 2022, , .	1.1	5
33	Double <i>de novo</i> mutations of <i>ELANE</i> (<i>ELA2</i>) in a patient with severe congenital neutropenia requiring high-dose G-CSF therapy. <i>British Journal of Haematology</i> , 2009, 147, 587-590.	1.2	4
34	Partial tetrasomy 14 associated with multiple malformations. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1284-1290.	0.7	4
35	Testing strategies to reduce morbidity and mortality from Lynch syndrome. <i>Scandinavian Journal of Gastroenterology</i> , 2018, 53, 1535-1540.	0.6	4
36	Haplotype analysis suggest that the MLH1 c.2059C>T mutation is a Swedish founder mutation. <i>Familial Cancer</i> , 2018, 17, 531-537.	0.9	3

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37	Massive parallel sequencing in a family with rectal cancer. <i>Hereditary Cancer in Clinical Practice</i> , 2021, 19, 23.	0.6	3
38	Pathogenic variant in the <i>COL2A1</i> gene is associated with Spondyloepiphyseal dysplasia type Stanescu. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 266-269.	0.7	2
39	Altered CSF levels of monoamines in hereditary spastic paraparesis 10. <i>Neurology: Genetics</i> , 2019, 5, e344.	0.9	2
40	Sequencing for germline mutations in Swedish breast cancer families reveals novel breast cancer risk genes. <i>Scientific Reports</i> , 2021, 11, 14737.	1.6	2
41	Merged Testing for Colorectal Cancer Syndromes and Reevaluation of Genetic Variants Improve Diagnostic Yield: results from a nationwide prospective cohort. <i>Genes Chromosomes and Cancer</i> , 2022, , .	1.5	2
42	Discordant Reporting of a Previously Undescribed Pathogenic Germline BRCA2 Variant in Blood and Tumor Tissue in a Patient With Pancreatic Adenocarcinoma. <i>JCO Precision Oncology</i> , 2021, 5, 974-980.	1.5	1
43	Increased risk for uterine cancer among first-degree relatives to Swedish gastric cancer patients. <i>Hereditary Cancer in Clinical Practice</i> , 2020, 18, 12.	0.6	0
44	Identification of known and novel familial cancer genes in Swedish colorectal cancer families. <i>International Journal of Cancer</i> , 2021, 149, 627-634.	2.3	0