Kristina Lagerstedt-Robinson

List of Publications by Year
in descending order

44 papers	1,436 citations	516710 16 h-index	345221 36 g-index
46 all docs	46 docs citations	46 times ranked	3134 citing authors

#	Article	IF	Citations
1	A complex DICER1 syndrome phenotype associated with a germline pathogenic variant affecting the RNase IIIa domain of DICER1. Journal of Medical Genetics, 2022, 59, 141-146.	3.2	9
2	X-linked Malformation Deafness: Neurodevelopmental Symptoms Are Common in Children With IP3 Malformation and Mutation in POU3F4. Ear and Hearing, 2022, 43, 53-69.	2.1	12
3	Detection of germline mosaicism in fathers of children with intellectual disability syndromes caused by de novo variants. Molecular Genetics & Genomic Medicine, 2022, 10, e1880.	1.2	10
4	A retrospective two centre study of Birt-Hogg-DubÃ© syndrome reveals a pathogenic founder mutation in FLCN in the Swedish population. PLoS ONE, 2022, 17, e0264056.	2.5	5
5	PatientMatcher: A customizable Pythonâ€based openâ€source tool for matching undiagnosed rare disease patients via the Matchmaker Exchange network. Human Mutation, 2022, , .	2.5	5
6	Merged Testing for Colorectal Cancer Syndromes and Reâ€evaluation of Genetic Variants Improve Diagnostic Yield: results from a nationâ€wide prospective cohort. Genes Chromosomes and Cancer, 2022, , .	2.8	2
7	Identification of known and novel familial cancer genes in Swedish colorectal cancer families. International Journal of Cancer, 2021, 149, 627-634.	5.1	0
8	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.	8.2	116
9	Massive parallel sequencing in a family with rectal cancer. Hereditary Cancer in Clinical Practice, 2021, 19, 23.	1.5	3
10	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.	2.3	19
11	Discordant Reporting of a Previously Undescribed Pathogenic Germline BRCA2 Variant in Blood and Tumor Tissue in a Patient With Pancreatic Adenocarcinoma. JCO Precision Oncology, 2021, 5, 974-980.	3.0	1
12	Sequencing for germline mutations in Swedish breast cancer families reveals novel breast cancer risk genes. Scientific Reports, 2021, 11, 14737.	3.3	2
13	Novel Mutation of the TGF-Î² 3 Protein (Loeys-Dietz Type 5) Associated With Aortic and Carotid Dissections. Neurology: Genetics, 2021, 7, e625.	1.9	5
14	SLC1A3 variant associated with hemiplegic migraine and acetazolamide-responsive MRS changes. Neurology: Genetics, 2020, 6, e474.	1.9	9
15	Increased risk for uterine cancer among first-degree relatives to Swedish gastric cancer patients. Hereditary Cancer in Clinical Practice, 2020, 18, 12.	1.5	0
16	Genetic analyses supporting colorectal, gastric, and prostate cancer syndromes. Genes Chromosomes and Cancer, 2019, 58, 775-782.	2.8	6
17	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.	8.2	88
18	The Apparent Genetic Anticipation in PMS2-Associated Lynch Syndrome Families Is Explained by Birth-cohort Effect. Cancer Epidemiology Biomarkers and Prevention, 2019, 28, 1010-1014.	2.5	6

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19	Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions. Nature Communications, 2019, 10, 1396.	12.8	11
20	Parafibromin immunostainings of parathyroid tumors in clinical routine: a near-decade experience from a tertiary center. Modern Pathology, 2019, 32, 1082-1094.	5.5	35
21	Altered CSF levels of monoamines in hereditary spastic paraparesis 10. Neurology: Genetics, 2019, 5, e344.	1.9	2
22	Haplotype analysis suggest that the MLH1 c.2059Câ€‰>â€‰T mutation is a Swedish founder mutation. Familial Cancer, 2018, 17, 531-537.	1.9	3
23	Testing strategies to reduce morbidity and mortality from Lynch syndrome. Scandinavian Journal of Gastroenterology, 2018, 53, 1535-1540.	1.5	4
24	A retrospective study of extracolonic, non-endometrial cancer in Swedish Lynch syndrome families. Hereditary Cancer in Clinical Practice, 2018, 16, 16.	1.5	11
25	Chorea, psychosis, acanthocytosis, and prolonged survival associated with ELAC2 mutations. Neurology, 2018, 91, 710-712.	1.1	8
26	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.	2.0	29
27	Benign paroxysmal torticollis of infancy does not lead to neurological sequelae. Developmental Medicine and Child Neurology, 2018, 60, 1251-1255.	2.1	17
28	Genetic anticipation in Swedish Lynch syndrome families. PLoS Genetics, 2017, 13, e1007012.	3.5	14
29	Mismatch repair gene mutation spectrum in the Swedish Lynch syndrome population. Oncology Reports, 2016, 36, 2823-2835.	2.6	43
30	Pathogenenic variant in the COL2A1 gene is associated with Spondyloepiphyseal dysplasia type Stanescu. American Journal of Medical Genetics, Part A, 2016, 170, 266-269.	1.2	2
31	A novel phenotype in N-glycosylation disorders: Gillessen-Kaesbachâ€"Nishimura skeletal dysplasia due to pathogenic variants in ALG9. European Journal of Human Genetics, 2016, 24, 198-207.	2.8	29
32	Targeted high-throughput sequencing for genetic diagnostics of hemophagocytic lymphohistiocytosis. Genome Medicine, 2015, 7, 130.	8.2	37
33	A Case with Bladder Exstrophy and Unbalanced X Chromosome Rearrangement. European Journal of Pediatric Surgery, 2014, 24, 353-359.	1.3	5
34	Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. Nature Genetics, 2014, 46, 107-115.	21.4	410
35	Partial tetrasomy 14 associated with multiple malformations. American Journal of Medical Genetics, Part A, 2013, 161, 1284-1290.	1.2	4
36	Novel Mutations Including Deletions of the EntireOFD1Gene in 30 Families with Type 1 Orofaciodigital Syndrome: A Study of the Extensive Clinical Variability. Human Mutation, 2013, 34, 237-247.	2.5	41

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37	Multigeneration Inheritance through Fertile XX Carriers of anNR0B1(DAX1) Locus Duplication in a Kindred of Females with Isolated XY Gonadal Dysgenesis. International Journal of Endocrinology, 2012, 2012, 1-7.	1.5	17
38	Incidence of severe congenital neutropenia in Sweden and risk of evolution to myelodysplastic syndrome/leukaemia. British Journal of Haematology, 2012, 158, 363-369.	2.5	53
39	Leiden open variation database of the MUTYH gene. Human Mutation, 2010, 31, 1205-1215.	2.5	72
40	Chimerism resulting from parthenogenetic activation and dispermic fertilization. American Journal of Medical Genetics, Part A, 2010, 152A, 2277-2286.	1.2	28
41	Mutational study of the MAMLD1-gene in hypospadias. European Journal of Medical Genetics, 2010, 53, 122-126.	1.3	51
42	Double de novo mutations of ELANE (ELA2) in a patient with severe congenital neutropenia requiring highâ€dose Gâ€CSF therapy. British Journal of Haematology, 2009, 147, 587-590.	2.5	4
43	No association between a promoter NOS1 polymorphism (rs41279104) and Infantile Hypertrophic Pyloric Stenosis. Journal of Human Genetics, 2009, 54, 706-708.	2.3	7
44	Lynch Syndrome (Hereditary Nonpolyposis Colorectal Cancer) Diagnostics. Journal of the National Cancer Institute, 2007, 99, 291-299.	6.3	201