

Cathrine Jespersgaard

List of Publications by Year in descending order

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Version: 2024-02-01

23
papers

755
citations

567281

15
h-index

642732

23
g-index

23
all docs

23
docs citations

23
times ranked

1600
citing authors

#	ARTICLE	IF	CITATIONS
1	Disease Concordance, Zygosity, and NOD2/CARD15 Status: Follow-Up of a Population-Based Cohort of Danish Twins with Inflammatory Bowel Disease. <i>American Journal of Gastroenterology</i> , 2005, 100, 2486-2492.	0.4	89
2	Capillary electrophoresis-based single strand DNA conformation analysis in high-throughput mutation screening. <i>Human Mutation</i> , 2003, 21, 455-465.	2.5	87
3	Genome-wide peripheral blood leukocyte DNA methylation microarrays identified a single association with inflammatory bowel diseases. <i>Inflammatory Bowel Diseases</i> , 2012, 18, 2334-2341.	1.9	80
4	Molecular genetic analysis using targeted NGS analysis of 677 individuals with retinal dystrophy. <i>Scientific Reports</i> , 2019, 9, 1219.	3.3	76
5	Phenotypes and genotypes in individuals with <i>SMC1A</i> variants. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2108-2125.	1.2	69
6	High-throughput single strand conformation polymorphism mutation detection by automated capillary array electrophoresis: validation of the method. <i>Human Mutation</i> , 2003, 21, 116-122.	2.5	52
7	Determination of Beta-Defensin Genomic Copy Number in Different Populations: A Comparison of Three Methods. <i>PLoS ONE</i> , 2011, 6, e16768.	2.5	39
8	A pathogenic haplotype, common in Europeans, causes autosomal recessive albinism and uncovers missing heritability in OCA1. <i>Scientific Reports</i> , 2019, 9, 645.	3.3	29
9	<i>MTACYB</i> mutations in hypertrophic cardiomyopathy. <i>Molecular Genetics & Genomic Medicine</i> , 2013, 1, 54-65.	1.2	28
10	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. <i>PLoS Genetics</i> , 2018, 14, e1007780.	3.5	28
11	Optimization of capillary array electrophoresis single-strand conformation polymorphism analysis for routine molecular diagnostics. <i>Electrophoresis</i> , 2006, 27, 3816-3822.	2.4	21
12	Alpha-Defensin DEFA1A3 Gene Copy Number Elevation in Danish Crohn's Disease Patients. <i>Digestive Diseases and Sciences</i> , 2011, 56, 3517-3524.	2.3	21
13	TS-EUROTRAIN: A European-Wide Investigation and Training Network on the Etiology and Pathophysiology of Gilles de la Tourette Syndrome. <i>Frontiers in Neuroscience</i> , 2016, 10, 384.	2.8	21
14	Single-strand conformation polymorphism analysis using capillary array electrophoresis for large-scale mutation detection. <i>Nature Protocols</i> , 2007, 2, 1458-1466.	12.0	20
15	Genetic and environmental factors as predictors of disease severity and extent at time of diagnosis in an inception cohort of inflammatory bowel disease, Copenhagen County and City 2003-2005. <i>Journal of Crohn's and Colitis</i> , 2008, 2, 162-169.	1.3	19
16	Deletion of 11q12.3-11q13.1 in a patient with intellectual disability and childhood facial features resembling Cornelia de Lange syndrome. <i>Gene</i> , 2015, 572, 130-134.	2.2	16
17	Polymorphisms in inflammation genes, tobacco smoke and furred pets and wheeze in children. <i>Pediatric Allergy and Immunology</i> , 2009, 20, 614-623.	2.6	11
18	Mosaic <i>MECP2</i> variants in males with classical Rett syndrome features, including stereotypical hand movements. <i>Clinical Genetics</i> , 2019, 95, 403-408.	2.0	10

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19	Genetic Variability in Beta-Defensins Is Not Associated with Susceptibility to Staphylococcus aureus Bacteremia. PLoS ONE, 2012, 7, e32315.	2.5	8
20	Proximal 21q deletion as a result of a de novo unbalanced t(12;21) translocation in a patient with dysmorphic features, hepatomegaly, thick myocardium and delayed psychomotor development. Molecular Cytogenetics, 2016, 9, 11.	0.9	8
21	Heterozygous mutations in GTP-cyclohydrolase-1 reduce BH4 biosynthesis but not pain sensitivity. Pain, 2018, 159, 1012-1024.	4.2	8
22	Bi-Allelic Pathogenic Variations in MERTK Including Deletions Are Associated with an Early Onset Progressive Form of Retinitis Pigmentosa. Genes, 2020, 11, 1517.	2.4	8
23	Genetic and Environmental Factors in Monozygotic Twins with Crohn's Disease and Their First-Degree Relatives: A Case Report. Digestion, 2005, 71, 262-265.	2.3	7