Cathrine Jespersgaard

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/10616432/publications.pdf

Version: 2024-02-01

23	755	567281 15	642732 23
papers	citations	h-index	g-index

23	23	23	1600
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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
2	A pathogenic haplotype, common in Europeans, causes autosomal recessive albinism and uncovers missing heritability in OCA1. Scientific Reports, 2019, 9, 645.	3.3	29
3	Molecular genetic analysis using targeted NGS analysis of 677 individuals with retinal dystrophy. Scientific Reports, 2019, 9, 1219.	3.3	76
4	Mosaic MECP2 variants in males with classical Rett syndrome features, including stereotypical hand movements. Clinical Genetics, 2019, 95, 403-408.	2.0	10
5	Heterozygous mutations in GTP-cyclohydrolase-1 reduce BH4 biosynthesis but not pain sensitivity. Pain, 2018, 159, 1012-1024.	4.2	8
6	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. PLoS Genetics, 2018, 14, e1007780.	3.5	28
7	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	1.2	69
8	TS-EUROTRAIN: A European-Wide Investigation and Training Network on the Etiology and Pathophysiology of Gilles de la Tourette Syndrome. Frontiers in Neuroscience, 2016, 10, 384.	2.8	21
9	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
10	Deletion of 11q12.3â€“11q13.1 in a patient with intellectual disability and childhood facial features resembling Cornelia de Lange syndrome. Gene, 2015, 572, 130-134.	2.2	16
11	MTâ€CYB mutations in hypertrophic cardiomyopathy. Molecular Genetics & Genomic Medicine, 2013, 1, 54-65.	1.2	28
12	Genetic Variability in Beta-Defensins Is Not Associated with Susceptibility to Staphylococcus aureus Bacteremia. PLoS ONE, 2012, 7, e32315.	2.5	8
13	Genome-wide peripheral blood leukocyte DNA methylation microarrays identified a single association with inflammatory bowel diseases. Inflammatory Bowel Diseases, 2012, 18, 2334-2341.	1.9	80
14	Alpha-Defensin DEFA1A3 Gene Copy Number Elevation in Danish Crohnâ€™s Disease Patients. Digestive Diseases and Sciences, 2011, 56, 3517-3524.	2.3	21
15	Determination of Beta-Defensin Genomic Copy Number in Different Populations: A Comparison of Three Methods. PLoS ONE, 2011, 6, e16768.	2.5	39
16	Polymorphisms in inflammation genes, tobacco smoke and furred pets and wheeze in children. Pediatric Allergy and Immunology, 2009, 20, 614-623.	2.6	11
17	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 2003â€“2005. Journal of Crohn's and Colitis, 2008, 2, 162-169.	1.3	19
18	Single-strand conformation polymorphism analysis using capillary array electrophoresis for large-scale mutation detection. Nature Protocols, 2007, 2, 1458-1466.	12.0	20

#	Article	IF	Citations
19	Optimization of capillary array electrophoresis single-strand conformation polymorphism analysis for routine molecular diagnostics. Electrophoresis, 2006, 27, 3816-3822.	2.4	21
20	Disease Concordance, Zygosity, and NOD2/CARD15 Status: Follow-Up of a Population-Based Cohort of Danish Twins with Inflammatory Bowel Disease. American Journal of Gastroenterology, 2005, 100, 2486-2492.	0.4	89
21	Genetic and Environmental Factors in Monozygotic Twins with Crohnâ€™s Disease and Their First-Degree Relatives: A Case Report. Digestion, 2005, 71, 262-265.	2.3	7
22	High-throughput single strand conformation polymorphism mutation detection by automated capillary array electrophoresis: validation of the method. Human Mutation, 2003, 21, 116-122.	2.5	52
23	Capillary electrophoresis-based single strand DNA conformation analysis in high-throughput mutation screening. Human Mutation, 2003, 21, 455-465.	2.5	87