Henrik Stranneheim

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/10581310/publications.pdf

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26	1,153	471509	580821
papers	citations	17 h-index	25 g-index

26	26	26	2590
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in SLC12A5 in epilepsy of infancy with migrating focal seizures. Nature Communications, 2015, 6, 8038.	12.8	160
2	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
3	Increased Throughput by Parallelization of Library Preparation for Massive Sequencing. PLoS ONE, 2010, 5, e10029.	2.5	114
4	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. American Journal of Human Genetics, 2016, 99, 735-743.	6.2	99
5	Rescue of primary ubiquinone deficiency due to a novelCOQ7defect using 2,4â€“dihydroxybensoic acid. Journal of Medical Genetics, 2015, 52, 779-783.	3.2	94
6	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
7	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. American Journal of Human Genetics, 2015, 97, 761-768.	6.2	58
8	Classification of DNA sequences using Bloom filters. Bioinformatics, 2010, 26, 1595-1600.	4.1	57
9	Rapid pulsed whole genome sequencing for comprehensive acute diagnostics of inborn errors of metabolism. BMC Genomics, 2014, 15, 1090.	2.8	54
10	Sun-Induced Nonsynonymous p53 Mutations Are Extensively Accumulated and Tolerated in Normal Appearing Human Skin. Journal of Investigative Dermatology, 2011, 131, 504-508.	0.7	49
11	Gene Expression Profiles in Paired Gingival Biopsies from Periodontitis-Affected and Healthy Tissues Revealed by Massively Parallel Sequencing. PLoS ONE, 2012, 7, e46440.	2.5	44
12	Comparison of total and cytoplasmic mRNA reveals global regulation by nuclear retention and miRNAs. BMC Genomics, 2012, 13, 574.	2.8	35
13	SQSTM1/p62-Directed Metabolic Reprogramming Is Essential for Normal Neurodifferentiation. Stem Cell Reports, 2019, 12, 696-711.	4.8	32
14	Epilepsy syndromes, etiologies, and the use of nextâ€generation sequencing in epilepsy presenting in the first 2 years of life: A populationâ€based study. Epilepsia, 2020, 61, 2486-2499.	5.1	24
15	Respiratory chain complex III deficiency due to mutated BCS1L: a novel phenotype with encephalomyopathy, partially phenocopied in a Bcs1l mutant mouse model. Orphanet Journal of Rare Diseases, 2017, 12, 73.	2.7	20
16	SLC12A2 mutations cause NKCC1 deficiency with encephalopathy and impaired secretory epithelia. Neurology: Genetics, 2020, 6, e478.	1.9	20
17	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
18	Scalable Transcriptome Preparation for Massive Parallel Sequencing. PLoS ONE, 2011, 6, e21910.	2.5	18

#	Article	IF	Citations
19	Biotin and Thiamine Responsive Basal Ganglia Disease â€“ A vital differential diagnosis in infants with severe encephalopathy. European Journal of Paediatric Neurology, 2016, 20, 457-461.	1.6	18
20	Chorea, psychosis, acanthocytosis, and prolonged survival associated with ELAC2 mutations. Neurology, 2018, 91, 710-712.	1.1	8
21	Severe congenital lactic acidosis and hypertrophic cardiomyopathy caused by an intronic variant in NDUFB7. Human Mutation, 2021, 42, 378-384.	2.5	8
22	Clinical Presentation, Genetic Etiology, and Coenzyme Q10 Levels in 55 Children with Combined Enzyme Deficiencies of the Mitochondrial Respiratory Chain. Journal of Pediatrics, 2021, 228, 240-251.e2.	1.8	6
23	Loqusdb: added value of an observations database of local genomic variation. BMC Bioinformatics, 2020, 21, 273.	2.6	5
24	A comparison between protein profiles of B cell subpopulations and mantle cell lymphoma cells. Proteome Science, 2009, 7, 43.	1.7	4
25	Novel Mutation m.10372A>G in MT-ND3 Causing Sensorimotor Axonal Polyneuropathy. Neurology: Genetics, 2021, 7, e566.	1.9	3
26	J10â€…Chorea, psychotic symptoms and long survival in a subject with ELAC2 mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A78.3-A79.	1.9	0