Karl Sperling

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8284814/publications.pdf

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25 papers	2,562 citations	623734 14 h-index	610901 24 g-index
31 all docs	31 docs citations	31 times ranked	2566 citing authors

#	Article	IF	Citations
1	Nibrin, a Novel DNA Double-Strand Break Repair Protein, Is Mutated in Nijmegen Breakage Syndrome. Cell, 1998, 93, 467-476.	28.9	989
2	Intracytoplasmic Sperm Injection May Increase the Risk of Imprinting Defects. American Journal of Human Genetics, 2002, 71, 162-164.	6.2	660
3	Nijmegen breakage syndrome: clinical manifestation of defective response to DNA double-strand breaks. DNA Repair, 2004, 3, 1207-1217.	2.8	212
4	Clinical ascertainment of Nijmegen breakage syndrome (NBS) and prevalence of the major mutation, 657del5, in three Slav populations. European Journal of Human Genetics, 2000, 8, 900-902.	2.8	130
5	Nibrin functions in Ig class-switch recombination. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 1584-1589.	7.1	98
6	Cancer Risk of Heterozygotes With the NBN Founder Mutation. Journal of the National Cancer Institute, 2007, 99, 1875-1880.	6.3	79
7	A novel mutation in the DNA-binding domain ofMAF at 16q23.1 associated with autosomal dominant â€œcerulean cataractâ€•in an Indian family. American Journal of Medical Genetics, Part A, 2006, 140A, 558-566.	1.2	74
8	Spectrum of mutations in the Fanconi anaemia group G gene, FANCG/XRCC9. European Journal of Human Genetics, 2000, 8, 861-868.	2.8	61
9	SV40 large T-antigen disturbs the formation of nuclear DNA-repair foci containing MRE11. Oncogene, 2002, 21, 4873-4878.	5.9	57
10	Phenotypic differences in Angelman syndrome patients: Imprinting mutations show less frequently microcephaly and hypopigmentation than deletions. , 1996, 66, 221-226.		35
11	Evidence for an increase in trisomy 21 (Down syndrome) in Europe after the Chernobyl reactor accident. Genetic Epidemiology, 2012, 36, 48-55.	1.3	33
12	Localisation of a Fanconi anaemia gene to chromosome 9p. European Journal of Human Genetics, 1998, 6, 501-508.	2.8	22
13	Multisite de novo mutations in human offspring after paternal exposure to ionizing radiation. Scientific Reports, 2018, 8, 14611.	3.3	22
14	The Slavic NBN Founder Mutation: A Role for Reproductive Fitness?. PLoS ONE, 2016, 11, e0167984.	2.5	21
15	From proteomics to genomics. Electrophoresis, 2001, 22, 2835-2837.	2.4	16
16	Nijmegen Breakage Syndrome fibroblasts and iPSCs: cellular models for uncovering disease-associated signaling pathways and establishing a screening platform for anti-oxidants. Scientific Reports, 2017, 7, 7516.	3.3	13
17	Directed Alternative Splicing in Nijmegen Breakage Syndrome: Proof of Principle Concerning Its Therapeutical Application. Molecular Therapy, 2016, 24, 117-124.	8.2	8
18	Pattern of Ultra-violet-light-induced Repair in Metaphase and Interphase Chromosomes. International Journal of Radiation Biology and Related Studies in Physics, Chemistry, and Medicine, 1978, 34, 575-582.	1.0	6

#	Article	IF	Citations
19	Image analysis of neutrophil nuclear morphology: Learning about phenotypic range and its reliable analysis from patients with pelgerâ€HuÃ«tâ€anomaly and treated with colchicine. Cytometry Part B - Clinical Cytometry, 2017, 92, 541-549.	1.5	6
20	Telomere attrition and dysfunction: a potential trigger of the progeroid phenotype in nijmegen breakage syndrome. Aging, 2020, 12, 12342-12375.	3.1	6
21	Die Bedeutung der Epigenese fÃ¼r das VerstÃndnis der Pathogenese aus humangenetischer Sicht. Journal Fur Verbraucherschutz Und Lebensmittelsicherheit, 2008, 3, 9-17.	1.4	3
22	Evidence for a pre-malignant cell line in a skin biopsy from a patient with Nijmegen breakage syndrome. Molecular Cytogenetics, 2018, 11, 17.	0.9	3
23	Transmission ratio distortion of mutations in the master regulator of centriole biogenesis PLK4. Human Genetics, 2022, 141, 1785-1794.	3.8	3
24	Phenotypic differences in Angelman syndrome patients: Imprinting mutations show less frequently microcephaly and hypopigmentation than deletions. American Journal of Medical Genetics Part A, 1996, 66, 221-226.	2.4	1
25	Yeast XRS2 and human NBN gene: Experimental evidence for homology using codon optimized cDNA. PLoS ONE, 2018, 13, e0207315.	2.5	0