

Silvia Bione

List of Publications by Year in descending order

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Version: 2024-02-01

40
papers

3,990
citations

257101

24
h-index

288905

40
g-index

41
all docs

41
docs citations

41
times ranked

3936
citing authors

#	ARTICLE	IF	CITATIONS
1	Identification of a novel X-linked gene responsible for Emery-Dreifuss muscular dystrophy. <i>Nature Genetics</i> , 1994, 8, 323-327.	9.4	857
2	A novel X-linked gene, G4.5. is responsible for Barth syndrome. <i>Nature Genetics</i> , 1996, 12, 385-389.	9.4	718
3	Different Mutations in the LMNA Gene Cause Autosomal Dominant and Autosomal Recessive Emery-Dreifuss Muscular Dystrophy. <i>American Journal of Human Genetics</i> , 2000, 66, 1407-1412.	2.6	384
4	A Human Homologue of the <i>Drosophila melanogaster</i> diaphanous Gene Is Disrupted in a Patient with Premature Ovarian Failure: Evidence for Conserved Function in Oogenesis and Implications for Human Sterility. <i>American Journal of Human Genetics</i> , 1998, 62, 533-541.	2.6	248
5	The X-Linked Gene G4.5 Is Responsible for Different Infantile Dilated Cardiomyopathies. <i>American Journal of Human Genetics</i> , 1997, 61, 862-867.	2.6	236
6	A family of transmembrane proteins with homology to the MET-hepatocyte growth factor receptor.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1996, 93, 674-678.	3.3	169
7	Influence of intermediate and uninterrupted FMR1 CGG expansions in premature ovarian failure manifestation. <i>Human Reproduction</i> , 2006, 21, 952-957.	0.4	162
8	BMP15 mutations associated with primary ovarian insufficiency cause a defective production of bioactive protein. <i>Human Mutation</i> , 2009, 30, 804-810.	1.1	126
9	Alterations in the expression, structure and function of progesterone receptor membrane component-1 (PGRMC1) in premature ovarian failure. <i>Human Molecular Genetics</i> , 2008, 17, 3776-3783.	1.4	114
10	Chromosomal rearrangements in Xq and premature ovarian failure: mapping of 25 new cases and review of the literature. <i>Human Reproduction</i> , 2006, 21, 1477-1483.	0.4	105
11	Identification of new mutations in the Emery-Dreifuss muscular dystrophy gene and evidence for genetic heterogeneity of the disease. <i>Human Molecular Genetics</i> , 1995, 4, 1859-1863.	1.4	93
12	Mutation analysis of two candidate genes for premature ovarian failure, DACH2 and POF1B. <i>Human Reproduction</i> , 2004, 19, 2759-2766.	0.4	82
13	Transcriptional organization of a 450-kb region of the human X chromosome in Xq28. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1993, 90, 10977-10981.	3.3	67
14	TERRA transcription destabilizes telomere integrity to initiate break-induced replication in human ALT cells. <i>Nature Communications</i> , 2021, 12, 3760.	5.8	60
15	An X chromosome-linked gene encoding a protein with characteristics of a rhoGAP predominantly expressed in hematopoietic cells.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1996, 93, 695-699.	3.3	50
16	Sequence variation at the human FOXO3 locus: a study of premature ovarian failure and primary amenorrhea. <i>Human Reproduction</i> , 2007, 23, 216-221.	0.4	49
17	Methylation and sequence analysis around Eagi sites: identification of 28 new CpG islands in XQ24-XQ28. <i>Nucleic Acids Research</i> , 1992, 20, 727-733.	6.5	44
18	A susceptibility gene for premature ovarian failure (POF) maps to proximal Xq28. <i>European Journal of Human Genetics</i> , 2004, 12, 829-834.	1.4	44

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19	Bi-allelic TARS Mutations Are Associated with Brittle Hair Phenotype. <i>American Journal of Human Genetics</i> , 2019, 105, 434-440.	2.6	42
20	A mutation in the X-linked Emeryâ€Dreifuss muscular dystrophy gene in a patient affected with conduction cardiomyopathy. <i>Neuromuscular Disorders</i> , 2001, 11, 411-413.	0.3	37
21	Heritability and Demographic Analyses in the Large Isolated Population of Val Borbera Suggest Advantages in Mapping Complex Traits Genes. <i>PLoS ONE</i> , 2009, 4, e7554.	1.1	37
22	Epigenetic control of the critical region for premature ovarian failure on autosomal genes translocated to the X chromosome: a hypothesis. <i>Human Genetics</i> , 2007, 121, 441-450.	1.8	35
23	A large-scale association study to assess the impact of known variants of the human INHA gene on premature ovarian failure. <i>Human Reproduction</i> , 2009, 24, 2023-2028.	0.4	30
24	Variation of hemoglobin levels in normal Italian populations from genetic isolates. <i>Haematologica</i> , 2008, 93, 1372-1375.	1.7	25
25	Are Myocardial Infarctionâ€Associated Single-Nucleotide Polymorphisms Associated With Ischemic Stroke?. <i>Stroke</i> , 2012, 43, 980-986.	1.0	25
26	X Chromosome Genes and Premature Ovarian Failure. <i>Seminars in Reproductive Medicine</i> , 2000, 18, 051-058.	0.5	23
27	Skewed X-chromosome inactivation is not associated with premature ovarian failure in a large cohort of Italian patients. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1349-1351.	0.7	16
28	Heterogeneous molecular mechanisms underlie attenuated familial adenomatous polyposis. <i>Genetics in Medicine</i> , 2007, 9, 836-841.	1.1	14
29	Unusual expression of emerin in a patient with X-linked Emeryâ€Dreifuss muscular dystrophy. <i>Neuromuscular Disorders</i> , 2000, 10, 567-571.	0.3	13
30	Association of a variant in the CHRNA5-A3-B4 gene cluster region to heavy smoking in the Italian population. <i>European Journal of Human Genetics</i> , 2011, 19, 593-596.	1.4	13
31	Spatial and temporal expression of POF1B, a gene expressed in epithelia. <i>Gene Expression Patterns</i> , 2007, 7, 529-534.	0.3	12
32	Cross-Analysis of Gene and miRNA Genome-Wide Expression Profiles in Human Fibroblasts at Different Stages of Transformation. <i>OMICS A Journal of Integrative Biology</i> , 2012, 16, 24-36.	1.0	12
33	Sequence and gene content in 52 kb including and centromeric to the G6PD gene in Xq28. <i>DNA Sequence</i> , 1995, 6, 1-11.	0.7	11
34	Reduced levels of prostaglandin I ₂ synthase: a distinctive feature of the cancer-free trichothiodystrophy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	8
35	A Comparative Transcriptional Map of a Region of 250 kb on the Human and Mouse X Chromosome between the G6PD and the FLN1 Genes. <i>Genomics</i> , 1995, 28, 377-382.	1.3	7
36	An Association Rule Mining Approach to Discover lncRNAs Expression Patterns in Cancer Datasets. <i>BioMed Research International</i> , 2015, 2015, 1-13.	0.9	7

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37	Highly Conserved Non-Coding Sequences and the 18q Critical Region for Short Stature: A Common Mechanism of Disease?. PLoS ONE, 2008, 3, e1460.	1.1	7
38	CorrelGenes: a new tool for the interpretation of the human transcriptome. BMC Bioinformatics, 2014, 15, S6.	1.2	4
39	A Role for Human DNA Polymerase δ in Alternative Lengthening of Telomeres. International Journal of Molecular Sciences, 2021, 22, 2365.	1.8	3
40	CorrelGenes: a new tool for the interpretation of the human transcriptome. EMBnet Journal, 2012, 18, 103.	0.2	1