Silvia Bione

List of Publications by Year
in descending order

40 papers	3,990 citations	257450 24 h-index	289244 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. Nature Genetics, 1994, 8, 323-327.	21.4	857
2	A novel X-linked gene, G4.5. is responsible for Barth syndrome. Nature Genetics, 1996, 12, 385-389.	21.4	718
3	Different Mutations in the LMNA Gene Cause Autosomal Dominant and Autosomal Recessive Emery-Dreifuss Muscular Dystrophy. American Journal of Human Genetics, 2000, 66, 1407-1412.	6.2	384
4	A Human Homologue of the Drosophila melanogaster diaphanous Gene Is Disrupted in a Patient with Premature Ovarian Failure: Evidence for Conserved Function in Oogenesis and Implications for Human Sterility. American Journal of Human Genetics, 1998, 62, 533-541.	6.2	248
5	The X-Linked Gene G4.5 Is Responsible for Different Infantile Dilated Cardiomyopathies. American Journal of Human Genetics, 1997, 61, 862-867.	6.2	236
6	A family of transmembrane proteins with homology to the MET-hepatocyte growth factor receptor.. Proceedings of the National Academy of Sciences of the United States of America, 1996, 93, 674-678.	7.1	169
7	Influence of intermediate and uninterrupted FMR1 CGG expansions in premature ovarian failure manifestation. Human Reproduction, 2006, 21, 952-957.	0.9	162
8	BMP15 mutations associated with primary ovarian insufficiency cause a defective production of bioactive protein. Human Mutation, 2009, 30, 804-810.	2.5	126
9	Alterations in the expression, structure and function of progesterone receptor membrane component-1 (PGRMC1) in premature ovarian failure. Human Molecular Genetics, 2008, 17, 3776-3783.	2.9	114
10	Chromosomal rearrangements in Xq and premature ovarian failure: mapping of 25 new cases and review of the literature. Human Reproduction, 2006, 21, 1477-1483.	0.9	105
11	Identification of new mutations in the Emery-Dreifuss muscular dystrophy gene and evidence for genetic heterogeneity of the disease. Human Molecular Genetics, 1995, 4, 1859-1863.	2.9	93
12	Mutation analysis of two candidate genes for premature ovarian failure, DACH2 and POF1B. Human Reproduction, 2004, 19, 2759-2766.	0.9	82
13	Transcriptional organization of a 450-kb region of the human X chromosome in Xq28. Proceedings of the National Academy of Sciences of the United States of America, 1993, 90, 10977-10981.	7.1	67
14	TERRA transcription destabilizes telomere integrity to initiate break-induced replication in human ALT cells. Nature Communications, 2021, 12, 3760.	12.8	60
15	An X chromosome-linked gene encoding a protein with characteristics of a rhoGAP predominantly expressed in hematopoietic cells.. Proceedings of the National Academy of Sciences of the United States of America, 1996, 93, 695-699.	7.1	50
16	Sequence variation at the human FOXO3 locus: a study of premature ovarian failure and primary amenorrhea. Human Reproduction, 2007, 23, 216-221.	0.9	49
17	Methylation and sequence analysis around Eagi sites: identification of 28 new CpG islands in XQ24-XQ28. Nucleic Acids Research, 1992, 20, 727-733.	14.5	44
18	A susceptibility gene for premature ovarian failure (POF) maps to proximal Xq28. European Journal of Human Genetics, 2004, 12, 829-834.	2.8	44

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19	Bi-allelic TARS Mutations Are Associated with Brittle Hair Phenotype. American Journal of Human Genetics, 2019, 105, 434-440.	6.2	42
20	A mutation in the X-linked Emeryâ€"Dreifuss muscular dystrophy gene in a patient affected with conduction cardiomyopathy. Neuromuscular Disorders, 2001, 11, 411-413.	0.6	37
21	Heritability and Demographic Analyses in the Large Isolated Population of Val Borbera Suggest Advantages in Mapping Complex Traits Genes. PLoS ONE, 2009, 4, e7554.	2.5	37
22	Epigenetic control of the critical region for premature ovarian failure on autosomal genes translocated to the X chromosome: a hypothesis. Human Genetics, 2007, 121, 441-450.	3.8	35
23	A large-scale association study to assess the impact of known variants of the human INHA gene on premature ovarian failure. Human Reproduction, 2009, 24, 2023-2028.	0.9	30
24	Variation of hemoglobin levels in normal Italian populations from genetic isolates. Haematologica, 2008, 93, 1372-1375.	3.5	25
25	Are Myocardial Infarctionâ€"Associated Single-Nucleotide Polymorphisms Associated With Ischemic Stroke?. Stroke, 2012, 43, 980-986.	2.0	25
26	X Chromosome Genes and Premature Ovarian Failure. Seminars in Reproductive Medicine, 2000, 18, 051-058.	1.1	23
27	Skewed X-chromosome inactivation is not associated with premature ovarian failure in a large cohort of Italian patients. American Journal of Medical Genetics, Part A, 2006, 140A, 1349-1351.	1.2	16
28	Heterogeneous molecular mechanisms underlie attenuated familial adenomatous polyposis. Genetics in Medicine, 2007, 9, 836-841.	2.4	14
29	Unusual expression of emerin in a patient with X-linked Emeryâ€"Dreifuss muscular dystrophy. Neuromuscular Disorders, 2000, 10, 567-571.	0.6	13
30	Association of a variant in the CHRNA5-A3-B4 gene cluster region to heavy smoking in the Italian population. European Journal of Human Genetics, 2011, 19, 593-596.	2.8	13
31	Spatial and temporal expression of POF1B, a gene expressed in epithelia. Gene Expression Patterns, 2007, 7, 529-534.	0.8	12
32	Cross-Analysis of Gene and miRNA Genome-Wide Expression Profiles in Human Fibroblasts at Different Stages of Transformation. OMICS A Journal of Integrative Biology, 2012, 16, 24-36.	2.0	12
33	Sequence and gene content in 52 kb including and centromeric to the G6PD gene in Xq28. DNA Sequence, 1995, 6, 1-11.	0.7	11
34	Reduced levels of prostaglandin I ₂ synthase: a distinctive feature of the cancer-free trichothiodystrophy. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	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. Genomics, 1995, 28, 377-382.	2.9	7
36	An Association Rule Mining Approach to Discover lncRNAs Expression Patterns in Cancer Datasets. BioMed Research International, 2015, 2015, 1-13.	1.9	7

#	Article	IF	Citations
37	Highly Conserved Non-Coding Sequences and the 18q Critical Region for Short Stature: A Common Mechanism of Disease?. PLoS ONE, 2008, 3, e1460.	2.5	7
38	CorrelaGenes: a new tool for the interpretation of the human transcriptome. BMC Bioinformatics, 2014, 15, S6.	2.6	4
39	A Role for Human DNA Polymerase Î» in Alternative Lengthening of Telomeres. International Journal of Molecular Sciences, 2021, 22, 2365.	4.1	3
40	CorrelaGenes: a new tool for the interpretation of the human transcriptome. EMBnet Journal, 2012, 18, 103.	0.6	1