Luciano Calzari

List of Publications by Year
in descending order

27 papers	674 citations	687220 13 h-index	580701 25 g-index
29 all docs	29 docs citations	29 times ranked	1030 citing authors

#	Article	IF	Citations
1	(Epi)genotypeâ€“phenotype correlations in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 183-190.	1.4	113
2	Stochastic epigenetic mutations (DNA methylation) increase exponentially in human aging and correlate with X chromosome inactivation skewing in females. Aging, 2015, 7, 568-578.	1.4	68
3	A multi-method approach to the molecular diagnosis of overt and borderline 11p15.5 defects underlying Silverâ€“Russell and Beckwithâ€“Wiedemann syndromes. Clinical Epigenetics, 2016, 8, 23.	1.8	54
4	Involvement of the yeast metacaspase Yca1 in Î”-programmed cell death. FEMS Yeast Research, 2004, 5, 141-147.	1.1	46
5	A KHDC3L mutation resulting in recurrent hydatidiform mole causes genome-wide DNA methylation loss in oocytes and persistent imprinting defects post-fertilisation. Genome Medicine, 2019, 11, 84.	3.6	45
6	Loss-of-function maternal-effect mutations of PADI6 are associated with familial and sporadic Beckwith-Wiedemann syndrome with multi-locus imprinting disturbance. Clinical Epigenetics, 2020, 12, 139.	1.8	40
7	Fetal growth patterns in Beckwithâ€“Wiedemann syndrome. Clinical Genetics, 2016, 90, 21-27.	1.0	34
8	Quantitative DNA methylation analysis improves epigenotype-phenotype correlations in Beckwith-Wiedemann syndrome. Epigenetics, 2013, 8, 1053-1060.	1.3	33
9	DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355.	1.0	28
10	The phenotypic variations of multi-locus imprinting disturbances associated with maternal-effect variants of NLRP5 range from overt imprinting disorder to apparently healthy phenotype. Clinical Epigenetics, 2019, 11, 190.	1.8	22
11	iPSC-derived neurons of CREBBP - and EP300 -mutated Rubinstein-Taybi syndrome patients show morphological alterations and hypoexcitability. Stem Cell Research, 2018, 30, 130-140.	0.3	19
12	Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. Genetical Research, 2019, 101, e3.	0.3	17
13	Genome-Wide DNA Methylation Analysis of a Cohort of 41 Patients Affected by Oculo-Auriculo-Vertebral Spectrum (OAVS). International Journal of Molecular Sciences, 2021, 22, 1190.	1.8	16
14	Transcriptome Analysis of iPSC-Derived Neurons from Rubinstein-Taybi Patients Reveals Deficits in Neuronal Differentiation. Molecular Neurobiology, 2020, 57, 3685-3701.	1.9	15
15	Taurine Administration Recovers Motor and Learning Deficits in an Angelman Syndrome Mouse Model. International Journal of Molecular Sciences, 2018, 19, 1088.	1.8	14
16	Ring Chromosome 20 Syndrome: Genetics, Clinical Characteristics, and Overlapping Phenotypes. Frontiers in Neurology, 2020, 11, 613035.	1.1	14
17	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. Clinical Epigenetics, 2022, 14, 41.	1.8	14
18	The Histone Deubiquitinating Enzyme Ubp10 Is Involved in rDNA Locus Control in Saccharomyces cerevisiae by Affecting Sir2p Association. Genetics, 2006, 174, 2249-2254.	1.2	13

#	Article	IF	Citations
19	Epigenome Wide Association and Stochastic Epigenetic Mutation Analysis on Cord Blood of Preterm Birth. International Journal of Molecular Sciences, 2020, 21, 5044.	1.8	12
20	Blood oxidative stress and metallothionein expression in Rett syndrome: Probing for markers. World Journal of Biological Psychiatry, 2016, 17, 198-209.	1.3	11
21	Molecular Etiology Disclosed by Array CGH in Patients With Silverâ€“Russell Syndrome or Similar Phenotypes. Frontiers in Genetics, 2019, 10, 955.	1.1	11
22	Extensive Placental Methylation Profiling in Normal Pregnancies. International Journal of Molecular Sciences, 2021, 22, 2136.	1.8	8
23	Generation of three iPSC lines (IALi002, IALi004, IALi003) from Rubinstein-Taybi syndrome 1 patients carrying CREBBP non sense c.4435G>T, p.(Gly1479*) and c.3474G>A, p.(Trp1158*) and missense c.4627G>T, p.(Asp1543Tyr) mutations. Stem Cell Research, 2019, 40, 101553.	0.3	6
24	Maternal Uniparental Disomy of Chromosome 20 (UPD(20)mat) as Differential Diagnosis of Silver Russell Syndrome: Identification of Three New Cases. Genes, 2021, 12, 588.	1.0	6
25	Germline variants in genes of the subcortical maternal complex and Multilocus Imprinting Disturbance are associated with miscarriage/infertility or Beckwithâ€“Wiedemann progeny. Clinical Epigenetics, 2022, 14, 43.	1.8	6
26	Genes for RNA-binding proteins involved in neural-specific functions and diseases are downregulated in Rubinstein-Taybi iNeurons. Neural Regeneration Research, 2022, 17, 5.	1.6	5
27	Interconnected Gene Networks Underpin the Clinical Overlap of HNRNPH1-Related and Rubinsteinâ€“Taybi Intellectual Disability Syndromes. Frontiers in Neuroscience, 2021, 15, 745684.	1.4	0