Laura Fontana

List of Publications by Year
in descending order

41 papers	837 citations	516710 16 h-index	526287 27 g-index
42 all docs	42 docs citations	42 times ranked	1612 citing authors

#	Article	IF	Citations
1	Genetics of Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser (<scp>MRKH</scp>) syndrome. Clinical Genetics, 2017, 91, 233-246.	2.0	117
2	Fragile X syndrome: a review of clinical and molecular diagnoses. Italian Journal of Pediatrics, 2017, 43, 39.	2.6	114
3	Decreased serum level of sphingosineâ€1â€phosphate: a novel predictor of clinical severity in COVIDâ€19. EMBO Molecular Medicine, 2021, 13, e13424.	6.9	70
4	Mitochondrial DNA content and methylation in fetal cord blood of pregnancies with placental insufficiency. Placenta, 2017, 55, 63-70.	1.5	47
5	Microtubule-associated protein/microtubule affinity-regulating kinase 4 (MARK4) plays a role in cell cycle progression and cytoskeletal dynamics. European Journal of Cell Biology, 2014, 93, 355-365.	3.6	36
6	Angiogenesis in human brain tumors: screening of drug response through a patient-specific cell platform for personalized therapy. Scientific Reports, 2018, 8, 8748.	3.3	29
7	The Genetic Landscape of Human Glioblastoma and Matched Primary Cancer Stem Cells Reveals Intratumour Similarity and Intertumour Heterogeneity. Stem Cells International, 2019, 2019, 1-12.	2.5	29
8	Beckwithâ€“Wiedemann syndrome prenatal diagnosis by methylation analysis in chorionic villi. Epigenetics, 2015, 10, 643-649.	2.7	26
9	Molecular Insights into the Classification of Luminal Breast Cancers: The Genomic Heterogeneity of Progesterone-Negative Tumors. International Journal of Molecular Sciences, 2019, 20, 510.	4.1	25
10	Differential Signature of the Centrosomal MARK4 Isoforms in Glioma. Analytical Cellular Pathology, 2011, 34, 319-338.	1.4	23
11	Constitutive BRCA1 Promoter Hypermethylation Can Be a Predisposing Event in Isolated Early-Onset Breast Cancer. Cancers, 2019, 11, 58.	3.7	22
12	Characterization of multi-locus imprinting disturbances and underlying genetic defects in patients with chromosome 11p15.5 related imprinting disorders. Epigenetics, 2018, 13, 897-909.	2.7	21
13	Rothmund-Thomson Syndrome: Insights from New Patients on the Genetic Variability Underpinning Clinical Presentation and Cancer Outcome. International Journal of Molecular Sciences, 2018, 19, 1103.	4.1	20
14	MGMT-Methylated Alleles Are Distributed Heterogeneously Within Glioma Samples Irrespective ofIDHStatus and Chromosome 10q Deletion. Journal of Neuropathology and Experimental Neurology, 2016, 75, 791-800.	1.7	19
15	A HS6ST2 gene variant associated with Xâ€linked intellectual disability and severe myopia in two male twins. Clinical Genetics, 2019, 95, 368-374.	2.0	18
16	Mass spectrometry-based assay for the molecular diagnosis of glioma: concomitant detection of chromosome 1p/19q codeletion, and IDH1, IDH2, and TERT mutation status. Oncotarget, 2017, 8, 57134-57148.	1.8	17
17	Familial gastrointestinal stromal tumors, lentigines, and cafÃ©â€auâ€lait macules associated with germline câ€kit mutation treated with imatinib. International Journal of Dermatology, 2017, 56, 195-201.	1.0	16
18	Molecular profiling of lung cancer specimens and liquid biopsies using MALDI-TOF mass spectrometry. Diagnostic Pathology, 2018, 13, 4.	2.0	16

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19	Clinical and Molecular Diagnosis of Beckwith-Wiedemann Syndrome with Single- or Multi-Locus Imprinting Disturbance. International Journal of Molecular Sciences, 2021, 22, 3445.	4.1	14
20	Novel physiological RECQL4 alternative transcript disclosed by molecular characterisation of Rothmundâ€“Thomson Syndrome sibs with mild phenotype. European Journal of Human Genetics, 2014, 22, 1298-1304.	2.8	13
21	A zebrafish model of Poikiloderma with Neutropenia recapitulates the human syndrome hallmarks and traces back neutropenia to the myeloid progenitor. Scientific Reports, 2015, 5, 15814.	3.3	13
22	Analysis of BRCA1 and RAD51C Promoter Methylation in Italian Families at High-Risk of Breast and Ovarian Cancer. Cancers, 2020, 12, 910.	3.7	13
23	Differential signature of the centrosomal MARK4 isoforms in glioma. Analytical Cellular Pathology, 2011, 34, 319-38.	1.4	13
24	A miRNome analysis of drug-free manic psychotic bipolar patients versus healthy controls. European Archives of Psychiatry and Clinical Neuroscience, 2020, 270, 893-900.	3.2	12
25	Profound alterations of the chromatin architecture at chromosome 11p15.5 in cells from Beckwith-Wiedemann and Silver-Russell syndromes patients. Scientific Reports, 2020, 10, 8275.	3.3	11
26	Suggestive evidence on the involvement of polypyrimidine-tract binding protein in regulating alternative splicing of MAP/microtubule affinity-regulating kinase 4 in glioma. Cancer Letters, 2015, 359, 87-96.	7.2	10
27	A novel splice site variant in ITPR1 gene underlying recessive Gillespie syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1427-1431.	1.2	9
28	(Epi)genetic profiling of extraembryonic and postnatal tissues from female monozygotic twins discordant for Beckwithâ€“Wiedemann syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1386.	1.2	9
29	A novel de novo DDX3X missense variant in a female with brachycephaly and intellectual disability: a case report. Italian Journal of Pediatrics, 2021, 47, 81.	2.6	9
30	Extensive Placental Methylation Profiling in Normal Pregnancies. International Journal of Molecular Sciences, 2021, 22, 2136.	4.1	8
31	Expanding the role of the splicing <scp>USB</scp>1 gene from Poikiloderma with Neutropenia to acquired myeloid neoplasms. British Journal of Haematology, 2015, 171, 557-565.	2.5	7
32	Revertant mosaicism for family mutations is not observed in BRCA1/2 phenocopies. PLoS ONE, 2017, 12, e0171663.	2.5	7
33	Preferential X Chromosome Inactivation as a Mechanism to Explain Female Preponderance in Myasthenia Gravis. Genes, 2022, 13, 696.	2.4	7
34	Forecasting the burden of COVID-19 hospitalized patients during the SARS-CoV-2 second wave in Lombardy, Italy. Panminerva Medica, 2021, 63, 86-87.	0.8	6
35	Sequence variants identification at the KCNQ1OT1:TSS differentially Methylated region in isolated omphalocele cases. BMC Medical Genetics, 2017, 18, 115.	2.1	3
36	Assessment of pregnancy dietary intake and association with maternal and neonatal outcomes. Pediatric Research, 2021, , .	2.3	3

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37	Cohesin Mutations Induce Chromatin Conformation Perturbation of the H19/IGF2 Imprinted Region and Gene Expression Dysregulation in Cornelia de Lange Syndrome Cell Lines. Biomolecules, 2021, 11, 1622.	4.0	3
38	Dysregulated ratio between MARK4 LÂand S isoforms during glioma progression. Cancer Genetics and Cytogenetics, 2010, 203, 45.	1.0	1
39	Serum Sphingosine-1-Phosphate as Novel Prognostic and Predictive Biomarker for COVID-19 Severity and Morbidity and Its Implications in Clinical Management. SSRN Electronic Journal, 0, , .	0.4	1
40	NGS sequencing proves as a powerful method to perform differential diagnosis in patients with inactivating PTH/PTHrP signaling disorders (iPPSD). Endocrine Abstracts, 0, , .	0.0	0
41	Hereditary : BRCA and Other. , 2020, , 23-41.		0