Elisa Frullanti

List of Publications by Year
in descending order

64 papers	2,394 citations	331670 21 h-index	233421 45 g-index
74 all docs	74 docs citations	74 times ranked	5956 citing authors

#	Article	IF	Citations
1	Genetic mechanisms of critical illness in COVID-19. Nature, 2021, 591, 92-98.	27.8	1,014
2	Association of Toll-like receptor 7 variants with life-threatening COVID-19 disease in males: findings from a nested case-control study. ELife, 2021, 10, .	6.0	145
3	Transcription Deregulation at the 15q25 Locus in Association with Lung Adenocarcinoma Risk. Clinical Cancer Research, 2009, 15, 1837-1842.	7.0	78
4	Imbalance of excitatory/inhibitory synaptic protein expression in iPSC-derived neurons from FOXG1+/â^' patients and in foxg1+/â^' mice. European Journal of Human Genetics, 2016, 24, 871-880.	2.8	54
5	Meta and pooled analyses of FGFR4 Gly388Arg polymorphism as a cancer prognostic factor. European Journal of Cancer Prevention, 2011, 20, 340-347.	1.3	52
6	Alport syndrome: impact of digenic inheritance in patients management. Clinical Genetics, 2017, 92, 34-44.	2.0	52
7	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. EBioMedicine, 2021, 65, 103246.	6.1	52
8	Association of lung adenocarcinoma clinical stage with gene expression pattern in noninvolved lung tissue. International Journal of Cancer, 2012, 131, E643-8.	5.1	49
9	Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1666-1676.	1.2	49
10	Rare variants in Toll-like receptor 7 results in functional impairment and downregulation of cytokine-mediated signaling in COVID-19 patients. Genes and Immunity, 2022, 23, 51-56.	4.1	41
11	Gene expression signature of non-involved lung tissue associated with survival in lung adenocarcinoma patients. Carcinogenesis, 2013, 34, 2767-2773.	2.8	40
12	FGFR4 Gly388Arg polymorphism may affect the clinical stage of patients with lung cancer by modulating the transcriptional profile of normal lung. International Journal of Cancer, 2009, 124, 2880-2885.	5.1	39
13	Telethon Network of Genetic Biobanks: a key service for diagnosis and research on rare diseases. Orphanet Journal of Rare Diseases, 2013, 8, 129.	2.7	39
14	Protective Role of a TMPRSS2 Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. Genes, 2021, 12, 596.	2.4	39
15	Promoter Polymorphisms and Transcript Levels of Nicotinic Receptor CHRNA5. Journal of the National Cancer Institute, 2010, 102, 1366-1370.	6.3	36
16	iPSC-derived neurons profiling reveals GABAergic circuit disruption and acetylated Î±-tubulin defect which improves after iHDAC6 treatment in Rett syndrome. Experimental Cell Research, 2018, 368, 225-235.	2.6	36
17	Employing a systematic approach to biobanking and analyzing clinical and genetic data for advancing COVID-19 research. European Journal of Human Genetics, 2021, 29, 745-759.	2.8	35
18	Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients. Lung Cancer, 2014, 85, 168-174.	2.0	30

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19	The polymorphism L412F in TLR3 inhibits autophagy and is a marker of severe COVID-19 in males. Autophagy, 2022, 18, 1662-1672.	9.1	25
20	Usefulness and Limitations of Comprehensive Characterization of mRNA Splicing Profiles in the Definition of the Clinical Relevance of BRCA1/2 Variants of Uncertain Significance. Cancers, 2019, 11, 295.	3.7	24
21	Analysis of the Phenotypes in the Rett Networked Database. International Journal of Genomics, 2019, 2019, 1-9.	1.6	23
22	MEIS2 gene is responsible for intellectual disability, cardiac defects and a distinct facial phenotype. European Journal of Medical Genetics, 2020, 63, 103627.	1.3	23
23	New frontiers to cure Alport syndrome: COL4A3 and COL4A5 gene editing in podocyte-lineage cells. European Journal of Human Genetics, 2020, 28, 480-490.	2.8	22
24	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. Human Genetics, 2022, 141, 147-173.	3.8	22
25	Exome sequencing coupled with mRNA analysis identifies NDUFAF6 as a Leigh gene. Molecular Genetics and Metabolism, 2016, 119, 214-222.	1.1	21
26	Omic Approach in Non-smoker Female with Lung Squamous Cell Carcinoma Pinpoints to Germline Susceptibility and Personalized Medicine. Cancer Research and Treatment, 2018, 50, 356-365.	3.0	20
27	Parent-of-origin effect of hypomorphic pathogenic variants and somatic mosaicism impact on phenotypic expression of retinoblastoma. European Journal of Human Genetics, 2018, 26, 1026-1037.	2.8	19
28	Cell-free DNA next-generation sequencing liquid biopsy as a new revolutionary approach for arteriovenous malformation. JVS Vascular Science, 2020, 1, 176-180.	1.1	17
29	Vinyl chloride exposure and cirrhosis: A systematic review and meta-analysis. Digestive and Liver Disease, 2012, 44, 775-779.	0.9	16
30	Germline polymorphisms and survival of lung adenocarcinoma patients: A genomeâ€wide study in two European patient series. International Journal of Cancer, 2015, 136, E262-71.	5.1	16
31	Urine-derived podocytes-lineage cells: A promising tool for precision medicine in Alport Syndrome. Human Mutation, 2018, 39, 302-314.	2.5	16
32	Severe COVID-19 in Hospitalized Carriers of Single CFTR Pathogenic Variants. Journal of Personalized Medicine, 2021, 11, 558.	2.5	16
33	Genome-wide association study in discordant sibships identifies multiple inherited susceptibility alleles linked to lung cancer. Carcinogenesis, 2010, 31, 462-465.	2.8	15
34	Mouse Pulmonary Adenoma Susceptibility 1 Locus Is an Expression QTL Modulating Kras-4A. PLoS Genetics, 2014, 10, e1004307.	3.5	15
35	A pilot study of next generation sequencingâ€“liquid biopsy on cell-free DNA as a novel non-invasive diagnostic tool for Klippelâ€“Trenaunay syndrome. Vascular, 2021, 29, 85-91.	0.9	14
36	A polygenic model with common variants may predict lung adenocarcinoma risk in humans. International Journal of Cancer, 2008, 123, 2327-2330.	5.1	13

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37	AAV-mediated FOXG1 gene editing in human Rett primary cells. European Journal of Human Genetics, 2020, 28, 1446-1458.	2.8	12
38	C9orf72 Intermediate Repeats Confer Genetic Risk for Severe COVID-19 Pneumonia Independently of Age. International Journal of Molecular Sciences, 2021, 22, 6991.	4.1	12
39	Multiple Genetic Loci Modulate Lung Adenocarcinoma Clinical Staging. Clinical Cancer Research, 2011, 17, 2410-2416.	7.0	11
40	SELP Asp603Asn and severe thrombosis in COVID-19 males. Journal of Hematology and Oncology, 2021, 14, 123.	17.0	11
41	Altered expression of neuropeptides in FoxG1-null heterozygous mutant mice. European Journal of Human Genetics, 2016, 24, 252-257.	2.8	10
42	High rate of HDR in gene editing of p.(Thr158Met) MECP2 mutational hotspot. European Journal of Human Genetics, 2020, 28, 1231-1242.	2.8	10
43	The phenomenon of multidrug resistance in glioblastomas. Hematology/ Oncology and Stem Cell Therapy, 2021, , .	0.9	10
44	Post-Mendelian Genetic Model in COVID-19. Cardiology and Cardiovascular Medicine, 2021, 05, .	0.2	10
45	A 5'-region polymorphism modulates promoter activity of the tumor suppressor gene MFSD2A. Molecular Cancer, 2011, 10, 81.	19.2	9
46	Twoâ€pointâ€NGS analysis of cancer genes in cellâ€free DNA of metastatic cancer patients. Cancer Medicine, 2020, 9, 2052-2061.	2.8	8
47	Personalized therapy in a GRIN1 mutated girl with intellectual disability and epilepsy. Clinical Dysmorphology, 2018, 27, 18-20.	0.3	7
48	MET somatic activating mutations are responsible for lymphovenous malformation and can be identified using cell-free DNA next generation sequencing liquid biopsy. Journal of Vascular Surgery: Venous and Lymphatic Disorders, 2021, 9, 740-744.	1.6	7
49	Potentially Treatable Disorder Diagnosed Post Mortem by Exome Analysis in a Boy with Respiratory Distress. International Journal of Molecular Sciences, 2016, 17, 306.	4.1	5
50	Germline mutations in lung cancer and personalized medicine. Familial Cancer, 2018, 17, 429-430.	1.9	5
51	Reply to the Letter to the Editor from Wang. Clinical Cancer Research, 2009, 15, 5599-5599.	7.0	4
52	Detection of Cryptic Mosaicism in X-linked Alport Syndrome Prompts to Re-evaluate Living-donor Kidney Transplantation. Transplantation, 2020, 104, 2360-2364.	1.0	4
53	A Genome Wide Copy Number Variations Analysis in Autism Spectrum Disorder (Asd) and Intellectual Disability (Id) in Italian Families. Journal of Genetic Syndromes & Gene Therapy, 2016, 7, .	0.2	3
54	Authorsâ€™ reply: Comment to â€œVinyl chloride exposure and cirrhosis: A systematic review and meta-analysisâ€• Digestive and Liver Disease, 2013, 45, 702.	0.9	2

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55	Lowâ€level <scp>TP</scp>53 mutational load antecedes clonal expansion in chronic lymphocytic leukaemia. British Journal of Haematology, 2019, 184, 657-659.	2.5	2
56	Shorter Androgen Receptor PolyQ Alleles Protect Against Life-Threatening COVID-19 Disease in Males. SSRN Electronic Journal, 0, , .	0.4	2
57	PIK3CA-CDKN2A clonal evolution in metastatic breast cancer and multiple points cell-free DNA analysis. Cancer Cell International, 2019, 19, 274.	4.1	1
58	Related expression of TRKA and P75 receptors and the changing copy number ofMYC-oncogenes determine the sensitivity of brain tumor cells to the treatment of the nerve growth factor in combination with cisplatin and temozolomide. Drug Metabolism and Personalized Therapy, 2021, .	0.6	1
59	Commentary: Potential Links between Hepadnavirus and Bornavirus Sequences in the Host Genome and Cancer. Frontiers in Microbiology, 2018, 9, 1649.	3.5	0
60	Liquid Biopsies. , 2020, , 495-500.		0
61	Specific clonal expansion at disease progression (PD) in solid cancers pinpointed by cell free DNA analysis.. Journal of Clinical Oncology, 2019, 37, e13144-e13144.	1.6	0
62	Noninvasive Genetic Testing: Adhesive Patch-Based Skin Biopsy and Buccal Swab. , 2020, , 491-494.		0
63	Private somatic mutations identified with liquid biopsy lead tumor progression in solid cancers. Journal of Cancer Metastasis and Treatment, 0, 2020, .	0.8	0
64	Related expression of TRKA and P75 receptors and the changing copy number ofMYC-oncogenes determine the sensitivity of brain tumor cells to the treatment of the nerve growth factor in combination with cisplatin and temozolomide. Drug Metabolism and Drug Interactions, 2020, 35, .	0.3	0