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	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. Human Genetics, 2022, 141, 147-173.	3.8	22
2	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
3	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
4	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
5	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
6	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
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 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
9	Protective Role of a TMPRSS2 Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. Genes, 2021, 12, 596.	2.4	39
10	The phenomenon of multidrug resistance in glioblastomas. Hematology/ Oncology and Stem Cell Therapy, 2021, , .	0.9	10
11	Severe COVID-19 in Hospitalized Carriers of Single CFTR Pathogenic Variants. Journal of Personalized Medicine, 2021, 11, 558.	2.5	16
12	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
13	SELP Asp603Asn and severe thrombosis in COVID-19 males. Journal of Hematology and Oncology, 2021, 14, 123.	17.0	11
14	Genetic mechanisms of critical illness in COVID-19. Nature, 2021, 591, 92-98.	27.8	1,014
15	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
16	Post-Mendelian Genetic Model in COVID-19. Cardiology and Cardiovascular Medicine, 2021, 05, .	0.2	10
17	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
18	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

#	Article	IF	Citations
19	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
20	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
21	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
22	AAV-mediated FOXG1 gene editing in human Rett primary cells. European Journal of Human Genetics, 2020, 28, 1446-1458.	2.8	12
23	Liquid Biopsies. , 2020, , 495-500.		0
24	Twoâ€pointâ€NGS analysis of cancer genes in cellâ€free DNA of metastatic cancer patients. Cancer Medicine, 2020, 9, 2052-2061.	2.8	8
25	Noninvasive Genetic Testing: Adhesive Patch-Based Skin Biopsy and Buccal Swab. , 2020, , 491-494.		0
26	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
27	PIK3CA-CDKN2A clonal evolution in metastatic breast cancer and multiple points cell-free DNA analysis. Cancer Cell International, 2019, 19, 274.	4.1	1
28	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
29	Analysis of the Phenotypes in the Rett Networked Database. International Journal of Genomics, 2019, 2019, 1-9.	1.6	23
30	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
31	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
32	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
33	Germline mutations in lung cancer and personalized medicine. Familial Cancer, 2018, 17, 429-430.	1.9	5
34	Personalized therapy in a GRIN1 mutated girl with intellectual disability and epilepsy. Clinical Dysmorphology, 2018, 27, 18-20.	0.3	7
35	Urine-derived podocytes-lineage cells: A promising tool for precision medicine in Alport Syndrome. Human Mutation, 2018, 39, 302-314.	2.5	16
36	Commentary: Potential Links between Hepadnavirus and Bornavirus Sequences in the Host Genome and Cancer. Frontiers in Microbiology, 2018, 9, 1649.	3.5	0

#	Article	IF	Citations
37	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
38	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
39	Alport syndrome: impact of digenic inheritance in patients management. Clinical Genetics, 2017, 92, 34-44.	2.0	52
40	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
41	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
42	Exome sequencing coupled with mRNA analysis identifies NDUFAF6 as a Leigh gene. Molecular Genetics and Metabolism, 2016, 119, 214-222.	1.1	21
43	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
44	Altered expression of neuropeptides in FoxG1-null heterozygous mutant mice. European Journal of Human Genetics, 2016, 24, 252-257.	2.8	10
45	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
46	Mouse Pulmonary Adenoma Susceptibility 1 Locus Is an Expression QTL Modulating Kras-4A. PLoS Genetics, 2014, 10, e1004307.	3.5	15
47	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
48	Oligogenic germline mutations identified in early non-smokers lung adenocarcinoma patients. Lung Cancer, 2014, 85, 168-174.	2.0	30
49	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
50	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
51	Gene expression signature of non-involved lung tissue associated with survival in lung adenocarcinoma patients. Carcinogenesis, 2013, 34, 2767-2773.	2.8	40
52	Vinyl chloride exposure and cirrhosis: A systematic review and meta-analysis. Digestive and Liver Disease, 2012, 44, 775-779.	0.9	16
53	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
54	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

#	Article	IF	Citations
55	A 5'-region polymorphism modulates promoter activity of the tumor suppressor gene MFSD2A. Molecular Cancer, 2011, 10, 81.	19.2	9
56	Multiple Genetic Loci Modulate Lung Adenocarcinoma Clinical Staging. Clinical Cancer Research, 2011, 17, 2410-2416.	7.0	11
57	Genome-wide association study in discordant sibships identifies multiple inherited susceptibility alleles linked to lung cancer. Carcinogenesis, 2010, 31, 462-465.	2.8	15
58	Promoter Polymorphisms and Transcript Levels of Nicotinic Receptor CHRNA5. Journal of the National Cancer Institute, 2010, 102, 1366-1370.	6.3	36
59	Transcription Deregulation at the 15q25 Locus in Association with Lung Adenocarcinoma Risk. Clinical Cancer Research, 2009, 15, 1837-1842.	7.0	78
60	Reply to the Letter to the Editor from Wang. Clinical Cancer Research, 2009, 15, 5599-5599.	7.0	4
61	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
62	A polygenic model with common variants may predict lung adenocarcinoma risk in humans. International Journal of Cancer, 2008, 123, 2327-2330.	5.1	13
63	Shorter Androgen Receptor PolyQ Alleles Protect Against Life-Threatening COVID-19 Disease in Males. SSRN Electronic Journal, 0, , .	0.4	2
64	Private somatic mutations identified with liquid biopsy lead tumor progression in solid cancers. Journal of Cancer Metastasis and Treatment, 0, 2020, .	0.8	0