Carmen SÃ-lvia Bertuzzo

List of Publications by Year
in descending order

44 papers	775 citations	471509 17 h-index	552781 26 g-index
48 all docs	48 docs citations	48 times ranked	1140 citing authors

#	Article	IF	Citations
1	Classification of CFTR mutation classes. Lancet Respiratory Medicine,the, 2016, 4, e37-e38.	10.7	115
2	Epidemiological and genetic characteristics associated with the severity of acute viral bronchiolitis by respiratory syncytial virus. Jornal De Pediatria, 2013, 89, 531-543.	2.0	55
3	Measurements of CFTR-Mediated Clâ˜ Secretion in Human Rectal Biopsies Constitute a Robust Biomarker for Cystic Fibrosis Diagnosis and Prognosis. PLoS ONE, 2012, 7, e47708.	2.5	52
4	Asthma: Gln27Glu and Arg16Gly polymorphisms of the beta2-adrenergic receptor gene as risk factors. Allergy, Asthma and Clinical Immunology, 2014, 10, 8.	2.0	38
5	CFTR genotype and clinical outcomes of adult patients carried as cystic fibrosis disease. Gene, 2014, 540, 183-190.	2.2	37
6	Association of clinical severity of cystic fibrosis with variants in the SLC gene family (SLC6A14,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 54	2.2	34
7	Novel, rare and common pathogenic variants in the CFTR gene screened by high-throughput sequencing technology and predicted by in silico tools. Scientific Reports, 2019, 9, 6234.	3.3	33
8	Association of TGF-β1, CD14, IL-4, IL-4R and ADAM33 gene polymorphisms with asthma severity in children and adolescents. Jornal De Pediatria, 2008, 84, 203-210.	2.0	33
9	Polymorphisms in the glutathione pathway modulate cystic fibrosis severity: a cross-sectional study. BMC Medical Genetics, 2014, 15, 27.	2.1	28
10	Extent of rescue of F508del-CFTR function by VX-809 and VX-770 in human nasal epithelial cells correlates with SNP rs7512462 in SLC26A9 gene in F508del/F508del Cystic Fibrosis patients. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 1323-1331.	3.8	28
11	Personalized Drug Therapy in Cystic Fibrosis: From Fiction to Reality. Current Drug Targets, 2015, 16, 1007-1017.	2.1	25
12	AssociaÃ§Ã£o entre os polimorfismos dos genes MBL2, TGF-Î²1 e CD14 com a gravidade da doenÃ§a pulmonar na fibrose cÃstica. Jornal Brasileiro De Pneumologia, 2009, 35, 334-342.	0.7	24
13	Genetic interaction of GSH metabolic pathway genes in cystic fibrosis. BMC Medical Genetics, 2013, 14, 60.	2.1	24
14	IL8 gene as modifier of cystic fibrosis: unraveling the factors which influence clinical variability. Human Genetics, 2016, 135, 881-894.	3.8	22
15	Chloride and sodium ion concentrations in saliva and sweat as a method to diagnose cystic fibrosis. Jornal De Pediatria, 2019, 95, 443-450.	2.0	22
16	MutaÃ§Ãµes do gene cystic fibrosis transmembrane conductance regulator e deleÃ§Ãµes dos genes glutationa S-transferase em pacientes com fibrose cÃstica no Brasil. Jornal Brasileiro De Pneumologia, 2012, 38, 50-56.	0.7	20
17	Cystic Fibrosis in Adults. Lung, 2007, 185, 81-87.	3.3	19
18	Association between the IVS4Gâ€‰>â€‰T mutation in the TCF7L2 gene and susceptibility to diabetes in cystic fibrosis patients. BMC Research Notes, 2012, 5, 561.	1.4	16

#	Article	IF	Citations
19	APC germline mutations in families with familial adenomatous polyposis. Oncology Reports, 2013, 30, 2081-2088.	2.6	16
20	Thymidylate synthase gene (TYMS) polymorphisms in sporadic and hereditary breast cancer. BMC Research Notes, 2012, 5, 676.	1.4	15
21	Screening for F508del as a first step in the molecular diagnosis of cystic fibrosis. Jornal Brasileiro De Pneumologia, 2013, 39, 306-316.	0.7	11
22	Hypertonic Saline as a Useful Tool for Sputum Induction and Pathogen Detection in Cystic Fibrosis. Lung, 2017, 195, 431-439.	3.3	9
23	Determining mutations in G6PC and SLC37A4 genes in a sample of Brazilian patients with glycogen storage disease types Ia and Ib. Genetics and Molecular Biology, 2013, 36, 502-506.	1.3	8
24	SLC23A2-05 (rs4987219) and KRAS-LCS6 (rs61764370) polymorphisms in patients with squamous cell carcinoma of the head and neck. Oncology Letters, 2014, 7, 1803-1811.	1.8	8
25	Frequency of 677C -> T and 1298A -> C polymorphisms in the 5,10-methylenetetrahydrofolate reductase (MTHFR) gene in Turner syndrome individuals. Genetics and Molecular Biology, 2006, 29, 41-44.	1.3	8
26	AvaliaÃ§Ã£o da concentraÃ§Ã£o de alfa 1-antitripsina e da presenÃ§a dos alelos S e Z em uma populaÃ§Ã£o de indivÃduos sintomÃ¡ticos respiratÃ³rios crÃ´nicos. Jornal Brasileiro De Pneumologia, 2008, 34, 1019-1025.	0.7	8
27	Nasal Potential Difference in Cystic Fibrosis considering SevereCFTRMutations. Disease Markers, 2015, 2015, 1-11.	1.3	7
28	Variants of estrogen receptor alpha and beta genes modify the severity of sporadic breast cancer. Gene, 2017, 608, 73-78.	2.2	7
29	Variants in the interleukin 8 gene and the response to inhaled bronchodilators in cystic fibrosis. Jornal De Pediatria, 2017, 93, 639-648.	2.0	7
30	Cystic fibrosis transmembrane conductance regulator mutations at a referral center for cystic fibrosis. Jornal Brasileiro De Pneumologia, 2013, 39, 555-561.	0.7	6
31	Pancreatic Insufficiency in Cystic Fibrosis. Pancreas, 2018, 47, 99-109.	1.1	5
32	Quality of sweat test (ST) based on the proportion of sweat sodium (Na) and sweat chloride (Cl) as diagnostic parameter of cystic fibrosis: are we on the right way?. Diagnostic Pathology, 2016, 11, 103.	2.0	4
33	Single nucleotide variants c.-13GÂâ†'ÂC (rs17429833) and c.108CÂâ†'ÂT (rs72466472) in the CLDN1 gene and increased risk for familial colorectal cancer. Gene, 2021, 768, 145304.	2.2	4
34	Prevalence of ΔF508 mutation in the cystic fibrosis transmembrane conductance regulator gene among cystic fibrosis patients from a Brazilian referral center. Jornal De Pediatria, 2012, 88, 531-4.	2.0	4
35	Burkholderia cepacia complex in cystic fibrosis in a Brazilian reference center. Medical Microbiology and Immunology, 2017, 206, 447-461.	4.8	3
36	A negative screening of rare genetic variants in the ADIPOQ and STATH genes in cystic fibrosis. Pulmonology, 2020, 26, 138-144.	2.1	2

#	Article	IF	Citations
37	Impact of CFTR large deletions and insertions on the clinical and laboratory severity of cystic fibrosis: a serial case report. Pulmonology, 2022, 28, 235-238.	2.1	2
38	Radiation from mammography: Diagnostic or cancer induction?. , 2011, , .		1
39	Preimplantation genetic diagnosis for cystic fibrosis: a case report. Einstein (Sao Paulo, Brazil), 2015, 13, 110-113.	0.7	1
40	Cystic fibrosis transmembrane regulator haplotypes in households of patients with cystic fibrosis. Gene, 2018, 641, 137-143.	2.2	1
41	ABCB1 variants (C1236T, rs1128503 and G2677T/A, rs2032582) do not show an association with recurrence and survival in patients with breast cancer undergoing anthracycline-based chemotherapy. Meta Gene, 2019, 21, 100596.	0.6	1
42	Chloride and sodium ion concentrations in saliva and sweat as a method to diagnose cystic fibrosis. Jornal De Pediatria (VersÃ£o Em PortuguÃªs), 2019, 95, 443-450.	0.2	1
43	Molecular screening of CFTR gene in Brazilian men with bilateral agenesis of the vas deferens. Human Fertility, 2006, 9, 53-56.	1.7	0
44	Association between the IVS4G > T mutation in the TCF7L2 gene and susceptibility to diabetes in cystic fibrosis patients. BMC Research Notes, 2012, 5, 2101791285670503.	1.4	0