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	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
2	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
3	A negative screening of rare genetic variants in the ADIPOQ and STATH genes in cystic fibrosis. Pulmonology, 2020, 26, 138-144.	2.1	2
4	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
5	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
6	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
7	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
8	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
9	Cystic fibrosis transmembrane regulator haplotypes in households of patients with cystic fibrosis. Gene, 2018, 641, 137-143.	2.2	1
10	Pancreatic Insufficiency in Cystic Fibrosis. Pancreas, 2018, 47, 99-109.	1.1	5
11	Variants of estrogen receptor alpha and beta genes modify the severity of sporadic breast cancer. Gene, 2017, 608, 73-78.	2.2	7
12	Hypertonic Saline as a Useful Tool for Sputum Induction and Pathogen Detection in Cystic Fibrosis. Lung, 2017, 195, 431-439.	3.3	9
13	Burkholderia cepacia complex in cystic fibrosis in a Brazilian reference center. Medical Microbiology and Immunology, 2017, 206, 447-461.	4.8	3
14	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
15	Association of clinical severity of cystic fibrosis with variants in the SLC gene family (SLC6A14,) Tj ETQq1 1 0.784314 rgBT /Overlock	2.2	34
16	IL8 gene as modifier of cystic fibrosis: unraveling the factors which influence clinical variability. Human Genetics, 2016, 135, 881-894.	3.8	22
17	Classification of CFTR mutation classes. Lancet Respiratory Medicine,the, 2016, 4, e37-e38.	10.7	115
18	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

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19	Nasal Potential Difference in Cystic Fibrosis considering SevereCFTRMutations. Disease Markers, 2015, 2015, 1-11.	1.3	7
20	Preimplantation genetic diagnosis for cystic fibrosis: a case report. Einstein (Sao Paulo, Brazil), 2015, 13, 110-113.	0.7	1
21	Personalized Drug Therapy in Cystic Fibrosis: From Fiction to Reality. Current Drug Targets, 2015, 16, 1007-1017.	2.1	25
22	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
23	CFTR genotype and clinical outcomes of adult patients carried as cystic fibrosis disease. Gene, 2014, 540, 183-190.	2.2	37
24	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
25	Polymorphisms in the glutathione pathway modulate cystic fibrosis severity: a cross-sectional study. BMC Medical Genetics, 2014, 15, 27.	2.1	28
26	Genetic interaction of GSH metabolic pathway genes in cystic fibrosis. BMC Medical Genetics, 2013, 14, 60.	2.1	24
27	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
28	APC germline mutations in families with familial adenomatous polyposis. Oncology Reports, 2013, 30, 2081-2088.	2.6	16
29	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
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	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
32	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
33	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
34	Thymidylate synthase gene (TYMS) polymorphisms in sporadic and hereditary breast cancer. BMC Research Notes, 2012, 5, 676.	1.4	15
35	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
36	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

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37	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
38	Radiation from mammography: Diagnostic or cancer induction?. , 2011, , .		1
39	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
40	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
41	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
42	Cystic Fibrosis in Adults. Lung, 2007, 185, 81-87.	3.3	19
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	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