Karen R Siklosi

List of Publications by Year
in descending order

40	2,178	430754 18	345118 36
papers	citations	h-index	g-index

41	41	41	2942
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Comparing encounter-based and annualized chronic pseudomonas infection definitions in cystic fibrosis. Journal of Cystic Fibrosis, 2022, 21, 40-44.	0.3	3
2	Complete CFTR gene sequencing in 5,058 individuals with cystic fibrosis informs variant-specific treatment. Journal of Cystic Fibrosis, 2022, 21, 463-470.	0.3	13
3	Accurate assignment of disease liability to genetic variants using only population data. Genetics in Medicine, 2022, 24, 87-99.	1.1	4
4	Genetic counseling access for parents of newborns who screen positive for cystic fibrosis: Consensus guidelines. Pediatric Pulmonology, 2022, 57, 894-902.	1.0	6
5	CFTRvariants are associated with chronic bronchitis in smokers. European Respiratory Journal, 2022, 60, 2101994.	3.1	6
6	Things come in threes: A new complex allele and a novel deletion within the CFTR gene complicate an accurate diagnosis of cystic fibrosis. Molecular Genetics & Genomic Medicine, 2022, 10, e1926.	0.6	2
7	CFTR bearing variant p.Phe312del exhibits function inconsistent with phenotype and negligible response to ivacaftor. JCI Insight, 2022, 7, .	2.3	3
8	DNA sequencing analysis of cystic fibrosis transmembrane conductance regulator gene identifies cystic fibrosisâ€associated variants in the Severe Asthma Research Program. Pediatric Pulmonology, 2022, 57, 1782-1788.	1.0	3
9	Caution advised in the use of CFTR modulator treatment for individuals harboring specific CFTR variants. Journal of Cystic Fibrosis, 2022, 21, 856-860.	0.3	8
10	The future of cystic fibrosis care: a global perspective. Lancet Respiratory Medicine,the, 2020, 8, 65-124.	5.2	573
11	Genetic Modifiers of Cystic Fibrosis-Related Diabetes Have Extensive Overlap With Type 2 Diabetes and Related Traits. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 1401-1415.	1.8	34
12	Predictive effects of low birth weight and small for gestational age status on respiratory and nutritional outcomes in cystic fibrosis. Journal of Cystic Fibrosis, 2020, 19, 888-895.	0.3	2
13	Cystic fibrosis transmembrane conductance regulator function, not TAS2R38 gene haplotypes, predict sinus surgery in children and young adults with cystic fibrosis. International Forum of Allergy and Rhinology, 2020, 10, 748-754.	1.5	7
14	Evaluation of both exonic and intronic variants for effects on RNA splicing allows for accurate assessment of the effectiveness of precision therapies. PLoS Genetics, 2020, 16, e1009100.	1.5	23
15	Title is missing!. , 2020, 16, e1009100.		0
16	Title is missing!. , 2020, 16, e1009100.		0
17	Title is missing!. , 2020, 16, e1009100.		0
18	Title is missing!. , 2020, 16, e1009100.		0

#	Article	IF	Citations
19	Practice variation of genetic counselor engagement in the cystic fibrosis newborn screenâ€positive diagnostic resolution process. Journal of Genetic Counseling, 2019, 28, 1178-1188.	0.9	8
20	Correlating Cystic Fibrosis Transmembrane Conductance Regulator Function with Clinical Features to Inform Precision Treatment of Cystic Fibrosis. American Journal of Respiratory and Critical Care Medicine, 2019, 199, 1116-1126.	2.5	76
21	The increasing challenge of genetic counseling for cystic fibrosis. Journal of Cystic Fibrosis, 2019, 18, 167-174.	0.3	17
22	Capitalizing on the heterogeneous effects of CFTR nonsense and frameshift variants to inform therapeutic strategy for cystic fibrosis. PLoS Genetics, 2018, 14, e1007723.	1.5	44
23	Functional Assays Are Essential for Interpretation of Missense Variants Associated with Variable Expressivity. American Journal of Human Genetics, 2018, 102, 1062-1077.	2.6	69
24	Residual function of cystic fibrosis mutants predicts response to small molecule CFTR modulators. JCI Insight, 2018, 3, .	2.3	86
25	Applying Cystic Fibrosis Transmembrane Conductance Regulator Genetics and CFTR2 Data to Facilitate Diagnoses. Journal of Pediatrics, 2017, 181, S27-S32.e1.	0.9	58
26	Ethnicity impacts the cystic fibrosis diagnosis: A note of caution. Journal of Cystic Fibrosis, 2017, 16, 488-491.	0.3	34
27	Diagnosis and Treatment of Cystic Fibrosis: A (Not-so) Simple Recessive Condition. Current Genetic Medicine Reports, 2017, 5, 91-99.	1.9	1
28	Systematic Computational Identification of Variants That Activate Exonic and Intronic Cryptic Splice Sites. American Journal of Human Genetics, 2017, 100, 751-765.	2.6	68
29	Benign and Deleterious Cystic Fibrosis Transmembrane Conductance Regulator Mutations Identified by Sequencing in Positive Cystic Fibrosis Newborn Screen Children from California. PLoS ONE, 2016, 11, e0155624.	1.1	27
30	Deep resequencing of CFTR in 762 F508del homozygotes reveals clusters of non-coding variants associated with cystic fibrosis disease traits. Human Genome Variation, 2016, 3, 16038.	0.4	34
31	Respiratory pathogens mediate the association between lung function and temperature in cystic fibrosis. Journal of Cystic Fibrosis, 2016, 15, 794-801.	0.3	14
32	Molecular Genetics of Cystic Fibrosis Transmembrane Conductance Regulator. Pediatric Clinics of North America, 2016, 63, 585-598.	0.9	38
33	Bias in CFTR screening panels. Genetics in Medicine, 2016, 18, 209-209.	1.1	3
34	Loss of carbonic anhydrase XII function in individuals with elevated sweat chloride concentration and pulmonary airway disease. Human Molecular Genetics, 2016, 25, 1923-1933.	1.4	32
35	Benign outcome among positive cystic fibrosis newborn screen children with non-CF-causing variants. Journal of Cystic Fibrosis, 2015, 14, 714-719.	0.3	35
36	Missense variants in CFTR nucleotide-binding domains predict quantitative phenotypes associated with cystic fibrosis disease severity. Human Molecular Genetics, 2015, 24, 1908-1917.	1.4	11

#	Article	IF	Citations
37	Genome-wide association meta-analysis identifies five modifier loci of lung disease severity in cystic fibrosis. Nature Communications, 2015, 6, 8382.	5.8	242
38	Experimental Assessment of Splicing Variants Using Expression Minigenes and Comparison with In Silico Predictions. Human Mutation, 2014, 35, 1249-1259.	1.1	56
39	Defining the disease liability of variants in the cystic fibrosis transmembrane conductance regulator gene. Nature Genetics, 2013, 45, 1160-1167.	9.4	513
40	Development, validation, and implementation of a questionnaire assessing disease knowledge and understanding in adult cystic fibrosis patients. Journal of Cystic Fibrosis, 2010, 9, 400-405.	0.3	24