Alejandro Sifrim

List of Publications by Year in descending order

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1478505 1872680 6 779 6 6 citations h-index g-index papers 12 12 12 2667 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	Homozygous <i>GDF2</i> nonsense mutations result in a loss of circulating BMP9 and BMP10 and are associated with either PAH or an "HHTâ€likeâ€syndrome in children. Molecular Genetics & amp; Genomic Medicine, 2021, 9, e1685.	1.2	19
2	A sensitive and affordable multiplex RT-qPCR assay for SARS-CoV-2 detection. PLoS Biology, 2020, 18, e3001030.	5.6	32
3	PolyQ Tract Toxicity in SCA1 is Length Dependent in the Absence of CAG Repeat Interruption. Frontiers in Cellular Neuroscience, 2018, 12, 200.	3.7	18
4	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	2.4	46
5	Long QT syndrome and left ventricular noncompaction in 4 family members across 2 generations with KCNQ1 mutation. European Journal of Medical Genetics, 2017, 60, 233-238.	1.3	13
6	Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. Lancet, The, 2015, 385, 1305-1314.	13.7	651