

# Alejandro Sifrim

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2063914/publications.pdf>

Version: 2024-02-01

6  
papers

779  
citations

1478505

6  
h-index

1872680

6  
g-index

12  
all docs

12  
docs citations

12  
times ranked

2667  
citing authors

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