Avinash V Dharmadhikari

List of Publications by Year
in descending order

22 papers	1,576 citations	516710 16 h-index	794594 19 g-index
25 all docs	25 docs citations	25 times ranked	3152 citing authors

#	Article	IF	Citations
1	Whole Exome Sequencing detects PYGM variants in two adults with McArdle disease. Journal of Physical Education and Sports Management, 2022, , mcs.a006173.	1.2	1
2	Casual Genetic Variants in Stillbirth. Obstetrical and Gynecological Survey, 2021, 76, 79-81.	0.4	0
3	1007 Umbilical cord segment collection allows for comprehensive genetic diagnostic testing at delivery. American Journal of Obstetrics and Gynecology, 2021, 224, S624.	1.3	0
4	13. Revisiting centromeric polymorphisms: Implications for POC and prenatal interphase FISH studies. Cancer Genetics, 2021, 252-253, S5.	0.4	0
5	Compound heterozygous inheritance of two novel COQ2 variants results in familial coenzyme Q deficiency. Orphanet Journal of Rare Diseases, 2020, 15, 320.	2.7	8
6	Causal Genetic Variants in Stillbirth. New England Journal of Medicine, 2020, 383, 1107-1116.	27.0	67
7	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. Genome Medicine, 2019, 11, 30.	8.2	42
8	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	6.2	59
9	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. Genome Medicine, 2018, 10, 74.	8.2	105
10	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
11	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. Human Genetics, 2016, 135, 569-586.	3.8	85
12	Lethal lung hypoplasia and vascular defects in mice with conditionalFoxf1overexpression. Biology Open, 2016, 5, 1595-1606.	1.2	20
13	Genomic and Epigenetic Complexity of the FOXF1 Locus in 16q24.1: Implications for Development and Disease. Current Genomics, 2015, 16, 107-116.	1.6	51
14	Molecular and clinical analyses of 16q24.1 duplications involving FOXF1 identify an evolutionarily unstable large minisatellite. BMC Medical Genetics, 2014, 15, 128.	2.1	11
15	Two deletions overlapping a distant FOXF1 enhancer unravel the role of lncRNA LINC01081 in etiology of alveolar capillary dysplasia with misalignment of pulmonary veins. American Journal of Medical Genetics, Part A, 2014, 164, 2013-2019.	1.2	46
16	Comparative Analyses of Lung Transcriptomes in Patients with Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins and in Foxf1 Heterozygous Knockout Mice. PLoS ONE, 2014, 9, e94390.	2.5	31
17	CD19 expression in acute leukemia is not restricted to the cytogenetically aberrant populations. Leukemia and Lymphoma, 2013, 54, 1517-1520.	1.3	19
18	Small noncoding differentially methylated copy-number variants, including lncRNA genes, cause a lethal lung developmental disorder. Genome Research, 2013, 23, 23-33.	5.5	127

#	Article	IF	Citations
19	NovelFOXF1Mutations in Sporadic and Familial Cases of Alveolar Capillary Dysplasia with Misaligned Pulmonary Veins Imply a Role for its DNA Binding Domain. Human Mutation, 2013, 34, 801-811.	2.5	97
20	Small rare recurrent deletions and reciprocal duplications in 2q21.1, including brain-specific ARHGEF4 and GPR148. Human Molecular Genetics, 2012, 21, 3345-3355.	2.9	22
21	Recurrent deletions and reciprocal duplications of 10q11.21q11.23 including CHAT and SLC18A3 are likely mediated by complex low-copy repeats. Human Mutation, 2012, 33, 165-179.	2.5	45
22	Chromosome Catastrophes Involve Replication Mechanisms Generating Complex Genomic Rearrangements. Cell, 2011, 146, 889-903.	28.9	391