Ascia Eskin

List of Publications by Year
in descending order

21 papers	866 citations	623734 14 h-index	794594 19 g-index
22 all docs	22 docs citations	22 times ranked	2089 citing authors

#	Article	IF	Citations
1	Transcriptional regulation of macrophage cholesterol efflux and atherogenesis by a long noncoding RNA. Nature Medicine, 2018, 24, 304-312.	30.7	171
2	Diagnostic utility of transcriptome sequencing for rare Mendelian diseases. Genetics in Medicine, 2020, 22, 490-499.	2.4	136
3	EGFR Mutation-Induced Alternative Splicing of Max Contributes to Growth of Glycolytic Tumors in Brain Cancer. Cell Metabolism, 2013, 17, 1000-1008.	16.2	130
4	DMD genotype correlations from the Duchenne Registry: Endogenous exon skipping is a factor in prolonged ambulation for individuals with a defined mutation subtype. Human Mutation, 2018, 39, 1193-1202.	2.5	65
5	Metabolic characterization of isocitrate dehydrogenase (IDH) mutant and IDH wildtype gliomaspheres uncovers cell type-specific vulnerabilities. Cancer & Metabolism, 2018, 6, 4.	5.0	55
6	A homozygous loss-of-function CAMK2A mutation causes growth delay, frequent seizures and severe intellectual disability. ELife, 2018, 7, .	6.0	53
7	Sex-Specific Effects of Testosterone on the Sexually Dimorphic Transcriptome and Epigenome of Embryonic Neural Stem/Progenitor Cells. Scientific Reports, 2016, 6, 36916.	3.3	41
8	A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. Cilia, 2016, 5, 8.	1.8	37
9	Loss of the scavenger mRNA decapping enzyme DCPS causes syndromic intellectual disability with neuromuscular defects. Human Molecular Genetics, 2015, 24, 3163-3171.	2.9	31
10	Failure to up-regulate transcription of genes necessary for muscle adaptation underlies limb girdle muscular dystrophy 2A (calpainopathy). Human Molecular Genetics, 2016, 25, 2194-2207.	2.9	25
11	Online Self-Report Data for Duchenne Muscular Dystrophy Confirms Natural History and Can Be Used to Assess for Therapeutic Benefits. PLOS Currents, 2014, 6, .	1.4	25
12	Ethanol-induced differential gene expression and acetyl-CoA metabolism in a longevity model of the nematode Caenorhabditis elegans. Experimental Gerontology, 2015, 61, 20-30.	2.8	21
13	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. American Journal of Human Genetics, 2018, 103, 948-967.	6.2	18
14	Genomic predictors of remission to antidepressant treatment in geriatric depression using genomeâ€wide expression analyses: a pilot study. International Journal of Geriatric Psychiatry, 2016, 31, 510-517.	2.7	17
15	Identification of novel candidate genes for 46,XY disorders of sex development (DSD) using a C57BL/6J-Y POS mouse model. Biology of Sex Differences, 2018, 9, 8.	4.1	14
16	Mitchell-Riley syndrome iPSC exhibit reduced pancreatic endoderm differentiation due to an RFX6 mutation. Development (Cambridge), 2020, 147, .	2.5	10
17	Dusp6 is a genetic modifier of growth through enhanced ERK activity. Human Molecular Genetics, 2018, 28, 279-289.	2.9	6
18	Recessive ciliopathy mutations in primary endocardial fibroelastosis: a rare neonatal cardiomyopathy in a case of Alstrom syndrome. Journal of Molecular Medicine, 2021, 99, 1623-1638.	3.9	4

#	Article	IF	Citations
19	Mutation in TWINKLE in a Large Iranian Family with Progressive External Ophthalmoplegia, Myopathy, Dysphagia and Dysphonia, and Behavior Change. Archives of Iranian Medicine, 2016, 19, 87-91.	0.6	4
20	Leveraging ancestry to improve causal variant identification in exome sequencing for monogenic disorders. European Journal of Human Genetics, 2016, 24, 113-119.	2.8	3
21	Role of Mir-34 Upregulation in Disruption of c-Myc, c-Myb and NOTCH Signaling in Diamond-Blackfan Anemia. Blood, 2016, 128, 3895-3895.	1.4	0