

# Ascia Eskin

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/4299132/publications.pdf>

Version: 2024-02-01

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	Recessive ciliopathy mutations in primary endocardial fibroelastosis: a rare neonatal cardiomyopathy in a case of Alstrom syndrome. <i>Journal of Molecular Medicine</i> , 2021, 99, 1623-1638.	3.9	4
2	Diagnostic utility of transcriptome sequencing for rare Mendelian diseases. <i>Genetics in Medicine</i> , 2020, 22, 490-499.	2.4	136
3	Mitchell-Riley syndrome iPSC exhibit reduced pancreatic endoderm differentiation due to an <i>RFX6</i> mutation. <i>Development (Cambridge)</i> , 2020, 147, .	2.5	10
4	Transcriptional regulation of macrophage cholesterol efflux and atherogenesis by a long noncoding RNA. <i>Nature Medicine</i> , 2018, 24, 304-312.	30.7	171
5	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2018, 103, 948-967.	6.2	18
6	Dusp6 is a genetic modifier of growth through enhanced ERK activity. <i>Human Molecular Genetics</i> , 2018, 28, 279-289.	2.9	6
7	A homozygous loss-of-function CAMK2A mutation causes growth delay, frequent seizures and severe intellectual disability. <i>ELife</i> , 2018, 7, .	6.0	53
8	Metabolic characterization of isocitrate dehydrogenase (IDH) mutant and IDH wildtype gliomaspheres uncovers cell type-specific vulnerabilities. <i>Cancer &amp; Metabolism</i> , 2018, 6, 4.	5.0	55
9	Identification of novel candidate genes for 46,XY disorders of sex development (DSD) using a C57BL/6J-Y POS mouse model. <i>Biology of Sex Differences</i> , 2018, 9, 8.	4.1	14
10	<i>DMD</i> genotype correlations from the Duchenne Registry: Endogenous exon skipping is a factor in prolonged ambulation for individuals with a defined mutation subtype. <i>Human Mutation</i> , 2018, 39, 1193-1202.	2.5	65
11	Genomic predictors of remission to antidepressant treatment in geriatric depression using genome-wide expression analyses: a pilot study. <i>International Journal of Geriatric Psychiatry</i> , 2016, 31, 510-517.	2.7	17
12	Sex-Specific Effects of Testosterone on the Sexually Dimorphic Transcriptome and Epigenome of Embryonic Neural Stem/Progenitor Cells. <i>Scientific Reports</i> , 2016, 6, 36916.	3.3	41
13	A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. <i>Cilia</i> , 2016, 5, 8.	1.8	37
14	Failure to up-regulate transcription of genes necessary for muscle adaptation underlies limb girdle muscular dystrophy 2A (calpainopathy). <i>Human Molecular Genetics</i> , 2016, 25, 2194-2207.	2.9	25
15	Leveraging ancestry to improve causal variant identification in exome sequencing for monogenic disorders. <i>European Journal of Human Genetics</i> , 2016, 24, 113-119.	2.8	3
16	Role of Mir-34 Upregulation in Disruption of c-Myc, c-Myb and NOTCH Signaling in Diamond-Blackfan Anemia. <i>Blood</i> , 2016, 128, 3895-3895.	1.4	0
17	Mutation in TWINKLE in a Large Iranian Family with Progressive External Ophthalmoplegia, Myopathy, Dysphagia and Dysphonia, and Behavior Change. <i>Archives of Iranian Medicine</i> , 2016, 19, 87-91.	0.6	4
18	Loss of the scavenger mRNA decapping enzyme DCPS causes syndromic intellectual disability with neuromuscular defects. <i>Human Molecular Genetics</i> , 2015, 24, 3163-3171.	2.9	31

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19	Ethanol-induced differential gene expression and acetyl-CoA metabolism in a longevity model of the nematode <i>Caenorhabditis elegans</i> . <i>Experimental Gerontology</i> , 2015, 61, 20-30.	2.8	21
20	Online Self-Report Data for Duchenne Muscular Dystrophy Confirms Natural History and Can Be Used to Assess for Therapeutic Benefits. <i>PLOS Currents</i> , 2014, 6, .	1.4	25
21	EGFR Mutation-Induced Alternative Splicing of Max Contributes to Growth of Glycolytic Tumors in Brain Cancer. <i>Cell Metabolism</i> , 2013, 17, 1000-1008.	16.2	130