

Shay Tzur

List of Publications by Year in descending order

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Version: 2024-02-01

42
papers

2,831
citations

279798

23
h-index

276875

41
g-index

43
all docs

43
docs citations

43
times ranked

4321
citing authors

#	ARTICLE	IF	CITATIONS
1	The Genomic History of the Bronze Age Southern Levant. <i>Cell</i> , 2020, 181, 1146-1157.e11.	28.9	51
2	Lysine acetyltransferase 8 is involved in cerebral development and syndromic intellectual disability. <i>Journal of Clinical Investigation</i> , 2020, 130, 1431-1445.	8.2	40
3	Primary Coenzyme Q deficiency Due to Novel ADCK3 Variants, Studies in Fibroblasts and Review of Literature. <i>Neurochemical Research</i> , 2019, 44, 2372-2384.	3.3	15
4	Gain-of-function mutation of microRNA-140 in human skeletal dysplasia. <i>Nature Medicine</i> , 2019, 25, 583-590.	30.7	86
5	Biallelic mutations in EXOC3L2 cause a novel syndrome that affects the brain, kidney and blood. <i>Journal of Medical Genetics</i> , 2019, 56, 340-346.	3.2	9
6	Variant in SCYL1 gene causes aberrant splicing in a family with cerebellar ataxia, recurrent episodes of liver failure, and growth retardation. <i>European Journal of Human Genetics</i> , 2019, 27, 263-268.	2.8	22
7	Whole-exome sequencing reveals POC5 as a novel gene associated with autosomal recessive retinitis pigmentosa. <i>Human Molecular Genetics</i> , 2018, 27, 614-624.	2.9	26
8	A de novo GABRA2 missense mutation in severe early-onset epileptic encephalopathy with a choreiform movement disorder. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 516-524.	1.6	23
9	A null variant in the apolipoprotein L3 gene is associated with non-diabetic nephropathy. <i>Nephrology Dialysis Transplantation</i> , 2018, 33, 323-330.	0.7	25
10	Big data analysis of human mitochondrial DNA substitution models: a regression approach. <i>BMC Genomics</i> , 2018, 19, 759.	2.8	5
11	Strictly conserved tri-nucleotide motif "CAT" is associated with TAS DNA protein-binding sites in human mitochondrial DNA control region. <i>Mitochondrial DNA Part A: DNA Mapping, Sequencing, and Analysis</i> , 2017, 28, 250-253.	0.7	3
12	Deficiency of the sphingosine-1-phosphate lyase SGPL1 is associated with congenital nephrotic syndrome and congenital adrenal calcifications. <i>Human Mutation</i> , 2017, 38, 365-372.	2.5	71
13	A Biallelic Mutation in the Homologous Recombination Repair Gene SPIDR Is Associated With Human Gonadal Dysgenesis. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 681-688.	3.6	47
14	Clinical Heterogeneity and Phenotypic Expansion of NaPi-IIa Associated Disease. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 4604-4614.	3.6	22
15	The genetic variation in the R1a clade among the Ashkenazi Levites™ Y chromosome. <i>Scientific Reports</i> , 2017, 7, 14969.	3.3	13
16	Exome sequencing identified mutations in CASK and MYBPC3 as the cause of a complex dilated cardiomyopathy phenotype. <i>Genetical Research</i> , 2016, 98, e8.	0.9	3
17	Congenital dilated cardiomyopathy caused by biallelic mutations in Filamin C. <i>European Journal of Human Genetics</i> , 2016, 24, 1792-1796.	2.8	36
18	Intellectual disability and non-compaction cardiomyopathy with a de novo NONO mutation identified by exome sequencing. <i>European Journal of Human Genetics</i> , 2016, 24, 1635-1638.	2.8	22

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19	Exome sequencing identified a novel <i>de novo</i> OPA1 mutation in a consanguineous family presenting with optic atrophy. <i>Genetical Research</i> , 2016, 98, e10.	0.9	5
20	Lethal neonatal rigidity and multifocal seizure syndrome – Report of another family with a BRAT1 mutation. <i>European Journal of Paediatric Neurology</i> , 2015, 19, 240-242.	1.6	28
21	Mutations in <i>TAX1BP3</i> Cause Dilated Cardiomyopathy with Septo-Optic Dysplasia. <i>Human Mutation</i> , 2015, 36, 439-442.	2.5	21
22	A founder effect for p47phox Trp193Ter chronic granulomatous disease in Kavkazi Jews. <i>Blood Cells, Molecules, and Diseases</i> , 2015, 55, 320-327.	1.4	12
23	Exome Sequencing Reveals SYCE1 Mutation Associated With Autosomal Recessive Primary Ovarian Insufficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014, 99, E2129-E2132.	3.6	128
24	Phylogenetic applications of whole Y-chromosome sequences and the Near Eastern origin of Ashkenazi Levites. <i>Nature Communications</i> , 2013, 4, 2928.	12.8	31
25	Possible incipient sympatric ecological speciation in blind mole rats (<i>Spalax</i>). <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 2587-2592.	7.1	58
26	No Evidence from Genome-Wide Data of a Khazar Origin for the Ashkenazi Jews. <i>Human Biology</i> , 2013, 85, 859-900.	0.2	68
27	High Population Frequencies of APOL1 Risk Variants Are Associated with Increased Prevalence of Non-Diabetic Chronic Kidney Disease in the Igbo People from South-Eastern Nigeria. <i>Nephron Clinical Practice</i> , 2013, 123, 123-128.	2.3	82
28	No Evidence from Genome-wide Data of a Khazar Origin fo the Ashkenazi Jews. <i>Human Biology</i> , 2013, 85, 859.	0.2	30
29	APOL1 Risk Variants Predict Histopathology and Progression to ESRD in HIV-Related Kidney Disease. <i>Journal of the American Society of Nephrology: JASN</i> , 2012, 23, 343-350.	6.1	107
30	APOL1 allelic variants are associated with lower age of dialysis initiation and thereby increased dialysis vintage in African and Hispanic Americans with non-diabetic end-stage kidney disease. <i>Nephrology Dialysis Transplantation</i> , 2012, 27, 1498-1505.	0.7	85
31	Linkage disequilibrium analysis reveals an albuminuria risk haplotype containing three missense mutations in the cubilin gene with striking differences among European and African ancestry populations. <i>BMC Nephrology</i> , 2012, 13, 142.	1.8	7
32	The population genetics of chronic kidney disease: insights from the MYH9 – APOL1 locus. <i>Nature Reviews Nephrology</i> , 2011, 7, 313-326.	9.6	58
33	Absence of APOL1 Risk Variants Protects against HIV-Associated Nephropathy in the Ethiopian Population. <i>American Journal of Nephrology</i> , 2011, 34, 452-459.	3.1	66
34	Missense mutations in the APOL1 gene are highly associated with end stage kidney disease risk previously attributed to the MYH9 gene. <i>Human Genetics</i> , 2010, 128, 345-350.	3.8	539
35	African ancestry allelic variation at the MYH9 gene contributes to increased susceptibility to non-diabetic end-stage kidney disease in Hispanic Americans. <i>Human Molecular Genetics</i> , 2010, 19, 1816-1827.	2.9	75
36	A novel 154-bp deletion in the human mitochondrial DNA control region in healthy individuals. <i>Human Mutation</i> , 2008, 29, 1387-1391.	2.5	11

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37	The Dawn of Human Matrilineal Diversity. <i>American Journal of Human Genetics</i> , 2008, 82, 1130-1140.	6.2	392
38	Maternal traces of deep common ancestry and asymmetric gene flow between Pygmy hunter-gatherers and Bantu-speaking farmers. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2008, 105, 1596-1601.	7.1	157
39	Counting the Founders: The Matrilineal Genetic Ancestry of the Jewish Diaspora. <i>PLoS ONE</i> , 2008, 3, e2062.	2.5	101
40	The Genographic Project Public Participation Mitochondrial DNA Database. <i>PLoS Genetics</i> , 2007, 3, e104.	3.5	99
41	The Matrilineal Ancestry of Ashkenazi Jewry: Portrait of a Recent Founder Event. <i>American Journal of Human Genetics</i> , 2006, 78, 487-497.	6.2	140
42	Odour-genes covariance within a natural population of subterranean <i>Spalax galili</i> blind mole rats. <i>Biological Journal of the Linnean Society</i> , 0, 96, 483-490.	1.6	12