

# Tamar Harel

## List of Publications by Year in descending order

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Version: 2024-02-01

51  
papers

2,621  
citations

331670

21  
h-index

206112

48  
g-index

52  
all docs

52  
docs citations

52  
times ranked

6279  
citing authors

#	ARTICLE	IF	CITATIONS
1	Infantile SOD1 deficiency syndrome caused by a homozygous <i>SOD1</i> variant with absence of enzyme activity. <i>Brain</i> , 2022, 145, 872-878.	7.6	10
2	RECON syndrome is a genome instability disorder caused by mutations in the DNA helicase RECQL1. <i>Journal of Clinical Investigation</i> , 2022, 132, .	8.2	21
3	CRISPR/Cas9-induced gene conversion between ATAD3 paralogs. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100092.	1.7	1
4	Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity. <i>American Journal of Human Genetics</i> , 2022, 109, 518-532.	6.2	8
5	De novo variants in <i>EMC1</i> lead to neurodevelopmental delay and cerebellar degeneration and affect glial function in <i>Drosophila</i> . <i>Human Molecular Genetics</i> , 2022, 31, 3231-3244.	2.9	5
6	Orbital nodular fasciitis in child with biallelic germline RBL2 variant. <i>European Journal of Medical Genetics</i> , 2022, 65, 104513.	1.3	0
7	<i>De novo</i> heterozygous variants in <i>SLC30A7</i> are a candidate cause for Joubert syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2360-2366.	1.2	3
8	De novo variants in <i>SIAH1</i> , encoding an E3 ubiquitin ligase, are associated with developmental delay, hypotonia and dysmorphic features. <i>Journal of Medical Genetics</i> , 2021, 58, 205-212.	3.2	6
9	De novo variant in <i>AMOTL1</i> in infant with cleft lip and palate, imperforate anus and dysmorphic features. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 190-195.	1.2	3
10	Parental exome analysis identifies shared carrier status for a second recessive disorder in couples with an affected child. <i>European Journal of Human Genetics</i> , 2021, 29, 455-462.	2.8	8
11	Homozygous variant in <i>MADD</i> , encoding a Rab guanine nucleotide exchange factor, results in pleiotropic effects and a multisystemic disorder. <i>European Journal of Human Genetics</i> , 2021, 29, 977-987.	2.8	6
12	Functional interpretation of <i>ATAD3A</i> variants in neuro-mitochondrial phenotypes. <i>Genome Medicine</i> , 2021, 13, 55.	8.2	16
13	Biallelic deletion in a minimal <i>CAPN15</i> intron in siblings with a recognizable syndrome of congenital malformations and developmental delay. <i>Clinical Genetics</i> , 2021, 99, 577-582.	2.0	9
14	Levodopa-responsive dystonia caused by biallelic <i>PRKN</i> exon inversion invisible to exome sequencing. <i>Brain Communications</i> , 2021, 3, fcb197.	3.3	5
15	Response to Hall et al.. <i>American Journal of Human Genetics</i> , 2020, 107, 1188-1189.	6.2	0
16	Mutations in <i>MYLPH</i> Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogyposis. <i>American Journal of Human Genetics</i> , 2020, 107, 293-310.	6.2	21
17	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. <i>Nature Communications</i> , 2020, 11, 4625.	12.8	47
18	Whole-genome sequencing reveals complex chromosome rearrangement disrupting <i>NIPBL</i> in infant with Cornelia de Lange syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1143-1151.	1.2	17

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19	Grandparental genotyping enhances exome variant interpretation. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 689-696.	1.2	4
20	Recurrent De Novo NAHR Reciprocal Duplications in the ATAD3 Gene Cluster Cause a Neurogenetic Trait with Perturbed Cholesterol and Mitochondrial Metabolism. <i>American Journal of Human Genetics</i> , 2020, 106, 272-279.	6.2	33
21	Loss of function mutations in CCDC32 cause a congenital syndrome characterized by craniofacial, cardiac and neurodevelopmental anomalies. <i>Human Molecular Genetics</i> , 2020, 29, 1489-1497.	2.9	6
22	Fetal exome sequencing: yield and limitations in a tertiary referral center. <i>Ultrasound in Obstetrics and Gynecology</i> , 2019, 53, 80-86.	1.7	44
23	Homozygous frameshift variant in NTNG2, encoding a synaptic cell adhesion molecule, in individuals with developmental delay, hypotonia, and autistic features. <i>Neurogenetics</i> , 2019, 20, 209-213.	1.4	7
24	Homozygous variants in <i>MAPRE2</i> and <i>CDON</i> in individual with skin folds, growth delay, retinal coloboma, and pyloric stenosis. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2454-2458.	1.2	9
25	Pathogenic Variants in NUP214 Cause "Plugged" Nuclear Pore Channels and Acute Febrile Encephalopathy. <i>American Journal of Human Genetics</i> , 2019, 105, 48-64.	6.2	29
26	Homozygous stop-gain variant in LRRC32, encoding a TGF $\beta$ 2 receptor, associated with cleft palate, proliferative retinopathy, and developmental delay. <i>European Journal of Human Genetics</i> , 2019, 27, 1315-1319.	2.8	7
27	Biallelic variants in AGTPBP1, involved in tubulin deglutamylation, are associated with cerebellar degeneration and motor neuropathy. <i>European Journal of Human Genetics</i> , 2019, 27, 1419-1426.	2.8	25
28	Insights into genetics, human biology and disease gleaned from family based genomic studies. <i>Genetics in Medicine</i> , 2019, 21, 798-812.	2.4	161
29	MARS variant associated with both recessive interstitial lung and liver disease and dominant Charcot-Marie-Tooth disease. <i>European Journal of Medical Genetics</i> , 2018, 61, 616-620.	1.3	14
30	Phenotypic expansion illuminates multilocus pathogenic variation. <i>Genetics in Medicine</i> , 2018, 20, 1528-1537.	2.4	104
31	Comprehensive genomic analysis of patients with disorders of cerebral cortical development. <i>European Journal of Human Genetics</i> , 2018, 26, 1121-1131.	2.8	35
32	Genomic disorders 20 years on—"mechanisms for clinical manifestations. <i>Clinical Genetics</i> , 2018, 93, 439-449.	2.0	102
33	Identification of a pathogenic PMP2 variant in a multi-generational family with CMT type 1: Clinical gene panels versus genome-wide approaches to molecular diagnosis. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 302-304.	1.1	13
34	Homozygous mutation in MFSD2A, encoding a lysolipid transporter for docosahexanoic acid, is associated with microcephaly and hypomyelination. <i>Neurogenetics</i> , 2018, 19, 227-235.	1.4	45
35	22q11.2q13 duplication including <i>SOX10</i> causes sex-reversal and peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome, and Hirschsprung disease. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1066-1070.	1.2	23
36	Lessons learned from additional research analyses of unsolved clinical exome cases. <i>Genome Medicine</i> , 2017, 9, 26.	8.2	184

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37	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. <i>New England Journal of Medicine</i> , 2017, 376, 21-31.	27.0	565
38	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 716-724.	6.2	66
39	Homozygous null variant in <i>CRADD</i> , encoding an adaptor protein that mediates apoptosis, is associated with lissencephaly. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2539-2544.	1.2	18
40	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. <i>PLoS Genetics</i> , 2017, 13, e1006905.	3.5	80
41	Nonrecurrent PMP22-RAI1 contiguous gene deletions arise from replication-based mechanisms and result in Smith-Magenis syndrome with evident peripheral neuropathy. <i>Human Genetics</i> , 2016, 135, 1161-1174.	3.8	4
42	Bi-allelic Mutations in PKD1L1 Are Associated with Laterality Defects in Humans. <i>American Journal of Human Genetics</i> , 2016, 99, 886-893.	6.2	57
43	Recurrent De Novo and Biallelic Variation of ATAD3A, Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. <i>American Journal of Human Genetics</i> , 2016, 99, 831-845.	6.2	146
44	Identification of a RAI1-associated disease network through integration of exome sequencing, transcriptomics, and 3D genomics. <i>Genome Medicine</i> , 2016, 8, 105.	8.2	20
45	Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. <i>American Journal of Human Genetics</i> , 2016, 98, 562-570.	6.2	66
46	The role of combined SNV and CNV burden in patients with distal symmetric polyneuropathy. <i>Genetics in Medicine</i> , 2016, 18, 443-451.	2.4	18
47	Atypical presentation of moyamoya disease in an infant with a de novo <i>RNF213</i> variant. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2742-2747.	1.2	15
48	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. <i>Cell Reports</i> , 2015, 12, 1169-1183.	6.4	211
49	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. <i>Neuron</i> , 2015, 88, 499-513.	8.1	258
50	Nonrecurrent 17p11.2p12 Rearrangement Events that Result in Two Concomitant Genomic Disorders: The PMP22-RAI1 Contiguous Gene Duplication Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 691-707.	6.2	33
51	Charcot-Marie-Tooth disease and pathways to molecular based therapies. <i>Clinical Genetics</i> , 2014, 86, 422-431.	2.0	28