Tamar Harel

List of Publications by Year
in descending order

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	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	27.0	565
2	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	8.1	258
3	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell Reports, 2015, 12, 1169-1183.	6.4	211
4	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	8.2	184
5	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	2.4	161
6	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	6.2	146
7	Phenotypic expansion illuminates multilocus pathogenic variation. Genetics in Medicine, 2018, 20, 1528-1537.	2.4	104
8	Genomic disorders 20 years onâ€"mechanisms for clinical manifestations. Clinical Genetics, 2018, 93, 439-449.	2.0	102
9	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	3.5	80
10	Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. American Journal of Human Genetics, 2016, 98, 562-570.	6.2	66
11	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	6.2	66
12	Bi-allelic Mutations in PKD1L1 Are Associated with Laterality Defects in Humans. American Journal of Human Genetics, 2016, 99, 886-893.	6.2	57
13	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. Nature Communications, 2020, 11, 4625.	12.8	47
14	Homozygous mutation in MFSD2A, encoding a lysolipid transporter for docosahexanoic acid, is associated with microcephaly and hypomyelination. Neurogenetics, 2018, 19, 227-235.	1.4	45
15	Fetal exome sequencing: yield and limitations in a tertiary referral center. Ultrasound in Obstetrics and Gynecology, 2019, 53, 80-86.	1.7	44
16	Comprehensive genomic analysis of patients with disorders of cerebral cortical development. European Journal of Human Genetics, 2018, 26, 1121-1131.	2.8	35
17	Nonrecurrent 17p11.2p12 Rearrangement Events that Result in Two Concomitant Genomic Disorders: The PMP22-RAI1 Contiguous Gene Duplication Syndrome. American Journal of Human Genetics, 2015, 97, 691-707.	6.2	33
18	Recurrent De Novo NAHR Reciprocal Duplications in the ATAD3 Gene Cluster Cause a Neurogenetic Trait with Perturbed Cholesterol and Mitochondrial Metabolism. American Journal of Human Genetics, 2020, 106, 272-279.	6.2	33

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19	Pathogenic Variants in NUP214 Cause â€œPluggedâ€•Nuclear Pore Channels and Acute Febrile Encephalopathy. American Journal of Human Genetics, 2019, 105, 48-64.	6.2	29
20	Charcotâ€“Marieâ€“Tooth disease and pathways to molecular based therapies. Clinical Genetics, 2014, 86, 422-431.	2.0	28
21	Biallelic variants in AGTPBP1, involved in tubulin deglutamylation, are associated with cerebellar degeneration and motor neuropathy. European Journal of Human Genetics, 2019, 27, 1419-1426.	2.8	25
22	22q11.2q13 duplication including SOX10 causes sexâ€reversal and peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome, and Hirschsprung disease. American Journal of Medical Genetics, Part A, 2017, 173, 1066-1070.	1.2	23
23	Mutations in MYLPF Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogryposis. American Journal of Human Genetics, 2020, 107, 293-310.	6.2	21
24	RECON syndrome is a genome instability disorder caused by mutations in the DNA helicase RECQL1. Journal of Clinical Investigation, 2022, 132, .	8.2	21
25	Identification of a RAI1-associated disease network through integration of exome sequencing, transcriptomics, and 3D genomics. Genome Medicine, 2016, 8, 105.	8.2	20
26	The role of combined SNV and CNV burden in patients with distal symmetric polyneuropathy. Genetics in Medicine, 2016, 18, 443-451.	2.4	18
27	Homozygous null variant in CRADD, encoding an adaptor protein that mediates apoptosis, is associated with lissencephaly. American Journal of Medical Genetics, Part A, 2017, 173, 2539-2544.	1.2	18
28	Wholeâ€genome sequencing reveals complex chromosome rearrangement disrupting <scp>NIPBL</scp> in infant with Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1143-1151.	1.2	17
29	Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes. Genome Medicine, 2021, 13, 55.	8.2	16
30	Atypical presentation of moyamoya disease in an infant with a de novo RNF213 variant. American Journal of Medical Genetics, Part A, 2015, 167, 2742-2747.	1.2	15
31	MARS variant associated with both recessive interstitial lung and liver disease and dominant Charcot-Marie-Tooth disease. European Journal of Medical Genetics, 2018, 61, 616-620.	1.3	14
32	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. Molecular Genetics and Metabolism, 2018, 125, 302-304.	1.1	13
33	Infantile SOD1 deficiency syndrome caused by a homozygous SOD1 variant with absence of enzyme activity. Brain, 2022, 145, 872-878.	7.6	10
34	Homozygous variants in MAPRE2 and CDON in individual with skin folds, growth delay, retinal coloboma, and pyloric stenosis. American Journal of Medical Genetics, Part A, 2019, 179, 2454-2458.	1.2	9
35	Biallelic deletion in a minimal <scp>CAPN15</scp> intron in siblings with a recognizable syndrome of congenital malformations and developmental delay. Clinical Genetics, 2021, 99, 577-582.	2.0	9
36	Parental exome analysis identifies shared carrier status for a second recessive disorder in couples with an affected child. European Journal of Human Genetics, 2021, 29, 455-462.	2.8	8

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37	Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity. American Journal of Human Genetics, 2022, 109, 518-532.	6.2	8
38	Homozygous frameshift variant in NTNG2, encoding a synaptic cell adhesion molecule, in individuals with developmental delay, hypotonia, and autistic features. Neurogenetics, 2019, 20, 209-213.	1.4	7
39	Homozygous stop-gain variant in LRRC32, encoding a TGFÎ² receptor, associated with cleft palate, proliferative retinopathy, and developmental delay. European Journal of Human Genetics, 2019, 27, 1315-1319.	2.8	7
40	Loss of function mutations in CCDC32 cause a congenital syndrome characterized by craniofacial, cardiac and neurodevelopmental anomalies. Human Molecular Genetics, 2020, 29, 1489-1497.	2.9	6
41	De novo variants in SIAH1, encoding an E3 ubiquitin ligase, are associated with developmental delay, hypotonia and dysmorphic features. Journal of Medical Genetics, 2021, 58, 205-212.	3.2	6
42	Homozygous variant in MADD, encoding a Rab guanine nucleotide exchange factor, results in pleiotropic effects and a multisystemic disorder. European Journal of Human Genetics, 2021, 29, 977-987.	2.8	6
43	Levodopa-responsive dystonia caused by biallelic PRKN exon inversion invisible to exome sequencing. Brain Communications, 2021, 3, fcab197.	3.3	5
44	De novo variants in EMC1 lead to neurodevelopmental delay and cerebellar degeneration and affect glial function in Drosophila. Human Molecular Genetics, 2022, 31, 3231-3244.	2.9	5
45	Nonrecurrent PMP22-RAI1 contiguous gene deletions arise from replication-based mechanisms and result in Smithâ€“Magenis syndrome with evident peripheral neuropathy. Human Genetics, 2016, 135, 1161-1174.	3.8	4
46	Grandparental genotyping enhances exome variant interpretation. American Journal of Medical Genetics, Part A, 2020, 182, 689-696.	1.2	4
47	De novo variant in AMOTL1 in infant with cleft lip and palate, imperforate anus and dysmorphic features. American Journal of Medical Genetics, Part A, 2021, 185, 190-195.	1.2	3
48	De novo heterozygous variants in <scp>SLC30A7</scp> are a candidate cause for Joubert syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2360-2366.	1.2	3
49	CRISPR/Cas9-induced gene conversion between ATAD3 paralogs. Human Genetics and Genomics Advances, 2022, 3, 100092.	1.7	1
50	Response to Hall etÂal.. American Journal of Human Genetics, 2020, 107, 1188-1189.	6.2	0
51	Orbital nodular fasciitis in child with biallelic germline RBL2 variant. European Journal of Medical Genetics, 2022, 65, 104513.	1.3	0