

Tom Walsh

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

67 papers	6,965 citations	29 h-index	69 g-index
69 ext. papers	8,227 ext. citations	10.4 avg, IF	5.06 L-index

#	Paper	IF	Citations
67	Rare structural variants disrupt multiple genes in neurodevelopmental pathways in schizophrenia. <i>Science</i> , 2008 , 320, 539-43	33.3	1443
66	Inherited DNA-Repair Gene Mutations in Men with Metastatic Prostate Cancer. <i>New England Journal of Medicine</i> , 2016 , 375, 443-53	59.2	791
65	Spectrum of mutations in BRCA1, BRCA2, CHEK2, and TP53 in families at high risk of breast cancer. <i>JAMA - Journal of the American Medical Association</i> , 2006 , 295, 1379-88	27.4	502
64	Inherited Mutations in Women With Ovarian Carcinoma. <i>JAMA Oncology</i> , 2016 , 2, 482-90	13.4	410
63	Detection of inherited mutations for breast and ovarian cancer using genomic capture and massively parallel sequencing. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010 , 107, 12629-33	11.5	372
62	Germline ETV6 mutations in familial thrombocytopenia and hematologic malignancy. <i>Nature Genetics</i> , 2015 , 47, 180-5	36.3	239
61	Whole exome sequencing and homozygosity mapping identify mutation in the cell polarity protein GPSM2 as the cause of nonsyndromic hearing loss DFNB82. <i>American Journal of Human Genetics</i> , 2010 , 87, 90-4	11	223
60	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. <i>Nature Genetics</i> , 2013 , 45, 1226-1231	36.3	205
59	Validation and implementation of targeted capture and sequencing for the detection of actionable mutation, copy number variation, and gene rearrangement in clinical cancer specimens. <i>Journal of Molecular Diagnostics</i> , 2014 , 16, 56-67	5.1	203
58	From fliesVeyes to our ears: mutations in a human class III myosin cause progressive nonsyndromic hearing loss DFNB30. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2002 , 99, 7518-23	11.5	202
57	Complex MSH2 and MSH6 mutations in hypermutated microsatellite unstable advanced prostate cancer. <i>Nature Communications</i> , 2014 , 5, 4988	17.4	182
56	Targeted genomic capture and massively parallel sequencing to identify genes for hereditary hearing loss in Middle Eastern families. <i>Genome Biology</i> , 2011 , 12, R89	18.3	163
55	Accurate and exact CNV identification from targeted high-throughput sequence data. <i>BMC Genomics</i> , 2011 , 12, 184	4.5	156
54	Perrault syndrome is caused by recessive mutations in CLPP, encoding a mitochondrial ATP-dependent chambered protease. <i>American Journal of Human Genetics</i> , 2013 , 92, 605-13	11	152
53	Novel germ line DDX41 mutations define families with a lower age of MDS/AML onset and lymphoid malignancies. <i>Blood</i> , 2016 , 127, 1017-23	2.2	117
52	Mutations in Homologous Recombination Genes and Outcomes in Ovarian Carcinoma Patients in GOG 218: An NRG Oncology/Gynecologic Oncology Group Study. <i>Clinical Cancer Research</i> , 2018 , 24, 777-783	12.9	112
51	Mitochondrial serine protease HTRA2 p.G399S in a kindred with essential tremor and Parkinson disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014 , 111, 18285-90	11.5	96

50	Genomic duplication and overexpression of TJP2/ZO-2 leads to altered expression of apoptosis genes in progressive nonsyndromic hearing loss DFNA51. <i>American Journal of Human Genetics</i> , 2010 , 87, 101-9	11	82
49	Genomic analysis of bone marrow failure and myelodysplastic syndromes reveals phenotypic and diagnostic complexity. <i>Haematologica</i> , 2015 , 100, 42-8	6.6	81
48	Genetic features of myelodysplastic syndrome and aplastic anemia in pediatric and young adult patients. <i>Haematologica</i> , 2016 , 101, 1343-1350	6.6	74
47	Mutations in Twinkle primase-helicase cause Perrault syndrome with neurologic features. <i>Neurology</i> , 2014 , 83, 2054-61	6.5	73
46	Somatic Mosaic Mutations in PPM1D and TP53 in the Blood of Women With Ovarian Carcinoma. <i>JAMA Oncology</i> , 2016 , 2, 370-2	13.4	68
45	Inherited predisposition to malignant mesothelioma and overall survival following platinum chemotherapy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019 , 116, 9008-9013	11.5	67
44	Improving performance of multigene panels for genomic analysis of cancer predisposition. <i>Genetics in Medicine</i> , 2016 , 18, 974-81	8.1	60
43	Genetic Predisposition to Breast Cancer Due to Mutations Other Than BRCA1 and BRCA2 Founder Alleles Among Ashkenazi Jewish Women. <i>JAMA Oncology</i> , 2017 , 3, 1647-1653	13.4	42
42	Genomic analysis of a heterogeneous Mendelian phenotype: multiple novel alleles for inherited hearing loss in the Palestinian population. <i>Human Genomics</i> , 2006 , 2, 203-11	6.8	39
41	A girl with West syndrome and autistic features harboring a de novo TBL1XR1 mutation. <i>Journal of Human Genetics</i> , 2014 , 59, 581-3	4.3	34
40	Identifying Inherited and Acquired Genetic Factors Involved in Poor Stem Cell Mobilization and Donor-Derived Malignancy. <i>Biology of Blood and Marrow Transplantation</i> , 2016 , 22, 2100-2103	4.7	31
39	BARD1 is necessary for ubiquitylation of nucleosomal histone H2A and for transcriptional regulation of estrogen metabolism genes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018 , 115, 1316-1321	11.5	27
38	Features of Patients With Hereditary Mixed Polyposis Syndrome Caused by Duplication of GREM1 and Implications for Screening and Surveillance. <i>Gastroenterology</i> , 2017 , 152, 1876-1880.e1	13.3	25
37	Intensive Surveillance with Biannual Dynamic Contrast-Enhanced Magnetic Resonance Imaging Downstages Breast Cancer in Mutation Carriers. <i>Clinical Cancer Research</i> , 2019 , 25, 1786-1794	12.9	23
36	Mitochondrial peptidase loss-of-function in childhood cerebellar atrophy. <i>Journal of Medical Genetics</i> , 2018 , 55, 599-606	5.8	22
35	Vesicular acetylcholine transporter defect underlies devastating congenital myasthenia syndrome. <i>Neurology</i> , 2017 , 88, 1021-1028	6.5	21
34	MYO3A Causes Human Dominant Deafness and Interacts with Protocadherin 15-CD2 Isoform. <i>Human Mutation</i> , 2016 , 37, 481-7	4.7	21
33	Genomic analysis of inherited breast cancer among Palestinian women: Genetic heterogeneity and a founder mutation in TP53. <i>International Journal of Cancer</i> , 2017 , 141, 750-756	7.5	20

32	Mechanism for survival of homozygous nonsense mutations in the tumor suppressor gene. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018 , 115, 5241-5246	11.5	20
31	Using Somatic Mutations from Tumors to Classify Variants in Mismatch Repair Genes. <i>American Journal of Human Genetics</i> , 2018 , 103, 19-29	11	19
30	Family-Specific Variants and the Limits of Human Genetics. <i>Trends in Molecular Medicine</i> , 2016 , 22, 925-934	11.5	18
29	Characterization of splice-altering mutations in inherited predisposition to cancer. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019 ,	11.5	17
28	Targeted long-read sequencing identifies missing disease-causing variation. <i>American Journal of Human Genetics</i> , 2021 , 108, 1436-1449	11	16
27	De novo mutation in with epigenetic effects on neurodevelopment. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018 , 115, 1558-1563	11.5	15
26	Genetics of hearing loss in the Arab population of Northern Israel. <i>European Journal of Human Genetics</i> , 2018 , 26, 1840-1847	5.3	15
25	Mutation of KREMEN1, a modulator of Wnt signaling, is responsible for ectodermal dysplasia including oligodontia in Palestinian families. <i>European Journal of Human Genetics</i> , 2016 , 24, 1430-5	5.3	15
24	Systematic misclassification of missense variants in BRCA1 and BRCA2 "coldspots". <i>Genetics in Medicine</i> , 2020 , 22, 825-830	8.1	14
23	Identification of a new BRCA2 large genomic deletion associated with high risk male breast cancer. <i>Hereditary Cancer in Clinical Practice</i> , 2015 , 13, 2	2.3	13
22	Genomic analysis of inherited hearing loss in the Palestinian population. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020 , 117, 20070-20076	11.5	12
21	Germline variants drive myelodysplastic syndrome in young adults. <i>Leukemia</i> , 2021 , 35, 2439-2444	10.7	11
20	Infantile onset spinocerebellar ataxia caused by compound heterozygosity for Twinkle mutations and modeling of Twinkle mutations causing recessive disease. <i>Journal of Physical Education and Sports Management</i> , 2016 , 2, a001107	2.8	9
19	Genetic characterization of early onset ovarian carcinoma. <i>Gynecologic Oncology</i> , 2016 , 140, 221-5	4.9	8
18	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene ATOH1. <i>Clinical Genetics</i> , 2020 , 98, 353-364	4	8
17	CRISPR-Cas9/long-read sequencing approach to identify cryptic mutations in and other tumour suppressor genes. <i>Journal of Medical Genetics</i> , 2021 , 58, 850-852	5.8	7
16	A defect in GPI synthesis as a suggested mechanism for the role of ARV1 in intellectual disability and seizures. <i>Neurogenetics</i> , 2020 , 21, 259-267	3	5
15	Effects of germline and somatic events in candidate BRCA-like genes on breast-tumor signatures. <i>PLoS ONE</i> , 2020 , 15, e0239197	3.7	4

14	Molecular diagnosis of childhood immune dysregulation, polyendocrinopathy, and enteropathy, and implications for clinical management. <i>Journal of Allergy and Clinical Immunology</i> , 2021 ,	11.5	4
13	Variable Features of Juvenile Polyposis Syndrome With Gastric Involvement Among Patients With a Large Genomic Deletion of BMPR1A. <i>Clinical and Translational Gastroenterology</i> , 2019 , 10, e00054	4.2	4
12	Inherited predisposition to breast cancer in the Carolina Breast Cancer Study. <i>Npj Breast Cancer</i> , 2021 , 7, 6	7.8	3
11	Reply to Tzoulis et al.: Genetic and clinical heterogeneity of essential tremor. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015 , 112, E2269	11.5	1
10	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene ATOH1		1
9	Testing Ashkenazi Jewish Women for Mutations Predisposing to Breast Cancer in Genes Other Than BRCA1 and BRCA2-Reply. <i>JAMA Oncology</i> , 2018 , 4, 1012-1013	13.4	1
8	Mutational spectrum of breast cancer susceptibility genes among women ascertained in a cancer risk clinic in Northeast Brazil.. <i>Breast Cancer Research and Treatment</i> , 2022 , 1	4.4	0
7	Detection of Mutations in Inherited Bone Marrow Failure and Myelodysplastic Syndrome Genes Using Genomic Capture and Massively Parallel Sequencing in Clinical Diagnostics. <i>Blood</i> , 2016 , 128, 1507-1507		
6	Genetic Heterogeneity and Core Clinical Features of NOG-Related-Symphalangism Spectrum Disorder. <i>Otology and Neurotology</i> , 2021 , 42, e1143-e1151	2.6	
5	Effects of germline and somatic events in candidate BRCA-like genes on breast-tumor signatures 2020 , 15, e0239197		
4	Effects of germline and somatic events in candidate BRCA-like genes on breast-tumor signatures 2020 , 15, e0239197		
3	Effects of germline and somatic events in candidate BRCA-like genes on breast-tumor signatures 2020 , 15, e0239197		
2	Effects of germline and somatic events in candidate BRCA-like genes on breast-tumor signatures 2020 , 15, e0239197		
1	Revisiting Genetic Testing for Patients with Negative Results: IPEX and FOXP3. <i>Journal of Clinical Immunology</i> ,	5.7	