Tom Walsh

List of Publications by Year in Descending Order

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	Mutational spectrum of breast cancer susceptibility genes among women ascertained in a cancer risk clinic in Northeast Brazil.. *Breast Cancer Research and Treatment*, **2022**, 1	4.4	0
66	CRISPR-Cas9/long-read sequencing approach to identify cryptic mutations in and other tumour suppressor genes. *Journal of Medical Genetics*, **2021**, 58, 850-852	5.8	7
65	Molecular diagnosis of childhood immune dysregulation, polyendocrinopathy, and enteropathy, and implications for clinical management. *Journal of Allergy and Clinical Immunology*, **2021**,	11.5	4
64	Genetic Heterogeneity and Core Clinical Features of NOG-Related-Symphalangism Spectrum Disorder. *Otology and Neurotology*, **2021**, 42, e1143-e1151	2.6	
63	Inherited predisposition to breast cancer in the Carolina Breast Cancer Study. *Npj Breast Cancer*, **2021**, 7, 6	7.8	3
62	Germline variants drive myelodysplastic syndrome in young adults. *Leukemia*, **2021**, 35, 2439-2444	10.7	11
61	Targeted long-read sequencing identifies missing disease-causing variation. *American Journal of Human Genetics*, **2021**, 108, 1436-1449	11	16
60	A defect in GPI synthesis as a suggested mechanism for the role of ARV1 in intellectual disability and seizures. *Neurogenetics*, **2020**, 21, 259-267	3	5
59	Systematic misclassification of missense variants in BRCA1 and BRCA2 "coldspots". *Genetics in Medicine*, **2020**, 22, 825-830	8.1	14
58	Effects of germline and somatic events in candidate BRCA-like genes on breast-tumor signatures. *PLoS ONE*, **2020**, 15, e0239197	3.7	4
57	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene ATOH1. *Clinical Genetics*, **2020**, 98, 353-364	4	8
56	Genomic analysis of inherited hearing loss in the Palestinian population. *Proceedings of the National Academy of Sciences of the United States of America*, **2020**, 117, 20070-20076	11.5	12
55	Effects of germline and somatic events in candidate BRCA-like genes on breast-tumor signatures **2020**, 15, e0239197		
54	Effects of germline and somatic events in candidate BRCA-like genes on breast-tumor signatures **2020**, 15, e0239197		
53	Effects of germline and somatic events in candidate BRCA-like genes on breast-tumor signatures **2020**, 15, e0239197		
52	Effects of germline and somatic events in candidate BRCA-like genes on breast-tumor signatures **2020**, 15, e0239197		
51	Inherited predisposition to malignant mesothelioma and overall survival following platinum chemotherapy. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 9008-9013	11.5	67

50	Variable Features of Juvenile Polyposis Syndrome With Gastric Involvement Among Patients With a Large Genomic Deletion of BMPR1A. *Clinical and Translational Gastroenterology*, **2019**, 10, e00054	4.2	4
49	Characterization of splice-altering mutations in inherited predisposition to cancer. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**,	11.5	17
48	Intensive Surveillance with Biannual Dynamic Contrast-Enhanced Magnetic Resonance Imaging Downstages Breast Cancer in Mutation Carriers. *Clinical Cancer Research*, **2019**, 25, 1786-1794	12.9	23
47	De novo mutation in with epigenetic effects on neurodevelopment. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 1558-1563	11.5	15
46	BARD1 is necessary for ubiquitylation of nucleosomal histone H2A and for transcriptional regulation of estrogen metabolism genes. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 1316-1321	11.5	27
45	Mechanism for survival of homozygous nonsense mutations in the tumor suppressor gene. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 5241-5246	11.5	20
44	Genetics of hearing loss in the Arab population of Northern Israel. *European Journal of Human Genetics*, **2018**, 26, 1840-1847	5.3	15
43	Using Somatic Mutations from Tumors to Classify Variants in Mismatch Repair Genes. *American Journal of Human Genetics*, **2018**, 103, 19-29	11	19
42	Mutations in Homologous Recombination Genes and Outcomes in Ovarian Carcinoma Patients in GOG 218: An NRG Oncology/Gynecologic Oncology Group Study. *Clinical Cancer Research*, **2018**, 24, 777-783	12.9	112
41	Mitochondrial peptidase loss-of-function in childhood cerebellar atrophy. *Journal of Medical Genetics*, **2018**, 55, 599-606	5.8	22
40	Testing Ashkenazi Jewish Women for Mutations Predisposing to Breast Cancer in Genes Other Than BRCA1 and BRCA2-Reply. *JAMA Oncology*, **2018**, 4, 1012-1013	13.4	1
39	Features of Patients With Hereditary Mixed Polyposis Syndrome Caused by Duplication of GREM1 and Implications for Screening and Surveillance. *Gastroenterology*, **2017**, 152, 1876-1880.e1	13.3	25
38	Vesicular acetylcholine transporter defect underlies devastating congenital myasthenia syndrome. *Neurology*, **2017**, 88, 1021-1028	6.5	21
37	Genomic analysis of inherited breast cancer among Palestinian women: Genetic heterogeneity and a founder mutation in TP53. *International Journal of Cancer*, **2017**, 141, 750-756	7.5	20
36	Genetic Predisposition to Breast Cancer Due to Mutations Other Than BRCA1 and BRCA2 Founder Alleles Among Ashkenazi Jewish Women. *JAMA Oncology*, **2017**, 3, 1647-1653	13.4	42
35	Identifying Inherited and Acquired Genetic Factors Involved in Poor Stem Cell Mobilization and Donor-Derived Malignancy. *Biology of Blood and Marrow Transplantation*, **2016**, 22, 2100-2103	4.7	31
34	Family-Specific Variants and the Limits of Human Genetics. *Trends in Molecular Medicine*, **2016**, 22, 925-934	14.5	18
33	Genetic features of myelodysplastic syndrome and aplastic anemia in pediatric and young adult patients. *Haematologica*, **2016**, 101, 1343-1350	6.6	74

32	Infantile onset spinocerebellar ataxia caused by compound heterozygosity for Twinkle mutations and modeling of Twinkle mutations causing recessive disease. *Journal of Physical Education and Sports Management*, **2016**, 2, a001107	2.8	9
31	Improving performance of multigene panels for genomic analysis of cancer predisposition. *Genetics in Medicine*, **2016**, 18, 974-81	8.1	60
30	Somatic Mosaic Mutations in PPM1D and TP53 in the Blood of Women With Ovarian Carcinoma. *JAMA Oncology*, **2016**, 2, 370-2	13.4	68
29	Inherited Mutations in Women With Ovarian Carcinoma. *JAMA Oncology*, **2016**, 2, 482-90	13.4	410
28	Genetic characterization of early onset ovarian carcinoma. *Gynecologic Oncology*, **2016**, 140, 221-5	4.9	8
27	Detection of Mutations in Inherited Bone Marrow Failure and Myelodysplastic Syndrome Genes Using Genomic Capture and Massively Parallel Sequencing in Clinical Diagnostics. *Blood*, **2016**, 128, 1507-1507	2.2	
26	MYO3A Causes Human Dominant Deafness and Interacts with Protocadherin 15-CD2 Isoform. *Human Mutation*, **2016**, 37, 481-7	4.7	21
25	Inherited DNA-Repair Gene Mutations in Men with Metastatic Prostate Cancer. *New England Journal of Medicine*, **2016**, 375, 443-53	59.2	791
24	Mutation of KREMEN1, a modulator of Wnt signaling, is responsible for ectodermal dysplasia including oligodontia in Palestinian families. *European Journal of Human Genetics*, **2016**, 24, 1430-5	5.3	15
23	Novel germ line DDX41 mutations define families with a lower age of MDS/AML onset and lymphoid malignancies. *Blood*, **2016**, 127, 1017-23	2.2	117
22	Identification of a new BRCA2 large genomic deletion associated with high risk male breast cancer. *Hereditary Cancer in Clinical Practice*, **2015**, 13, 2	2.3	13
21	Reply to Tzoulis et al.: Genetic and clinical heterogeneity of essential tremor. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, E2269	11.5	1
20	Genomic analysis of bone marrow failure and myelodysplastic syndromes reveals phenotypic and diagnostic complexity. *Haematologica*, **2015**, 100, 42-8	6.6	81
19	Germline ETV6 mutations in familial thrombocytopenia and hematologic malignancy. *Nature Genetics*, **2015**, 47, 180-5	36.3	239
18	Validation and implementation of targeted capture and sequencing for the detection of actionable mutation, copy number variation, and gene rearrangement in clinical cancer specimens. *Journal of Molecular Diagnostics*, **2014**, 16, 56-67	5.1	203
17	Mutations in Twinkle primase-helicase cause Perrault syndrome with neurologic features. *Neurology*, **2014**, 83, 2054-61	6.5	73
16	A girl with West syndrome and autistic features harboring a de novo TBL1XR1 mutation. *Journal of Human Genetics*, **2014**, 59, 581-3	4.3	34
15	Complex MSH2 and MSH6 mutations in hypermutated microsatellite unstable advanced prostate cancer. *Nature Communications*, **2014**, 5, 4988	17.4	182

14	Mitochondrial serine protease HTRA2 p.G399S in a kindred with essential tremor and Parkinson disease. *Proceedings of the National Academy of Sciences of the United States of America*, **2014**, 111, 18285-90	11.5	96
13	A recurrent germline PAX5 mutation confers susceptibility to pre-B cell acute lymphoblastic leukemia. *Nature Genetics*, **2013**, 45, 1226-1231	36.3	205
12	Perrault syndrome is caused by recessive mutations in CLPP, encoding a mitochondrial ATP-dependent chambered protease. *American Journal of Human Genetics*, **2013**, 92, 605-13	11	152
11	Targeted genomic capture and massively parallel sequencing to identify genes for hereditary hearing loss in Middle Eastern families. *Genome Biology*, **2011**, 12, R89	18.3	163
10	Accurate and exact CNV identification from targeted high-throughput sequence data. *BMC Genomics*, **2011**, 12, 184	4.5	156
9	Detection of inherited mutations for breast and ovarian cancer using genomic capture and massively parallel sequencing. *Proceedings of the National Academy of Sciences of the United States of America*, **2010**, 107, 12629-33	11.5	372
8	Whole exome sequencing and homozygosity mapping identify mutation in the cell polarity protein GPSM2 as the cause of nonsyndromic hearing loss DFNB82. *American Journal of Human Genetics*, **2010**, 87, 90-4	11	223
7	Genomic duplication and overexpression of TJP2/ZO-2 leads to altered expression of apoptosis genes in progressive nonsyndromic hearing loss DFNA51. *American Journal of Human Genetics*, **2010**, 87, 101-9	11	82
6	Rare structural variants disrupt multiple genes in neurodevelopmental pathways in schizophrenia. *Science*, **2008**, 320, 539-43	33.3	1443
5	Genomic analysis of a heterogeneous Mendelian phenotype: multiple novel alleles for inherited hearing loss in the Palestinian population. *Human Genomics*, **2006**, 2, 203-11	6.8	39
4	Spectrum of mutations in BRCA1, BRCA2, CHEK2, and TP53 in families at high risk of breast cancer. *JAMA - Journal of the American Medical Association*, **2006**, 295, 1379-88	27.4	502
3	From fliesVeyes to our ears: mutations in a human class III myosin cause progressive nonsyndromic hearing loss DFNB30. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 7518-23	11.5	202
2	Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene ATOH1		1
1	Revisiting Genetic Testing for Patients with Negative Results: IPEX and FOXP3. *Journal of Clinical Immunology*,	5.7	