Terry-Lynn Young

List of Publications by Year
in descending order

39	1,426	430874	361022
papers	citations	18 h-index	35 g-index

42	42	42	1944
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Arrhythmogenic Right Ventricular Cardiomyopathy Type 5 Is a Fully Penetrant, Lethal Arrhythmic Disorder Caused by a Missense Mutation in the TMEM43 Gene. American Journal of Human Genetics, 2008, 82, 809-821.	6.2	431
2	Mutational spectrum of theWFS1 gene in Wolfram syndrome, nonsyndromic hearing impairment, diabetes mellitus, and psychiatric disease. Human Mutation, 2003, 22, 275-287.	2.5	160
3	A Fifth Locus for Bardet-Biedl Syndrome Maps to Chromosome 2q31. American Journal of Human Genetics, 1999, 64, 900-904.	6.2	117
4	The natural history of a genetic subtype ofÂarrhythmogenic right ventricular cardiomyopathy caused by a p.S358L mutation in <scp>TMEM43</scp>. Clinical Genetics, 2013, 83, 321-331.	2.0	72
5	Genetic Heterogeneity of Bardetâ€“Biedl Syndrome in a Distinct Canadian Population: Evidence for a Fifth Locus. Genomics, 1999, 55, 2-9.	2.9	63
6	The TMEM43 Newfoundland mutation p.S358L causing ARVC-5 was imported from Europe and increases the stiffness of the cell nucleus. European Heart Journal, 2015, 36, 872-881.	2.2	56
7	Canadian Bardet-Biedl syndrome family reduces the critical region of BBS3 (3p) and presents with a variable phenotype. American Journal of Medical Genetics Part A, 1998, 78, 461-467.	2.4	49
8	A Founder Effect in the Newfoundland Population Reduces the Bardet-Biedl Syndrome I (BBS1) Interval to 1 cM. American Journal of Human Genetics, 1999, 65, 1680-1687.	6.2	45
9	Recurrent missense mutations in TMEM43 (ARVD5) due to founder effects cause arrhythmogenic cardiomyopathies in the UK and Canada. European Heart Journal, 2013, 34, 1002-1011.	2.2	44
10	Long-Term Clinical Outcome of Arrhythmogenic Right Ventricular Cardiomyopathy in Individuals With a p.S358L Mutation in TMEM43 Following Implantable Cardioverter Defibrillator Therapy. Circulation: Arrhythmia and Electrophysiology, 2016, 9, .	4.8	37
11	Profound, prelingual nonsyndromic deafness maps to chromosome 10q21 and is caused by a novel missense mutation in the Usher syndrome type IF gene PCDH15. European Journal of Human Genetics, 2009, 17, 554-564.	2.8	33
12	A novel, non-stop mutation in FOXE3 causes an autosomal dominant form of variable anterior segment dysgenesis including Peters anomaly. European Journal of Human Genetics, 2011, 19, 293-299.	2.8	31
13	Characterization of a new full length TMPRSS3 isoform and identification of mutant alleles responsible for nonsyndromic recessive deafness in Newfoundland and Pakistan. BMC Medical Genetics, 2004, 5, 24.	2.1	30
14	Using nextâ€generation sequencing for the diagnosis of rare disorders: a family with retinitis pigmentosa and skeletal abnormalities. Journal of Pathology, 2011, 225, 12-18.	4.5	29
15	Variable neurologic phenotype in a GEFS+ family with a novel mutation in SCN1A. Seizure: the Journal of the British Epilepsy Association, 2009, 18, 492-497.	2.0	26
16	A Novel Deletion inSMPXCauses a Rare form of X-Linked Progressive Hearing Loss in Two Families Due to a Founder Effect. Human Mutation, 2013, 34, 66-69.	2.5	21
17	Translation of research discoveries to clinical care in arrhythmogenic right ventricular cardiomyopathy in Newfoundland and Labrador: Lessons for health policy in genetic disease. Genetics in Medicine, 2009, 11, 859-865.	2.4	18
18	Molecular Genetics of Achromatopsia in Newfoundland Reveal Genetic Heterogeneity, Founder Effects and the First Cases of Jalili Syndrome in North America. Ophthalmic Genetics, 2013, 34, 119-129.	1.2	18

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19	Exercise and arrhythmic risk in TMEM43 p.S358L arrhythmogenic right ventricular cardiomyopathy. Heart Rhythm, 2020, 17, 1159-1166.	0.7	16
20	Identification of a novel in-frame deletion in KCNQ4 (DFNA2A) and evidence of multiple phenocopies of unknown origin in a family with ADSNHL. European Journal of Human Genetics, 2013, 21, 1112-1119.	2.8	15
21	Autosomal recessive Bardetâ€“Biedl syndrome: first-degree relatives have no predisposition to metabolic and renal disorders. Kidney International, 2009, 76, 215-223.	5.2	14
22	A common variant in CLDN14 causes precipitous, prelingual sensorineural hearing loss in multiple families due to founder effect. Human Genetics, 2017, 136, 107-118.	3.8	14
23	â€˜It had to be doneâ€™: genetic testing decisions for arrhythmogenic right ventricular cardiomyopathy. Clinical Genetics, 2015, 88, 344-351.	2.0	13
24	X-Linked Hearing Loss: Two Gene Mutation Examples Provide Generalizable Implications for Clinical Care. American Journal of Audiology, 2014, 23, 190-200.	1.2	11
25	Novel Usher syndrome pathogenic variants identified in cases with hearing and vision loss. BMC Medical Genetics, 2019, 20, 68.	2.1	10
26	The genetic architecture of Stargardt macular dystrophy (STGD1): a longitudinal 40-year study in a genetic isolate. European Journal of Human Genetics, 2020, 28, 925-937.	2.8	10
27	A pathogenic deletion in Forkhead Box L1 (FOXL1) identifies the first otosclerosis (OTSC) gene. Human Genetics, 2022, 141, 965-979.	3.8	7
28	Autosomal dominant non-syndromic hearing loss maps to DFNA33 (13q34) and co-segregates with splice and frameshift variants in ATP11A, a phospholipid flippase gene. Human Genetics, 2022, 141, 431-444.	3.8	7
29	A dominant RAD51C pathogenic splicing variant predisposes to breast and ovarian cancer in the Newfoundland population due to founder effect. Molecular Genetics & Genomic Medicine, 2020, 8, e1070.	1.2	6
30	EFFECTS OF SEVERAL INSECT GROWTH REGULATORS ON EGG HATCH AND SUBSEQUENT DEVELOPMENT IN THE CABBAGE MAGGOT DELIA RADICUM (L.) (DIPTERA: ANTHOMYIIDAE). Canadian Entomologist, 1987, 119, 481-488.	0.8	5
31	High incidence of pediatric idiopathic epilepsy is associated with familial and autosomal dominant disease in Eastern Newfoundland. Epilepsy Research, 2012, 98, 140-147.	1.6	5
32	Perceived economic burden associated with an inherited cardiac condition: a qualitative inquiry with families affected by arrhythmogenic right ventricular cardiomyopathy. Genetics in Medicine, 2016, 18, 584-592.	2.4	5
33	Mutation of foxl1 Results in Reduced Cartilage Markers in a Zebrafish Model of Otosclerosis. Genes, 2022, 13, 1107.	2.4	4
34	Effect of the juvenoid methoprene on the hemolymph composition of the cabbage maggotDelia radicum (Diptera: Anthomyiidae). Experientia, 1987, 43, 902-903.	1.2	2
35	Psychological Distress and Quality of Life in Participants Undergoing Genetic Testing for Arrhythmogenic Right Ventricular Cardiomyopathy Caused by <i>TMEM43</i> p.S358L: Is It Time to Offer Population-Based Genetic Screening?. Public Health Genomics, 2021, 24, 253-260.	1.0	1
36	â€œSomething is just not right with my hearingâ€¦ early experiences of adults living with hearing loss. International Journal of Audiology, 2022, 61, 787-797.	1.7	1

#	Article	IF	Citations
37	Effects of Two Insect Growth Regulators on the Larval and Pupal Stages of the Cabbage Maggot (Diptera: Anthomyiidae). Journal of Economic Entomology, 1989, 82, 1040-1045.	1.8	0
38	Empirical and physiological assessment of in vitro growth in the mermithid nematode Romanomermis culicivorax. Canadian Journal of Zoology, 1990, 68, 511-516.	1.0	0
39	â€œThere are days I wish it wasnâ€™t there, and thereâ€™s days I realize Iâ€™m luckyâ€: A qualitative study of psychological sequelae to the implantable cardioverter defibrillator as a treatment for the prevention of sudden cardiac death in arrhythmogenic right ventricular cardiomyopathy. JRSM Cardiovascular Disease, 2017, 6, 204800401769861.	0.7	0