Nicholas Stong

List of Publications by Year
in descending order

58 papers	2,451 citations	236925 25 h-index	233421 45 g-index
68 all docs	68 docs citations	68 times ranked	5258 citing authors

#	Article	IF	Citations
1	Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples. Journal of Genetic Counseling, 2022, 31, 59-70.	1.6	3
2	Expanding the phenotypic spectrum of ARCN1-related syndrome. Genetics in Medicine, 2022, 24, 1227-1237.	2.4	5
3	Ancestry adjustment improves genome-wide estimates of regional intolerance. Genetics, 2022, , .	2.9	2
4	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	2.9	6
5	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
6	Family genetic result communication in rare and undiagnosed disease communities: Understanding the practice. Journal of Genetic Counseling, 2021, 30, 439-447.	1.6	4
7	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	2.4	18
8	Rare deleterious de novo missense variants in Rnf2/Ring2 are associated with a neurodevelopmental disorder with unique clinical features. Human Molecular Genetics, 2021, 30, 1283-1292.	2.9	17
9	Progressive cerebellar atrophy in a patient with complex II and III deficiency and a novel deleterious variant in SDHA: A Counseling Conundrum. Molecular Genetics & Genomic Medicine, 2021, 9, e1692.	1.2	1
10	<scp>TSPEAR</scp> variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study. American Journal of Medical Genetics, Part A, 2021, 185, 2417-2433.	1.2	10
11	De novo variants in <scp>TCF7L2</scp> are associated with a syndromic neurodevelopmental disorder. American Journal of Medical Genetics, Part A, 2021, 185, 2384-2390.	1.2	13
12	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13
13	One is the loneliest number: genotypic matchmaking using the electronic health record. Genetics in Medicine, 2021, 23, 1830-1832.	2.4	6
14	Integrative multi-omics identifies high risk multiple myeloma subgroup associated with significant DNA loss and dysregulated DNA repair and cell cycle pathways. BMC Medical Genomics, 2021, 14, 295.	1.5	5
15	DYRK1A pathogenic variants in two patients with syndromic intellectual disability and a review of the literature. Molecular Genetics & Genomic Medicine, 2020, 8, e1544.	1.2	8
16	Alternative transcripts in variant interpretation: the potential for missed diagnoses and misdiagnoses. Genetics in Medicine, 2020, 22, 1269-1275.	2.4	30
17	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	6.2	37
18	A pathogenic variant in the <scp>SETBP1</scp> hotspot results in a formeâ€fruste <scp>Schinzelâ€“Giedion</scp> syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1947-1951.	1.2	11

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19	Epileptic encephalopathy with features of rapidâ€onset dystonia Parkinsonism and alternating hemiplegia of childhood: a novel combination phenotype associated with ATP1A3 mutation. Epileptic Disorders, 2020, 22, 103-109.	1.3	4
20	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
21	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	6.2	43
22	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	1.5	6
23	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	6.2	29
24	Modification of the PROM1 disease phenotype by a mutation in ABCA4. Ophthalmic Genetics, 2019, 40, 369-375.	1.2	17
25	Whole-exome sequencing in the evaluation of fetal structural anomalies: a prospective cohort study. Lancet, The, 2019, 393, 758-767.	13.7	368
26	Improved Pathogenic Variant Localization via a Hierarchical Model of Sub-regional Intolerance. American Journal of Human Genetics, 2019, 104, 299-309.	6.2	29
27	Autism and developmental disability caused by KCNQ3 gainâ€ofâ€function variants. Annals of Neurology, 2019, 86, 181-192.	5.3	73
28	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. American Journal of Human Genetics, 2019, 104, 1127-1138.	6.2	59
29	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	2.5	19
30	IgG4â€related disease: Association with a rare gene variant expressed in cytotoxic T cells. Molecular Genetics & Genomic Medicine, 2019, 7, e686.	1.2	8
31	Whole genome sequencing reveals novel IGHMBP2 variant leading to unique cryptic spliceâ€site and Charcotâ€Marieâ€Tooth phenotype with early onset symptoms. Molecular Genetics & Genomic Medicine, 2019, 7, e00676.	1.2	18
32	A caseâ€"control collapsing analysis identifies retinal dystrophy genes associated with ophthalmic disease in patients with no pathogenic ABCA4 variants. Genetics in Medicine, 2019, 21, 2336-2344.	2.4	27
33	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. Epilepsia, 2019, 60, 797-806.	5.1	52
34	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
35	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
36	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59

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37	Late-onset pattern macular dystrophy mimicking ABCA4 and PRPH2 disease is caused by a homozygous frameshift mutation in ROM1. Journal of Physical Education and Sports Management, 2019, 5, a003624.	1.2	8
38	Heterozygous loss-of-function variants of MEIS2 cause a triad of palatal defects, congenital heart defects, and intellectual disability. European Journal of Human Genetics, 2019, 27, 278-290.	2.8	30
39	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	8.2	65
40	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	6.2	59
41	Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases. Genetics in Medicine, 2018, 20, 464-469.	2.4	42
42	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
43	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. PLoS Genetics, 2018, 14, e1007671.	3.5	16
44	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	5.3	44
45	Further evidence for the involvement of EFL1 in a Shwachmanâ€"Diamond-like syndrome and expansion of the phenotypic features. Journal of Physical Education and Sports Management, 2018, 4, a003046.	1.2	29
46	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
47	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. Human Molecular Genetics, 2018, 27, 2454-2465.	2.9	54
48	Exome sequencing of an adolescent with nonalcoholic fatty liver disease identifies a clinically actionable case of Wilson disease. Journal of Physical Education and Sports Management, 2018, 4, a003087.	1.2	3
49	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
50	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	6.2	35
51	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	6.2	181
52	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
53	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	6.2	43
54	A Novel Mutation in Junctional Plakoglobin Causing Lethal Congenital Epidermolysis Bullosa. Journal of Pediatrics, 2017, 191, 266-269.e1.	1.8	6

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55	Mapping H4K20me3 onto the chromatin landscape of senescent cells indicates a function in control of cell senescence and tumor suppression through preservation of genetic and epigenetic stability. Genome Biology, 2016, 17, 158.	8.8	65
56	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	6.2	68
57	Subtelomeric p53 binding prevents accumulation of <scp>DNA</scp> damage at human telomeres. EMBO Journal, 2016, 35, 193-207.	7.8	52
58	Subtelomeric CTCF and cohesin binding site organization using improved subtelomere assemblies and a novel annotation pipeline. Genome Research, 2014, 24, 1039-1050.	5.5	64