

Nicholas Stong

List of Publications by Year in descending order

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Version: 2024-02-01

58
papers

2,451
citations

270111

25
h-index

263392

45
g-index

68
all docs

68
docs citations

68
times ranked

5659
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples. <i>Journal of Genetic Counseling</i> , 2022, 31, 59-70.	0.9	3
2	Expanding the phenotypic spectrum of ARCN1-related syndrome. <i>Genetics in Medicine</i> , 2022, 24, 1227-1237.	1.1	5
3	Ancestry adjustment improves genome-wide estimates of regional intolerance. <i>Genetics</i> , 2022, , .	1.2	2
4	The microRNA processor <i>DROSHA</i> is a candidate gene for a severe progressive neurological disorder. <i>Human Molecular Genetics</i> , 2022, 31, 2934-2950.	1.4	6
5	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. <i>Genetics in Medicine</i> , 2021, 23, 653-660.	1.1	20
6	Family genetic result communication in rare and undiagnosed disease communities: Understanding the practice. <i>Journal of Genetic Counseling</i> , 2021, 30, 439-447.	0.9	4
7	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. <i>Genetics in Medicine</i> , 2021, 23, 259-271.	1.1	18
8	Rare deleterious <i>de novo</i> missense variants in <i>Rnf2/Ring2</i> are associated with a neurodevelopmental disorder with unique clinical features. <i>Human Molecular Genetics</i> , 2021, 30, 1283-1292.	1.4	17
9	Progressive cerebellar atrophy in a patient with complex II and III deficiency and a novel deleterious variant in SDHA: A Counseling Conundrum. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1692.	0.6	1
10	<i>TSPEAR</i> variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2417-2433.	0.7	10
11	De novo variants in <i>TCF7L2</i> are associated with a syndromic neurodevelopmental disorder. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2384-2390.	0.7	13
12	<i>CSNK2B</i> : A broad spectrum of neurodevelopmental disability and epilepsy severity. <i>Epilepsia</i> , 2021, 62, e103-e109.	2.6	13
13	One is the loneliest number: genotypic matchmaking using the electronic health record. <i>Genetics in Medicine</i> , 2021, 23, 1830-1832.	1.1	6
14	Integrative multi-omics identifies high risk multiple myeloma subgroup associated with significant DNA loss and dysregulated DNA repair and cell cycle pathways. <i>BMC Medical Genomics</i> , 2021, 14, 295.	0.7	5
15	<i>DYRK1A</i> pathogenic variants in two patients with syndromic intellectual disability and a review of the literature. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1544.	0.6	8
16	Alternative transcripts in variant interpretation: the potential for missed diagnoses and misdiagnoses. <i>Genetics in Medicine</i> , 2020, 22, 1269-1275.	1.1	30
17	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. <i>American Journal of Human Genetics</i> , 2020, 106, 570-583.	2.6	37
18	A pathogenic variant in the <i>SETBP1</i> hotspot results in a form of <i>Schinzel-Giedion</i> syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1947-1951.	0.7	11

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19	Epileptic encephalopathy with features of rapid-onset dystonia Parkinsonism and alternating hemiplegia of childhood: a novel combination phenotype associated with <i>ATP1A3</i> mutation. <i>Epileptic Disorders</i> , 2020, 22, 103-109.	0.7	4
20	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. <i>Genetics in Medicine</i> , 2019, 21, 161-172.	1.1	60
21	De Novo Variants in <i>WDR37</i> Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. <i>American Journal of Human Genetics</i> , 2019, 105, 413-424.	2.6	43
22	Magnetic Resonance Imaging characteristics in case of <i>TOR1AIP1</i> muscular dystrophy. <i>Clinical Imaging</i> , 2019, 58, 108-113.	0.8	6
23	De Novo Pathogenic Variants in <i>N-cadherin</i> Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. <i>American Journal of Human Genetics</i> , 2019, 105, 854-868.	2.6	29
24	Modification of the <i>PROM1</i> disease phenotype by a mutation in <i>ABCA4</i> . <i>Ophthalmic Genetics</i> , 2019, 40, 369-375.	0.5	17
25	Whole-exome sequencing in the evaluation of fetal structural anomalies: a prospective cohort study. <i>Lancet, The</i> , 2019, 393, 758-767.	6.3	368
26	Improved Pathogenic Variant Localization via a Hierarchical Model of Sub-regional Intolerance. <i>American Journal of Human Genetics</i> , 2019, 104, 299-309.	2.6	29
27	Autism and developmental disability caused by <i>KCNQ3</i> gain-of-function variants. <i>Annals of Neurology</i> , 2019, 86, 181-192.	2.8	73
28	Lysosomal Storage and Albinism Due to Effects of a De Novo <i>CLCN7</i> Variant on Lysosomal Acidification. <i>American Journal of Human Genetics</i> , 2019, 104, 1127-1138.	2.6	59
29	Heterozygous variants in <i>MYBPC1</i> are associated with an expanded neuromuscular phenotype beyond arthrogryposis. <i>Human Mutation</i> , 2019, 40, 1115-1126.	1.1	19
30	IgG4-related disease: Association with a rare gene variant expressed in cytotoxic T cells. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e686.	0.6	8
31	Whole genome sequencing reveals novel <i>IGHMBP2</i> variant leading to unique cryptic splice site and Charcot-Marie-Tooth phenotype with early onset symptoms. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00676.	0.6	18
32	A case-control collapsing analysis identifies retinal dystrophy genes associated with ophthalmic disease in patients with no pathogenic <i>ABCA4</i> variants. <i>Genetics in Medicine</i> , 2019, 21, 2336-2344.	1.1	27
33	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. <i>Epilepsia</i> , 2019, 60, 797-806.	2.6	52
34	Missense Variants in the Histone Acetyltransferase Complex Component Gene <i>TRRAP</i> Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	2.6	30
35	Bi-allelic Variants in <i>TONSL</i> Cause <i>SPONASTRIME</i> Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. <i>American Journal of Human Genetics</i> , 2019, 104, 422-438.	2.6	27
36	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in <i>SMARCC2</i> Cause a Syndrome with Intellectual Disability and Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 104, 164-178.	2.6	59

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37	Late-onset pattern macular dystrophy mimicking <i>ABCA4</i> and <i>PRPH2</i> disease is caused by a homozygous frameshift mutation in <i>ROM1</i> . <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a003624.	0.5	8
38	Heterozygous loss-of-function variants of <i>MEIS2</i> cause a triad of palatal defects, congenital heart defects, and intellectual disability. <i>European Journal of Human Genetics</i> , 2019, 27, 278-290.	1.4	30
39	<i>SSBP1</i> mutations cause mtDNA depletion underlying a complex optic atrophy disorder. <i>Journal of Clinical Investigation</i> , 2019, 130, 108-125.	3.9	65
40	Biallelic Mutations in <i>ATP5F1D</i> , which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. <i>American Journal of Human Genetics</i> , 2018, 102, 494-504.	2.6	59
41	Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases. <i>Genetics in Medicine</i> , 2018, 20, 464-469.	1.1	42
42	Loss of tubulin deglutamylase <i>CCP1</i> causes infantile-onset neurodegeneration. <i>EMBO Journal</i> , 2018, 37, .	3.5	86
43	De novo mutations in the GTP/GDP-binding region of <i>RALA</i> , a RAS-like small GTPase, cause intellectual disability and developmental delay. <i>PLoS Genetics</i> , 2018, 14, e1007671.	1.5	16
44	<i>NBEA</i> : Developmental disease gene with early generalized epilepsy phenotypes. <i>Annals of Neurology</i> , 2018, 84, 788-795.	2.8	44
45	Further evidence for the involvement of <i>EFL1</i> in a Shwachman-Diamond-like syndrome and expansion of the phenotypic features. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a003046.	0.5	29
46	<i>IRF2BPL</i> Is Associated with Neurological Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 103, 245-260.	2.6	69
47	Functional variants in <i>TBX2</i> are associated with a syndromic cardiovascular and skeletal developmental disorder. <i>Human Molecular Genetics</i> , 2018, 27, 2454-2465.	1.4	54
48	Exome sequencing of an adolescent with nonalcoholic fatty liver disease identifies a clinically actionable case of Wilson disease. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a003087.	0.5	3
49	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 185-192.	2.6	142
50	A Recurrent De Novo Variant in <i>NACC1</i> Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. <i>American Journal of Human Genetics</i> , 2017, 100, 343-351.	2.6	35
51	<i>MARRVEL</i> : Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. <i>American Journal of Human Genetics</i> , 2017, 100, 843-853.	2.6	181
52	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in <i>EBF3</i> . <i>American Journal of Human Genetics</i> , 2017, 100, 128-137.	2.6	96
53	De Novo Mutations in <i>PPP3CA</i> Cause Severe Neurodevelopmental Disease with Seizures. <i>American Journal of Human Genetics</i> , 2017, 101, 516-524.	2.6	43
54	A Novel Mutation in Junctional Plakoglobin Causing Lethal Congenital Epidermolysis Bullosa. <i>Journal of Pediatrics</i> , 2017, 191, 266-269.e1.	0.9	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. <i>Genome Biology</i> , 2016, 17, 158.	3.8	65
56	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. <i>American Journal of Human Genetics</i> , 2016, 99, 991-999.	2.6	68
57	Subtelomeric p53 binding prevents accumulation of <scp>DNA</scp> damage at human telomeres. <i>EMBO Journal</i> , 2016, 35, 193-207.	3.5	52
58	Subtelomeric CTCF and cohesin binding site organization using improved subtelomere assemblies and a novel annotation pipeline. <i>Genome Research</i> , 2014, 24, 1039-1050.	2.4	64