Yaping Yang

List of Publications by Year in descending order

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Version: 2024-02-01

109 papers	10,292 citations	53794 45 h-index	37204 96 g-index
115 all docs	115 docs citations	115 times ranked	17690 citing authors

#	Article	IF	Citations
1	Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders. New England Journal of Medicine, 2013, 369, 1502-1511.	27.0	1,717
2	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870.	7.4	1,171
3	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	27.0	565
4	Performance of ACMG-AMP Variant-Interpretation Guidelines among Nine Laboratories in the Clinical Sequencing Exploratory Research Consortium. American Journal of Human Genetics, 2016, 98, 1067-1076.	6.2	432
5	Diagnostic Yield of Clinical Tumor and Germline Whole-Exome Sequencing for Children With Solid Tumors. JAMA Oncology, 2016, 2, 616.	7.1	378
6	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
7	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245.	2.9	261
8	Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. Nature Communications, 2016, 7, 10713.	12.8	227
9	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	27.0	205
10	Molecular diagnostic experience of whole-exome sequencing in adult patients. Genetics in Medicine, 2016, 18, 678-685.	2.4	186
11	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	8.2	184
12	Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. Nature Genetics, 2015, 47, 926-932.	21.4	166
13	Non-invasive prenatal sequencing for multiple Mendelian monogenic disorders using circulating cell-free fetal DNA. Nature Medicine, 2019, 25, 439-447.	30.7	160
14	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	6.2	146
15	Exome sequencing resolves apparent incidental findings and reveals further complexity of SH3TC2 variant alleles causing Charcot-Marie-Tooth neuropathy. Genome Medicine, 2013, 5, 57.	8.2	143
16	Rapid Targeted Next-Generation Sequencing Platform for Molecular Screening and Clinical Genotyping in Subjects with Hemoglobinopathies. EBioMedicine, 2017, 23, 150-159.	6.1	138
17	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
18	De novo truncating mutations in ASXL3 are associated with a novel clinical phenotype with similarities to Bohring-Opitz syndrome. Genome Medicine, 2013, 5, 11.	8.2	128

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19	Mutations in signal recognition particle SRP54 cause syndromic neutropenia with Shwachman-Diamondâ€“like features. Journal of Clinical Investigation, 2017, 127, 4090-4103.	8.2	126
20	Heterozygous De Novo and Inherited Mutations in the Smooth Muscle Actin (ACTG2) Gene Underlie Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome. PLoS Genetics, 2014, 10, e1004258.	3.5	122
21	De Novo Mutations in CHD4 , an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. American Journal of Human Genetics, 2016, 99, 934-941.	6.2	111
22	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. Genome Medicine, 2018, 10, 74.	8.2	105
23	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	6.2	98
24	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52.	2.4	94
25	Mutations in PURA Cause Profound Neonatal Hypotonia, Seizures, and Encephalopathy in 5q31.3 Microdeletion Syndrome. American Journal of Human Genetics, 2014, 95, 579-583.	6.2	92
26	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
27	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
28	Exome sequencing in mostly consanguineous Arab families with neurologic disease provides a high potential molecular diagnosis rate. BMC Medical Genomics, 2016, 9, 42.	1.5	80
29	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	3.5	80
30	Evidence for feasibility of fetal trophoblastic cellâ€based noninvasive prenatal testing. Prenatal Diagnosis, 2016, 36, 1009-1019.	2.3	78
31	Whole-exome sequencing in the molecular diagnosis of individuals with congenital anomalies of the kidney and urinary tract and identification of a new causative gene. Genetics in Medicine, 2017, 19, 412-420.	2.4	73
32	The next generation of population-based spinal muscular atrophy carrier screening: comprehensive pan-ethnic SMN1 copy-number and sequence variant analysis by massively parallel sequencing. Genetics in Medicine, 2017, 19, 936-944.	2.4	70
33	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
34	A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. Genetics in Medicine, 2017, 19, 575-582.	2.4	68
35	Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. American Journal of Human Genetics, 2016, 98, 562-570.	6.2	66
36	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	6.2	66

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37	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. American Journal of Human Genetics, 2015, 97, 904-913.	6.2	65
38	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	6.2	61
39	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	6.2	59
40	De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. American Journal of Human Genetics, 2014, 94, 784-789.	6.2	57
41	Bi-allelic Mutations in PKD1L1 Are Associated with Laterality Defects in Humans. American Journal of Human Genetics, 2016, 99, 886-893.	6.2	57
42	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. American Journal of Human Genetics, 2018, 103, 154-162.	6.2	56
43	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. Genome Medicine, 2019, 11, 48.	8.2	55
44	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	6.2	54
45	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	2.4	52
46	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	8.2	50
47	A Founder Mutation in VPS11 Causes an Autosomal Recessive Leukoencephalopathy Linked to Autophagic Defects. PLoS Genetics, 2016, 12, e1005848.	3.5	50
48	Reproductive genetic counseling challenges associated with diagnostic exome sequencing in a large academic private reproductive genetic counseling practice. Prenatal Diagnosis, 2015, 35, 1022-1029.	2.3	47
49	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. American Journal of Human Genetics, 2016, 99, 720-727.	6.2	45
50	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106.	8.2	43
51	KCTD7 deficiency defines a distinct neurodegenerative disorder with a conserved autophagyâ€lysosome defect. Annals of Neurology, 2018, 84, 766-780.	5.3	42
52	Reliable detection of subchromosomal deletions and duplications using cellâ€free based noninvasive prenatal testing. Prenatal Diagnosis, 2018, 38, 1069-1078.	2.3	42
53	Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing. Journal of Genetic Counseling, 2019, 28, 1107-1118.	1.6	42
54	Loss-of-Function Mutations in FRRS1L Lead to an Epileptic-Dyskinetic Encephalopathy. American Journal of Human Genetics, 2016, 98, 1249-1255.	6.2	40

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55	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment GeneCHAMP1Cause Syndromic Intellectual Disability. Human Mutation, 2016, 37, 354-358.	2.5	40
56	Germline mutations in ABL1 cause an autosomal dominant syndrome characterized by congenital heart defects and skeletal malformations. Nature Genetics, 2017, 49, 613-617.	21.4	40
57	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. Genome Medicine, 2017, 9, 73.	8.2	39
58	A visual and curatorial approach to clinical variant prioritization and disease gene discovery in genome-wide diagnostics. Genome Medicine, 2016, 8, 13.	8.2	37
59	Haploinsufficiency of the E3 ubiquitin-protein ligase gene TRIP12 causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. Human Genetics, 2017, 136, 377-386.	3.8	36
60	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	2.4	36
61	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
62	Interpreting Incidentally Identified Variants in Genes Associated With Catecholaminergic Polymorphic Ventricular Tachycardia in a Large Cohort of Clinical Whole-Exome Genetic Test Referrals. Circulation: Arrhythmia and Electrophysiology, 2017, 10, .	4.8	35
63	Integrated analysis of metabolomic profiling and exome data supplements sequence variant interpretation, classification, and diagnosis. Genetics in Medicine, 2020, 22, 1560-1566.	2.4	35
64	Clinical and molecular characterization of de novo loss of function variants in HNRNPU. American Journal of Medical Genetics, Part A, 2017, 173, 2680-2689.	1.2	34
65	Genotype-phenotype correlations in individuals with pathogenicREREvariants. Human Mutation, 2018, 39, 666-675.	2.5	34
66	FHF1 (FGF12) epileptic encephalopathy. Neurology: Genetics, 2016, 2, e115.	1.9	32
67	Improvement of regressive autism symptoms in a child with TMLHE deficiency following carnitine supplementation. American Journal of Medical Genetics, Part A, 2015, 167, 2162-2167.	1.2	30
68	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	6.2	30
69	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
70	Low-level parental somatic mosaic SNVs in exomes from a large cohort of trios with diverse suspected Mendelian conditions. Genetics in Medicine, 2020, 22, 1768-1776.	2.4	30
71	Heterozygous variants inACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. Human Mutation, 2017, 38, 1365-1371.	2.5	27
72	Congenital heart defects and left ventricular non-compaction in males with loss-of-function variants inNONO. Journal of Medical Genetics, 2017, 54, 47-53.	3.2	24

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73	Whole exome sequencing identifies the first STRADA point mutation in a patient with polyhydramnios, megalencephaly, and symptomatic epilepsy syndrome (PMSE). American Journal of Medical Genetics, Part A, 2016, 170, 2181-2185.	1.2	23
74	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	8.2	23
75	Comparison of three whole genome amplification methods for detection of genomic aberrations in single cells. Prenatal Diagnosis, 2016, 36, 823-830.	2.3	22
76	Lysinuric protein intolerance presenting with multiple fractures. Molecular Genetics and Metabolism Reports, 2014, 1, 176-183.	1.1	20
77	Homozygous variants in pyrrolineâ€5â€carboxylate reductase 2 (PYCR2) in patients with progressive microcephaly and hypomyelinating leukodystrophy. American Journal of Medical Genetics, Part A, 2017, 173, 460-470.	1.2	20
78	Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review. American Journal of Medical Genetics, Part A, 2019, 179, 966-977.	1.2	20
79	DYRK1A-related intellectual disability: a syndrome associated with congenital anomalies of the kidney and urinary tract. Genetics in Medicine, 2019, 21, 2755-2764.	2.4	19
80	Atlas-CNV: a validated approach to call single-exon CNVs in the eMERGESeq gene panel. Genetics in Medicine, 2019, 21, 2135-2144.	2.4	19
81	Biallelic loss-of-function P4HTM gene variants cause hypotonia, hypoventilation, intellectual disability, dysautonomia, epilepsy, and eye abnormalities (HIDEA syndrome). Genetics in Medicine, 2019, 21, 2355-2363.	2.4	19
82	Case report and novel treatment of an autosomal recessive Leigh syndrome caused by shortâ€chain enoylâ€CoA hydratase deficiency. American Journal of Medical Genetics, Part A, 2019, 179, 803-807.	1.2	18
83	A de novo mutation in the X-linked PAK3 gene is the underlying cause of intellectual disability and macrocephaly in monozygotic twins. European Journal of Medical Genetics, 2017, 60, 212-216.	1.3	17
84	Further delineation of the phenotypic spectrum associated with hemizygous lossâ€ofâ€function variants in NONO. American Journal of Medical Genetics, Part A, 2020, 182, 652-658.	1.2	17
85	CRIPT exonic deletion and a novel missense mutation in a female with short stature, dysmorphic features, microcephaly, and pigmentary abnormalities. American Journal of Medical Genetics, Part A, 2016, 170, 2206-2211.	1.2	16
86	Increased bone turnover, osteoporosis, progressive tibial bowing, fractures, and scoliosis in a patient with a finalâ€exon SATB2 frameshift mutation. American Journal of Medical Genetics, Part A, 2016, 170, 3028-3032.	1.2	16
87	Increased diagnostic yield by reanalysis of data from a hearing loss gene panel. BMC Medical Genomics, 2019, 12, 76.	1.5	16
88	Atypical presentation of moyamoya disease in an infant with a de novo RNF213 variant. American Journal of Medical Genetics, Part A, 2015, 167, 2742-2747.	1.2	15
89	Biallelic loss of function variants in PPP1R21 cause a neurodevelopmental syndrome with impaired endocytic function. Human Mutation, 2019, 40, 267-280.	2.5	15
90	Approaches to carrier testing and results disclosure in translational genomics research: The clinical sequencing exploratory research consortium experience. Molecular Genetics & Genomic Medicine, 2018, 6, 898-909.	1.2	15

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91	<scp>Wolffâ€“Parkinsonâ€“White</scp> syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. American Journal of Medical Genetics, Part A, 2020, 182, 1387-1399.	1.2	14
92	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
93	Incidentally identified genetic variants in arrhythmogenic right ventricular cardiomyopathyâ€associated genes among children undergoing exome sequencing reflect healthy population variation. Molecular Genetics & Genomic Medicine, 2019, 7, e593.	1.2	13
94	Novel Missense Variants in ADAT3 as a Cause of Syndromic Intellectual Disability. Journal of Pediatric Genetics, 2019, 08, 244-251.	0.7	13
95	Value of Exome Sequencing in Diagnosis and Management of Recurrent Non-immune Hydrops Fetalis: A Retrospective Analysis. Frontiers in Genetics, 2021, 12, 616392.	2.3	13
96	A toolkit for genetics providers in followâ€up of patients with nonâ€diagnostic exome sequencing. Journal of Genetic Counseling, 2019, 28, 213-228.	1.6	11
97	BAZ2B haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. Human Mutation, 2020, 41, 921-925.	2.5	11
98	Characterization of the renal phenotype in RMND1 â€related mitochondrial disease. Molecular Genetics & Genomic Medicine, 2019, 7, e973.	1.2	10
99	The expanding clinical phenotype of germline ABL1 â€associated congenital heart defects and skeletal malformations syndrome. Human Mutation, 2020, 41, 1738-1744.	2.5	10
100	Renal cell carcinoma harboring somatic TSC2 mutations in a child with methylmalonic acidemia. Pediatric Blood and Cancer, 2017, 64, e26286.	1.5	9
101	Heterozygous variants in SPTBN1 cause intellectual disability and autism. American Journal of Medical Genetics, Part A, 2021, 185, 2037-2045.	1.2	9
102	De novo apparent loss-of-function mutations in PRR12 in three patients with intellectual disability and iris abnormalities. Human Genetics, 2018, 137, 257-264.	3.8	8
103	Phenotypic expansion of the <scp>BPTF</scp>â€related neurodevelopmental disorder with dysmorphic facies and distal limb anomalies. American Journal of Medical Genetics, Part A, 2021, 185, 1366-1378.	1.2	8
104	Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma. Neurology: Genetics, 2018, 4, e248.	1.9	7
105	<scp>PPP3CA</scp> truncating variants clustered in the regulatory domain cause earlyâ€onset refractory epilepsy. Clinical Genetics, 2021, 100, 227-233.	2.0	7
106	Methionyl-tRNA Formyltransferase (MTFMT) Deficiency Mimicking Acquired Demyelinating Disease. Journal of Child Neurology, 2016, 31, 215-219.	1.4	5
107	<scp>Genotypeâ€phenotype</scp> study and expansion of <scp>ARL6IP1â€related</scp> complicated hereditary spastic paraplegia. Clinical Genetics, 2021, 99, 477-480.	2.0	3
108	An Unusual Cause of Peroneal Neuropathy. Seminars in Pediatric Neurology, 2014, 21, 77-81.	2.0	2

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109	Signal-to-Noise Analysis Can Inform the Likelihood That Incidentally Identified Variants in Sarcomeric Genes Are Associated with Pediatric Cardiomyopathy. Journal of Personalized Medicine, 2022, 12, 733.	2.5	1