Dong Li

List of Publications by Year
in descending order

124 papers	8,246 citations	101543 36 h-index	51608 86 g-index
133 all docs	133 docs citations	133 times ranked	14885 citing authors

#	Article	IF	Citations
1	The diploid genome sequence of an Asian individual. Nature, 2008, 456, 60-65.	27.8	834
2	Aegilops tauschii draft genome sequence reveals a gene repertoire for wheat adaptation. Nature, 2013, 496, 91-95.	27.8	714
3	Draft genome of the wheat A-genome progenitor Triticum urartu. Nature, 2013, 496, 87-90.	27.8	700
4	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. Nature Genetics, 2019, 51, 1207-1214.	21.4	641
5	Genome-wide patterns of genetic variation among elite maize inbred lines. Nature Genetics, 2010, 42, 1027-1030.	21.4	439
6	Single baseâ€“resolution methylome of the silkworm reveals a sparse epigenomic map. Nature Biotechnology, 2010, 28, 516-520.	17.5	349
7	Complete Resequencing of 40 Genomes Reveals Domestication Events and Genes in Silkworm () Tj ETQq1 1 0.784314 rgBT /Overlock 1	12.6	342
8	Whole-genome sequencing of giant pandas provides insights into demographic history and local adaptation. Nature Genetics, 2013, 45, 67-71.	21.4	303
9	A genome-wide association study of anorexia nervosa. Molecular Psychiatry, 2014, 19, 1085-1094.	7.9	282
10	Draft genome sequence of the mulberry tree Morus notabilis. Nature Communications, 2013, 4, 2445.	12.8	277
11	Meta-analysis of shared genetic architecture across ten pediatric autoimmune diseases. Nature Medicine, 2015, 21, 1018-1027.	30.7	212
12	Molecular footprints of domestication and improvement in soybean revealed by whole genome re-sequencing. BMC Genomics, 2013, 14, 579.	2.8	186
13	Mutations in PDGFRB Cause Autosomal-Dominant Infantile Myofibromatosis. American Journal of Human Genetics, 2013, 92, 1001-1007.	6.2	174
14	GRIN2D Recurrent De Novo Dominant Mutation Causes a Severe Epileptic Encephalopathy Treatable with NMDA Receptor Channel Blockers. American Journal of Human Genetics, 2016, 99, 802-816.	6.2	138
15	ARAF recurrent mutation causes central conducting lymphatic anomaly treatable with a MEK inhibitor. Nature Medicine, 2019, 25, 1116-1122.	30.7	136
16	Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. Biological Psychiatry, 2022, 91, 313-327.	1.3	114
17	MicroRNAs of Bombyx mori identified by Solexa sequencing. BMC Genomics, 2010, 11, 148.	2.8	107
18	Autosomal Dominant Hypoparathyroidism Caused by Germline Mutation inGNA11: Phenotypic and Molecular Characterization. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1774-E1783.	3.6	79

#	Article	IF	Citations
19	CYP3A4 mutation causes vitamin Dâ€“dependent rickets type 3. Journal of Clinical Investigation, 2018, 128, 1913-1918.	8.2	77
20	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular Genetics and Metabolism, 2015, 114, 388-396.	1.1	76
21	Pathogenic variant in EPHB4 results in central conducting lymphatic anomaly. Human Molecular Genetics, 2018, 27, 3233-3245.	2.9	73
22	CYP3A4 Induction by Rifampin: An Alternative Pathway for Vitamin D Inactivation in Patients With CYP24A1 Mutations. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1440-1446.	3.6	72
23	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. Clinical Epigenetics, 2019, 11, 64.	4.1	71
24	KAT6A Syndrome: genotypeâ€“phenotype correlation in 76 patients with pathogenic KAT6A variants. Genetics in Medicine, 2019, 21, 850-860.	2.4	68
25	Genetic sharing and heritability of paediatric age of onset autoimmune diseases. Nature Communications, 2015, 6, 8442.	12.8	58
26	AGC1 Deficiency Causes Infantile Epilepsy, Abnormal Myelination, and Reduced N-Acetylaspartate. JIMD Reports, 2014, 14, 77-85.	1.5	57
27	Mutation in IRF2BP2 is responsible for a familial form of common variable immunodeficiency disorder. Journal of Allergy and Clinical Immunology, 2016, 138, 544-550.e4.	2.9	54
28	HDL (High-Density Lipoprotein) Metrics and Atherosclerotic Risk in Women. Arteriosclerosis, Thrombosis, and Vascular Biology, 2018, 38, 2236-2244.	2.4	52
29	Kaposiform lymphangiomatosis effectively treated with <scp>MEK</scp> inhibition. EMBO Molecular Medicine, 2020, 12, e12324.	6.9	51
30	Mutations in TBCK, Encoding TBC1-Domain-Containing Kinase, Lead to a Recognizable Syndrome of Intellectual Disability and Hypotonia. American Journal of Human Genetics, 2016, 98, 782-788.	6.2	50
31	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. American Journal of Human Genetics, 2019, 105, 283-301.	6.2	46
32	Metabolic Profiling and Transcriptome Analysis of Mulberry Leaves Provide Insights into Flavonoid Biosynthesis. Journal of Agricultural and Food Chemistry, 2020, 68, 1494-1504.	5.2	45
33	USMG5 Ashkenazi Jewish founder mutation impairs mitochondrial complex V dimerization and ATP synthesis. Human Molecular Genetics, 2018, 27, 3305-3312.	2.9	45
34	Monoallelic BMP2 Variants Predicted to Result in Haploinsufficiency Cause Craniofacial, Skeletal, and Cardiac Features Overlapping Those of 20p12 Deletions. American Journal of Human Genetics, 2017, 101, 985-994.	6.2	44
35	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	10.3	43
36	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42

#	Article	IF	Citations
37	Genetic diversity, molecular phylogeny and selection evidence of the silkworm mitochondria implicated by complete resequencing of 41 genomes. BMC Evolutionary Biology, 2010, 10, 81.	3.2	40
38	Mutations inSPECC1L, encoding sperm antigen with calponin homology and coiled-coil domains 1-like, are found in some cases of autosomal dominant Opitz G/BBB syndrome. Journal of Medical Genetics, 2015, 52, 104-110.	3.2	40
39	De novo variants in Myelin regulatory factor (MYRF) as candidates of a new syndrome of cardiac and urogenital anomalies. American Journal of Medical Genetics, Part A, 2018, 176, 969-972.	1.2	39
40	Mutations in topoisomerase IIÎ² result in a B cell immunodeficiency. Nature Communications, 2019, 10, 3644.	12.8	37
41	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. American Journal of Human Genetics, 2020, 107, 164-172.	6.2	37
42	Digenic Inheritance of PROKR2 and WDR11 Mutations in Pituitary Stalk Interruption Syndrome. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 2501-2507.	3.6	36
43	Exome Sequencing Reveals Mutations in AIRE as a Cause of Isolated Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1726-1733.	3.6	35
44	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
45	Late-onset hereditary hypophosphatemic rickets with hypercalciuria (HHRH) due to mutation of SLC34A3/NPT2c. Bone, 2017, 97, 15-19.	2.9	30
46	Shared genetic risk between eating disorderâ€•and substanceâ€useâ€related phenotypes: Evidence from genomeâ€wide association studies. Addiction Biology, 2021, 26, e12880.	2.6	28
47	Short stature and hypoparathyroidism in a child with Kenny-Caffey syndrome type 2 due to a novel mutation in FAM111A gene. International Journal of Pediatric Endocrinology (Springer), 2017, 2017, 1.	1.6	27
48	Expanding the SPECC1L mutation phenotypic spectrum to include Teebi hypertelorism syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2497-2502.	1.2	26
49	Association of Mutations in SLC12A1 Encoding the NKCC2 Cotransporter With Neonatal Primary Hyperparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2196-2200.	3.6	25
50	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	2.4	25
51	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. European Journal of Human Genetics, 2020, 28, 1422-1431.	2.8	25
52	Phenotypic spectrum associated with SPECC1L pathogenic variants: new families and critical review of the nosology of Teebi, Opitz GBBB, and Baraitser-Winter syndromes. European Journal of Medical Genetics, 2019, 62, 103588.	1.3	24
53	Identification and characterization of Sox genes in the silkworm, Bombyx mori. Molecular Biology Reports, 2011, 38, 3573-3584.	2.3	23
54	A genome-wide association study of anorexia nervosa suggests a risk locus implicated in dysregulated leptin signaling. Scientific Reports, 2017, 7, 3847.	3.3	23

#	Article	IF	Citations
55	Activating variants in <scp>PDGFRB</scp> result in a spectrum of disorders responsive to imatinib monotherapy. American Journal of Medical Genetics, Part A, 2020, 182, 1576-1591.	1.2	21
56	Novel truncating mutations in CTNND1 cause a dominant craniofacial and cardiac syndrome. Human Molecular Genetics, 2020, 29, 1900-1921.	2.9	21
57	Flavones Produced by Mulberry Flavone Synthase Type I Constitute a Defense Line against the Ultraviolet-B Stress. Plants, 2020, 9, 215.	3.5	21
58	SMARCE1, a rare cause of Coffinâ€“Siris Syndrome: Clinical description of three additional cases. American Journal of Medical Genetics, Part A, 2016, 170, 1967-1973.	1.2	18
59	Digenic Heterozygous Mutations in SLC34A3 and SLC34A1 Cause Dominant Hypophosphatemic Rickets with Hypercalciuria. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 2392-2400.	3.6	18
60	Enhanced B Cell Alloantigen Presentation and Its Epigenetic Dysregulation in Liver Transplant Rejection. American Journal of Transplantation, 2016, 16, 497-508.	4.7	17
61	Pathogenic variants in SMARCA5 , a chromatin remodeler, cause a range of syndromic neurodevelopmental features. Science Advances, 2021, 7, .	10.3	17
62	Delayed diagnosis of congenital myasthenia due to associated mitochondrial enzyme defect. Neuromuscular Disorders, 2015, 25, 257-261.	0.6	16
63	EP300 â€related Rubinsteinâ€“Taybi syndrome: Highlighted rare phenotypic findings and a genotypeâ€“phenotype metaâ€analysis of 74 patients. American Journal of Medical Genetics, Part A, 2020, 182, 2926-2938.	1.2	16
64	De novo loss-of-function variants in X-linked MED12 are associated with Hardikar syndrome in females. Genetics in Medicine, 2021, 23, 637-644.	2.4	16
65	Chromosome restructuring and number change during the evolution of Morus notabilis and Morus alba. Horticulture Research, 2022, 9, .	6.3	16
66	Treatment of severe Kaposiform lymphangiomatosis positive for NRAS mutation by MEK inhibition. Pediatric Research, 2023, 94, 1911-1915.	2.3	16
67	Coronary Artery Calcium on Noncontrast Thoracic Computerized Tomography Scans and All-Cause Mortality. Circulation, 2018, 138, 2437-2438.	1.6	15
68	Heterozygous Mutations in TBX1 as a Cause of Isolated Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 4023-4032.	3.6	15
69	Association of Rare Recurrent Copy Number Variants With Congenital Heart Defects Based on Next-Generation Sequencing Data From Family Trios. Frontiers in Genetics, 2019, 10, 819.	2.3	15
70	The variability of <scp>SMARCA4</scp>â€related <scp>Coffinâ€“Siris</scp> syndrome: Do nonsense candidate variants add to milder phenotypes?. American Journal of Medical Genetics, Part A, 2020, 182, 2058-2067.	1.2	14
71	MMHub, a database for the mulberry metabolome. Database: the Journal of Biological Databases and Curation, 2020, 2020, .	3.0	14
72	Exome sequencing reveals a nonsense mutation in MMP13 as a new cause of autosomal recessive metaphyseal anadysplasia. European Journal of Human Genetics, 2015, 23, 264-266.	2.8	13

#	Article	IF	Citations
73	Biliary-Atresia-Associated Mannosidase-1-Alpha-2 Gene Regulates Biliary and Ciliary Morphogenesis and Laterality. Frontiers in Physiology, 2020, 11, 538701.	2.8	13
74	Association of a rare NOTCH4 coding variant with systemic sclerosis: a family-based whole exome sequencing study. BMC Musculoskeletal Disorders, 2016, 17, 462.	1.9	12
75	High-throughput Molecular Analysis of Pseudohypoparathyroidism 1b Patients Reveals Novel Genetic and Epigenetic Defects. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e4603-e4620.	3.6	12
76	Expanding the phenotype of <scp>ASXL3</scp>â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	1.2	12
77	De novo variants in CACNA1E found in patients with intellectual disability, developmental regression and social cognition deficit but no seizures. Molecular Autism, 2021, 12, 69.	4.9	12
78	Safety and Effectiveness of JuvÃ©derm Ultra Plus Injectable Gel in Correcting Severe Nasolabial Folds in Chinese Subjects. Plastic and Reconstructive Surgery - Global Open, 2017, 5, e1133.	0.6	11
79	Expanding the phenotypic spectrum of TP63â€related disorders including the first set of monozygotic twins. American Journal of Medical Genetics, Part A, 2018, 176, 75-81.	1.2	11
80	Missense Mutations in NKAP Cause a Disorder of Transcriptional Regulation Characterized by Marfanoid Habitus and Cognitive Impairment. American Journal of Human Genetics, 2019, 105, 987-995.	6.2	11
81	Heterozygous de novo variants in <scp>CSNK1G1</scp> are associated with syndromic developmental delay and autism spectrum disorder. Clinical Genetics, 2020, 98, 571-576.	2.0	10
82	Rare Recurrent Variants in Noncoding Regions Impact Attention-Deficit Hyperactivity Disorder (ADHD) Gene Networks in Children of both African American and European American Ancestry. Genes, 2021, 12, 310.	2.4	10
83	The chromatin remodeler ISWI acts during Drosophila development to regulate adult sleep. Science Advances, 2021, 7, .	10.3	9
84	ALG13 Xâ€linked intellectual disability: New variants, glycosylation analysis, and expanded phenotypes. Journal of Inherited Metabolic Disease, 2021, 44, 1001-1012.	3.6	9
85	Increasing diagnostic yield by RNA-Sequencing in rare diseaseâ€"bypass hurdles of interpreting intronic or splice-altering variants. Annals of Translational Medicine, 2018, 6, 126-126.	1.7	9
86	Genetics etiologies and genotype phenotype correlations in a cohort of individuals with central conducting lymphatic anomaly. European Journal of Human Genetics, 2022, 30, 1022-1028.	2.8	9
87	Experiences with offering pro bono medical genetics services in the West Indies: Benefits to patients, physicians, and the community. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 1030-1041.	1.6	8
88	Expanding the clinical and phenotypic heterogeneity associated with biallelic variants in ACO2. Annals of Clinical and Translational Neurology, 2020, 7, 1013-1028.	3.7	8
89	A new syndrome of moyamoya disease, kidney dysplasia, aminotransferase elevation, and skin disease associated withÂde novo variants in <scp>RNF213</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 2168-2174.	1.2	8
90	BmSE, a SINE family with 3â€² ends of (ATTT) repeats in domesticated silkworm (Bombyx mori). Journal of Genetics and Genomics, 2010, 37, 125-135.	3.9	7

#	Article	IF	Citations
91	Value of whole exome sequencing for syndromic retinal dystrophy diagnosis in young patients. Clinical and Experimental Ophthalmology, 2015, 43, 132-138.	2.6	7
92	High-Quality Statin Trials Support the 2013 American College of Cardiology/American Heart Association Cholesterol Guidelines After the HOPE-3 Trial (Heart Outcomes Prevention Evaluation-3): MESA (The Multiethnic Study of Atherosclerosis). Circulation, 2017, 136, 1863-1865.	1.6	7
93	Extension of the mutational and clinical spectrum of SOX2 related disorders: Description of six new cases and a novel association with suprasellar teratoma. American Journal of Medical Genetics, Part A, 2018, 176, 2710-2719.	1.2	7
94	Isolated vocal cord paralysis in two siblings with compound heterozygous variants inMUSK: Expanding the phenotypic spectrum. American Journal of Medical Genetics, Part A, 2019, 179, 655-658.	1.2	7
95	Clinical variability of TUBB â€associated disorders: Diagnosis through reanalysis. American Journal of Medical Genetics, Part A, 2020, 182, 3035-3039.	1.2	7
96	Expanded phenotypic spectrum of <scp>JAG1</scp>â€associated diseases: Central conducting lymphatic anomaly with a pathogenic variant in <scp>JAG1</scp>. Clinical Genetics, 2021, 99, 742-743.	2.0	7
97	Ciliopathies: Coloring outside of the lines. American Journal of Medical Genetics, Part A, 2021, 185, 687-694.	1.2	7
98	PRKACB variants in skeletal disease or adrenocortical hyperplasia: effects on protein kinase A. Endocrine-Related Cancer, 2020, 27, 647-656.	3.1	7
99	Early Infantile Epileptic Encephalopathy in anSTXBP1Patient with Lactic Acidemia and Normal Mitochondrial Respiratory Chain Function. Case Reports in Genetics, 2016, 2016, 1-5.	0.2	6
100	Heterozygous Deletion Impacting SMARCAD1 in the Original Kindred with Absent Dermatoglyphs and Associated Features (Baird, 1964). Journal of Pediatrics, 2018, 194, 248-252.e2.	1.8	6
101	Proteomic Profiling Reveals Roles of Stress Response, Ca²⁺ Transient Dysregulation, and Novel Signaling Pathways in Alcoholâ€induced Cardiotoxicity. Alcoholism: Clinical and Experimental Research, 2020, 44, 2187-2199.	2.4	6
102	A DNA repair disorder caused by de novo monoallelic DDB1 variants is associated with a neurodevelopmental syndrome. American Journal of Human Genetics, 2021, 108, 749-756.	6.2	6
103	Intragenic Deletions of GNAS in Pseudohypoparathyroidism Type 1A Identify a New Region Affecting Methylation of Exon A/B. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e3197-e3206.	3.6	6
104	Variants in ADD1 cause intellectual disability, corpus callosum dysgenesis, and ventriculomegaly in humans. Genetics in Medicine, 2022, 24, 319-331.	2.4	6
105	Non-coding structural variation differentially impacts attention-deficit hyperactivity disorder (ADHD) gene networks in African American vs Caucasian children. Scientific Reports, 2020, 10, 15252.	3.3	5
106	A novel heterotaxy gene: Expansion of the phenotype of TTC21B â€spectrum disease. American Journal of Medical Genetics, Part A, 2021, 185, 1266-1269.	1.2	5
107	Variants in PHF8 cause a spectrum of X-linked neurodevelopmental disorders and facial dysmorphology. Human Genetics and Genomics Advances, 2022, 3, 100102.	1.7	5
108	Expanding the phenotypic spectrum of ARCN1-related syndrome. Genetics in Medicine, 2022, 24, 1227-1237.	2.4	5

#	Article	IF	Citations
109	Pathogenic variants in CDH11 impair cell adhesion and cause Teebi hypertelorism syndrome. Human Genetics, 2021, 140, 1061-1076.	3.8	4
110	BmHrp28 is a RNAâ€binding protein that binds to the femaleâ€specific exon 4 of Bombyx mori dsx preâ€mRNA. Insect Molecular Biology, 2009, 18, 795-803.	2.0	3
111	Generalized congenital epithelioid blue nevi (pigmented epithelioid melanocytomas) in an infant: Report of case and review of the literature. Journal of Cutaneous Pathology, 2019, 46, 954-959.	1.3	3
112	A homozygous truncating NALCN variant in two Afroâ€Caribbean siblings with hypotonia and dolichocephaly. American Journal of Medical Genetics, Part A, 2020, 182, 1877-1880.	1.2	3
113	Analysis of histone variant constraint and tissue expression suggests five potential novel human disease genes: H2AFY2, H2AFZ, H2AFY, H2AFV, H1F0. Human Genetics, 2022, 141, 1409-1421.	3.8	3
114	Further supporting <scp>SMARCC2</scp>â€related neurodevelopmental disorder through exome analysis and reanalysis in two patients. American Journal of Medical Genetics, Part A, 2022, 188, 878-882.	1.2	3
115	Application of exome sequencing to diagnose a novel presentation of the Cornelia de Lange syndrome in an Afroâ€Caribbean family. Molecular Genetics & Genomic Medicine, 2020, 8, e1318.	1.2	2
116	Chromosome 4q28.3q32.3 duplication in a patient with lymphatic malformations, craniosynostosis, and dysmorphic features. Clinical Dysmorphology, 2021, 30, 89-92.	0.3	2
117	Hypocalcemia as the Initial Presentation of Type 2 Bartter Syndrome: A Family Report. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e1679-e1688.	3.6	2
118	Novel PTH Gene Mutations Causing Isolated Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e2449-e2458.	3.6	2
119	Aortic coarctation and carotid artery aneurysm in a patient with hardikar syndrome: Cardiovascular implications for affected individuals. American Journal of Medical Genetics, Part A, 2016, 170, 482-486.	1.2	1
120	Discovery of a neuromuscular syndrome caused by biallelic variants in ASCC3. Human Genetics and Genomics Advances, 2021, 2, 100024.	1.7	1
121	Cleft palate morphology, genetic etiology, and risk of mortality in infants with Robin sequence. American Journal of Medical Genetics, Part A, 2021, 185, 3694-3700.	1.2	1
122	A novel unbalanced translocation between chromosomes 5p and 18q leading to dysmorphology and global developmental delay. Molecular Genetics & Genomic Medicine, 2022, , e1900.	1.2	1
123	Exome and <scp>RNAâ€Seq</scp> analyses of an incomplete penetrance variant in <scp> USP9X </scp> in femaleâ€specific syndromic intellectual disability. American Journal of Medical Genetics, Part A, 2022, , .	1.2	1
124	Contribution of Mendelian disorders in a population-based pediatric neurodegeneration cohort. Journal of Pediatrics, 2022, , .	1.8	0