Dong Li

List of Publications by Year in descending order

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

#	Article	IF	Citations
1	Treatment of severe Kaposiform lymphangiomatosis positive for NRAS mutation by MEK inhibition. Pediatric Research, 2023, 94, 1911-1915.	2.3	16
2	Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. Biological Psychiatry, 2022, 91, 313-327.	1.3	114
3	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
4	Variants in ADD1 cause intellectual disability, corpus callosum dysgenesis, and ventriculomegaly in humans. Genetics in Medicine, 2022, 24, 319-331.	2.4	6
5	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
6	Chromosome restructuring and number change during the evolution of <i>Morus notabilis</i> and <i>Morus alba</i> . Horticulture Research, 2022, 9, .	6.3	16
7	A novel unbalanced translocation between chromosomes 5p and 18q leading to dysmorphology and global developmental delay. Molecular Genetics & Enomic Medicine, 2022, , e1900.	1.2	1
8	Novel <i>PTH</i> Gene Mutations Causing Isolated Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e2449-e2458.	3.6	2
9	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
10	Expanding the phenotypic spectrum of ARCN1-related syndrome. Genetics in Medicine, 2022, 24, 1227-1237.	2.4	5
11	Exome and <scp>RNAâ€6eq</scp> analyses of an incomplete penetrance variant in <scp> <i>USP9X</i> </scp> in femaleâ€specific syndromic intellectual disability. American Journal of Medical Genetics, Part A, 2022, , .	1.2	1
12	Further supporting <scp><i>SMARCC2</i></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
13	Contribution of Mendelian disorders in a population-based pediatric neurodegeneration cohort. Journal of Pediatrics, 2022, , .	1.8	O
14	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
15	Shared genetic risk between eating disorder†and substanceâ€useâ€related phenotypes: Evidence from genomeâ€wide association studies. Addiction Biology, 2021, 26, e12880.	2.6	28
16	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
17	Expanded phenotypic spectrum of <scp><i>JAG1</i></scp> â€associated diseases: Central conducting lymphatic anomaly with a pathogenic variant in <scp><i>JAG1</i></scp> . Clinical Genetics, 2021, 99, 742-743.	2.0	7
18	A novel heterotaxy gene: Expansion of the phenotype of TTC21B $\hat{a} \in s$ pectrum disease. American Journal of Medical Genetics, Part A, 2021, 185, 1266-1269.	1.2	5

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19	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
20	The chromatin remodeler ISWI acts during <i> Drosophila < /i > development to regulate adult sleep. Science Advances, 2021, 7, .</i>	10.3	9
21	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
22	<i>ALG13</i> Xâ€inked intellectual disability: New variants, glycosylation analysis, and expanded phenotypes. Journal of Inherited Metabolic Disease, 2021, 44, 1001-1012.	3.6	9
23	Pathogenic variants in CDH11 impair cell adhesion and cause Teebi hypertelorism syndrome. Human Genetics, 2021, 140, 1061-1076.	3.8	4
24	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
25	Discovery of a neuromuscular syndrome caused by biallelic variants in ASCC3. Human Genetics and Genomics Advances, 2021, 2, 100024.	1.7	1
26	A new syndrome of moyamoya disease, kidney dysplasia, aminotransferase elevation, and skin disease associated withÂde novo variants in <scp><i>RNF213</i></scp> . American Journal of Medical Genetics, Part A, 2021, 185, 2168-2174.	1.2	8
27	Pathogenic variants in <i>SMARCA5</i> , a chromatin remodeler, cause a range of syndromic neurodevelopmental features. Science Advances, 2021, 7, .	10.3	17
28	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
29	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
30	Expanding the phenotype of <scp><i>ASXL3</i></scp> â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp><i>ASXL3</i></scp> . American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	1.2	12
31	Ciliopathies: Coloring outside of the lines. American Journal of Medical Genetics, Part A, 2021, 185, 687-694.	1.2	7
32	Chromosome 4q28.3q32.3 duplication in a patient with lymphatic malformations, craniosynostosis, and dysmorphic features. Clinical Dysmorphology, 2021, 30, 89-92.	0.3	2
33	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
34	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î ² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
35	Heterozygous de novo variants in <scp><i>CSNK1G1</i></scp> are associated with syndromic developmental delay and autism spectrum disorder. Clinical Genetics, 2020, 98, 571-576.	2.0	10
36	Clinical variability of TUBB â€associated disorders: Diagnosis through reanalysis. American Journal of Medical Genetics, Part A, 2020, 182, 3035-3039.	1.2	7

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37	EP300 â€related Rubinstein–Taybi syndrome: Highlighted rare phenotypic findings and a genotype–phenotype metaâ€analysis of 74 patients. American Journal of Medical Genetics, Part A, 2020, 182, 2926-2938.	1.2	16
38	The variability of <scp><i>SMARCA4</i></scp> â€related <scp>Coffin–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
39	Histone H3.3 beyond cancer: Germline mutations in <i>Histone 3 Family 3A and 3B</i> cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	10.3	43
40	Biliary-Atresia-Associated Mannosidase-1-Alpha-2 Gene Regulates Biliary and Ciliary Morphogenesis and Laterality. Frontiers in Physiology, 2020, 11, 538701.	2.8	13
41	Proteomic Profiling Reveals Roles of Stress Response, Ca ²⁺ Transient Dysregulation, and Novel Signaling Pathways in Alcoholâ€Induced Cardiotoxicity. Alcoholism: Clinical and Experimental Research, 2020, 44, 2187-2199.	2.4	6
42	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
43	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
44	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
45	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
46	Activating variants in <scp><i>PDGFRB</i></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
47	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
48	Application of exome sequencing to diagnose a novel presentation of the Cornelia de Lange syndrome in an Afroâ€Caribbean family. Molecular Genetics & Enomic Medicine, 2020, 8, e1318.	1.2	2
49	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
50	MMHub, a database for the mulberry metabolome. Database: the Journal of Biological Databases and Curation, 2020, 2020, .	3.0	14
51	Novel truncating mutations in CTNND1 cause a dominant craniofacial and cardiac syndrome. Human Molecular Genetics, 2020, 29, 1900-1921.	2.9	21
52	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
53	Flavones Produced by Mulberry Flavone Synthase Type I Constitute a Defense Line against the Ultraviolet-B Stress. Plants, 2020, 9, 215.	3.5	21
54	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

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55	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
56	Kaposiform lymphangiomatosis effectively treated with <scp>MEK</scp> inhibition. EMBO Molecular Medicine, 2020, 12, e12324.	6.9	51
57	PRKACB variants in skeletal disease or adrenocortical hyperplasia: effects on protein kinase A. Endocrine-Related Cancer, 2020, 27, 647-656.	3.1	7
58	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
59	Mutations in topoisomerase $\rm Ill^2$ result in a B cell immunodeficiency. Nature Communications, 2019, 10, 3644.	12.8	37
60	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
61	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
62	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
63	ARAF recurrent mutation causes central conducting lymphatic anomaly treatable with a MEK inhibitor. Nature Medicine, 2019, 25, 1116-1122.	30.7	136
64	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
65	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
66	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. Clinical Epigenetics, 2019, 11, 64.	4.1	71
67	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
68	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
69	KAT6A Syndrome: genotype–phenotype correlation in 76 patients with pathogenic KAT6A variants. Genetics in Medicine, 2019, 21, 850-860.	2.4	68
70	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
71	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
72	Expanding the phenotypic spectrum of <i>TP63</i> â€related disorders including the first set of monozygotic twins. American Journal of Medical Genetics, Part A, 2018, 176, 75-81.	1.2	11

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73	Extension of the mutational and clinical spectrum of <i>SOX2</i> 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
74	Coronary Artery Calcium on Noncontrast Thoracic Computerized Tomography Scans and All-Cause Mortality. Circulation, 2018, 138, 2437-2438.	1.6	15
75	Heterozygous Mutations in TBX1 as a Cause of Isolated Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 4023-4032.	3.6	15
76	HDL (High-Density Lipoprotein) Metrics and Atherosclerotic Risk in Women. Arteriosclerosis, Thrombosis, and Vascular Biology, 2018, 38, 2236-2244.	2.4	52
77	Pathogenic variant in EPHB4 results in central conducting lymphatic anomaly. Human Molecular Genetics, 2018, 27, 3233-3245.	2.9	73
78	USMG5 Ashkenazi Jewish founder mutation impairs mitochondrial complex V dimerization and ATP synthesis. Human Molecular Genetics, 2018, 27, 3305-3312.	2.9	45
79	CYP3A4 mutation causes vitamin D–dependent rickets type 3. Journal of Clinical Investigation, 2018, 128, 1913-1918.	8.2	77
80	Increasing diagnostic yield by RNA-Sequencing in rare disease—bypass hurdles of interpreting intronic or splice-altering variants. Annals of Translational Medicine, 2018, 6, 126-126.	1.7	9
81	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
82	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
83	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
84	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
85	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
86	Late-onset hereditary hypophosphatemic rickets with hypercalciuria (HHRH) due to mutation of SLC34A3/NPT2c. Bone, 2017, 97, 15-19.	2.9	30
87	Safety and Effectiveness of Juv $ ilde{A}$ ©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
88	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
89	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
90	Early Infantile Epileptic Encephalopathy in an <i>STXBP1</i> Patient with Lactic Acidemia and Normal Mitochondrial Respiratory Chain Function. Case Reports in Genetics, 2016, 2016, 1-5.	0.2	6

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91	<i>SMARCE1</i> , a rare cause of Coffin–Siris Syndrome: Clinical description of three additional cases. American Journal of Medical Genetics, Part A, 2016, 170, 1967-1973.	1.2	18
92	Enhanced B Cell Alloantigen Presentation and Its Epigenetic Dysregulation in Liver Transplant Rejection. American Journal of Transplantation, 2016, 16, 497-508.	4.7	17
93	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
94	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
95	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
96	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
97	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
98	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
99	Expanding the <i>SPECC1L</i> mutation phenotypic spectrum to include Teebi hypertelorism syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2497-2502.	1.2	26
100	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
101	Mutations in SPECC1L, 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
102	Value of whole exome sequencing for syndromic retinal dystrophy diagnosis in young patients. Clinical and Experimental Ophthalmology, 2015, 43, 132-138.	2.6	7
103	Delayed diagnosis of congenital myasthenia due to associated mitochondrial enzyme defect. Neuromuscular Disorders, 2015, 25, 257-261.	0.6	16
104	Genetic sharing and heritability of paediatric age of onset autoimmune diseases. Nature Communications, 2015, 6, 8442.	12.8	58
105	Meta-analysis of shared genetic architecture across ten pediatric autoimmune diseases. Nature Medicine, 2015, 21, 1018-1027.	30.7	212
106	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
107	Autosomal Dominant Hypoparathyroidism Caused by Germline Mutation in <i>GNA11</i> : Phenotypic and Molecular Characterization. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1774-E1783.	3.6	79
108	AGC1 Deficiency Causes Infantile Epilepsy, Abnormal Myelination, and Reduced N-Acetylaspartate. JIMD Reports, 2014, 14, 77-85.	1.5	57

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109	A genome-wide association study of anorexia nervosa. Molecular Psychiatry, 2014, 19, 1085-1094.	7.9	282
110	Molecular footprints of domestication and improvement in soybean revealed by whole genome re-sequencing. BMC Genomics, 2013, 14, 579.	2.8	186
111	Mutations in PDGFRB Cause Autosomal-Dominant Infantile Myofibromatosis. American Journal of Human Genetics, 2013, 92, 1001-1007.	6.2	174
112	Draft genome of the wheat A-genome progenitor Triticum urartu. Nature, 2013, 496, 87-90.	27.8	700
113	Aegilops tauschii draft genome sequence reveals a gene repertoire for wheat adaptation. Nature, 2013, 496, 91-95.	27.8	714
114	Whole-genome sequencing of giant pandas provides insights into demographic history and local adaptation. Nature Genetics, 2013, 45, 67-71.	21.4	303
115	Draft genome sequence of the mulberry tree Morus notabilis. Nature Communications, 2013, 4, 2445.	12.8	277
116	Identification and characterization of Sox genes in the silkworm, Bombyx mori. Molecular Biology Reports, 2011, 38, 3573-3584.	2.3	23
117	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
118	MicroRNAs of Bombyx mori identified by Solexa sequencing. BMC Genomics, 2010, 11, 148.	2.8	107
119	Single base–resolution methylome of the silkworm reveals a sparse epigenomic map. Nature Biotechnology, 2010, 28, 516-520.	17.5	349
120	Genome-wide patterns of genetic variation among elite maize inbred lines. Nature Genetics, 2010, 42, 1027-1030.	21.4	439
121	BmSE, a SINE family with $3\hat{a}\in^2$ ends of (ATTT) repeats in domesticated silkworm (Bombyx mori). Journal of Genetics and Genomics, 2010, 37, 125-135.	3.9	7
122	Complete Resequencing of 40 Genomes Reveals Domestication Events and Genes in Silkworm () Tj ETQq0 0 0 rg	gBT ₁ /Overlo	ock 10 Tf 50 2
123	BmHrp28 is a RNAâ€binding protein that binds to the femaleâ€specific exon 4 of <i>Bombyx mori dsx</i> preâ€mRNA. Insect Molecular Biology, 2009, 18, 795-803.	2.0	3
124	The diploid genome sequence of an Asian individual. Nature, 2008, 456, 60-65.	27.8	834