Jessica X Chong

List of Publications by Year
in descending order

56 papers	10,537 citations	172386 29 h-index	155592 55 g-index
68 all docs	68 docs citations	68 times ranked	23400 citing authors

#	Article	IF	Citations
1	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
2	Variantâ€level matching for diagnosis and discovery: Challenges and opportunities. Human Mutation, 2022, , .	1.1	11
3	Exome-wide rare variant analysis in familial essential tremor. Parkinsonism and Related Disorders, 2021, 82, 109-116.	1.1	11
4	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	13.7	45
5	Targeted long-read sequencing identifies missing disease-causing variation. American Journal of Human Genetics, 2021, 108, 1436-1449.	2.6	105
6	Biallelic variants in MESD, which encodes a WNT-signaling-related protein, in four new families with recessively inherited osteogenesis imperfecta. Human Genetics and Genomics Advances, 2021, 2, 100051.	1.0	3
7	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	1.1	24
8	Response to Hall etÂal.. American Journal of Human Genetics, 2020, 107, 1188-1189.	2.6	0
9	Biallelic mutations in LAMA5 disrupts a skeletal noncanonical focal adhesion pathway and produces a distinct bent bone dysplasia. EBioMedicine, 2020, 62, 103075.	2.7	7
10	Mutations in MYLPF Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogryposis. American Journal of Human Genetics, 2020, 107, 293-310.	2.6	21
11	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	1.1	25
12	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	13.7	6,140
13	Exome sequencing of family trios from the National Birth Defects Prevention Study: Tapping into a rich resource of genetic and environmental data. Birth Defects Research, 2019, 111, 1618-1632.	0.8	9
14	Mendelian Gene Discovery: Fast and Furious with No End in Sight. American Journal of Human Genetics, 2019, 105, 448-455.	2.6	166
15	Mutations in GDF11 and the extracellular antagonist, Follistatin, as a likely cause of Mendelian forms of orofacial clefting in humans. Human Mutation, 2019, 40, 1813-1825.	1.1	26
16	Front Cover, Volume 40, Issue 10. Human Mutation, 2019, 40, i.	1.1	0
17	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
18	Loss of function, missense, and intronic variants in NOTCH1 confer different risks for left ventricular outflow tract obstructive heart defects in two European cohorts. Genetic Epidemiology, 2019, 43, 215-226.	0.6	25

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19	A presynaptic congenital myasthenic syndrome attributed to a homozygous sequence variant in LAMA5. Annals of the New York Academy of Sciences, 2018, 1413, 119-125.	1.8	7
20	Plain-language medical vocabulary for precision diagnosis. Nature Genetics, 2018, 50, 474-476.	9.4	28
21	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	2.6	138
22	Variation in Cilia Protein Genes and Progression of Lung Disease in Cystic Fibrosis. Annals of the American Thoracic Society, 2018, 15, 440-448.	1.5	14
23	Genetic analysis of CHARGE syndrome identifies overlapping molecular biology. Genetics in Medicine, 2018, 20, 1022-1029.	1.1	43
24	ERCC4 variants identified in a cohort of patients with segmental progeroid syndromes. Human Mutation, 2018, 39, 255-265.	1.1	23
25	Mutations in the Epithelial Cadherin-p120-Catenin Complex Cause Mendelian Non-Syndromic Cleft Lip with or without Cleft Palate. American Journal of Human Genetics, 2018, 102, 1143-1157.	2.6	94
26	A postnatal role for embryonic myosin revealed by MYH3 mutations that alter TGFÎ² signaling and cause autosomal dominant spondylocarpotarsal synostosis. Scientific Reports, 2017, 7, 41803.	1.6	29
27	Molecular outcomes, clinical consequences, and genetic diagnosis of Oculocutaneous Albinism in Pakistani population. Scientific Reports, 2017, 7, 44185.	1.6	25
28	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
29	Presynaptic congenital myasthenic syndrome with a homozygous sequence variant in LAMA5 combines myopia, facial tics, and failure of neuromuscular transmission. American Journal of Medical Genetics, Part A, 2017, 173, 2240-2245.	0.7	29
30	Survival beyond the perinatal period expands the phenotypes caused by mutations in GLE1. American Journal of Medical Genetics, Part A, 2017, 173, 3098-3103.	0.7	10
31	Matchmaker Exchange. Current Protocols in Human Genetics, 2017, 95, 9.31.1-9.31.15.	3.5	47
32	DUOX2 Gene Mutation Manifesting as Resistance to Thyrotropin Phenotype. Thyroid, 2017, 27, 129-131.	2.4	19
33	Whole genome sequencing of extreme phenotypes identifies variants in CD101 and UBE2V1 associated with increased risk of sexually acquired HIV-1. PLoS Pathogens, 2017, 13, e1006703.	2.1	16
34	Recessive Inactivating Mutations in TBCK, Encoding a Rab GTPase-Activating Protein, Cause Severe Infantile Syndromic Encephalopathy. American Journal of Human Genetics, 2016, 98, 772-781.	2.6	43
35	MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. American Journal of Human Genetics, 2016, 99, 1229-1244.	2.6	91
36	Gene discovery for Mendelian conditions via social networking: de novo variants in KDM1A cause developmental delay and distinctive facial features. Genetics in Medicine, 2016, 18, 788-795.	1.1	88

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37	De Novo Mutations in NALCN Cause a Syndrome Characterized by Congenital Contractures of the Limbs and Face, Hypotonia, and Developmental Delay. American Journal of Human Genetics, 2015, 96, 462-473.	2.6	124
38	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
39	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	2.6	55
40	Exome Sequencing of Phenotypic Extremes Identifies CAV2 and TMC6 as Interacting Modifiers of Chronic Pseudomonas aeruginosa Infection in Cystic Fibrosis. PLoS Genetics, 2015, 11, e1005273.	1.5	39
41	Whole-Genome Sequencing of Individuals from a Founder Population Identifies Candidate Genes for Asthma. PLoS ONE, 2014, 9, e104396.	1.1	42
42	Solving Glycosylation Disorders: Fundamental Approaches Reveal Complicated Pathways. American Journal of Human Genetics, 2014, 94, 161-175.	2.6	222
43	Pathogenic Variants for Mendelian and Complex Traits in Exomes of 6,517 European and African Americans: Implications for the Return of Incidental Results. American Journal of Human Genetics, 2014, 95, 183-193.	2.6	78
44	Disclosure of Genetic Research Results to Members of a Founder Population. Journal of Genetic Counseling, 2014, 23, 984-991.	0.9	5
45	Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogryposis Type 5. American Journal of Human Genetics, 2014, 94, 734-744.	2.6	171
46	Practices and Policies of Clinical Exome Sequencing Providers: Analysis and Implications. American Journal of Medical Genetics, Part A, 2013, 161, 935-950.	0.7	60
47	Recessive TRAPPC11 Mutations Cause a Disease Spectrum of Limb Girdle Muscular Dystrophy and Myopathy with Movement Disorder and Intellectual Disability. American Journal of Human Genetics, 2013, 93, 181-190.	2.6	98
48	Mosaicism of the UDP-Galactose Transporter SLC35A2 Causes a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2013, 92, 632-636.	2.6	114
49	Homozygous Founder Mutation in Desmocollin-2 (DSC2) Causes Arrhythmogenic Cardiomyopathy in the Hutterite Population. Circulation: Cardiovascular Genetics, 2013, 6, 327-336.	5.1	47
50	Practices and Policies of Clinical Exome Sequencing Providers: Analysis and Implications. , 2013, 161, n/a-n/a.		2
51	Estimating the human mutation rate using autozygosity in a founder population. Nature Genetics, 2012, 44, 1277-1281.	9.4	202
52	A Population-Based Study of Autosomal-Recessive Disease-Causing Mutations in a Founder Population. American Journal of Human Genetics, 2012, 91, 608-620.	2.6	50
53	Accurate Imputation of Rare and Common Variants in a Founder Population From a Small Number of Sequenced Individuals. Genetic Epidemiology, 2012, 36, 312-319.	0.6	19
54	A shared founder mutation underlies restrictive dermopathy in Old Colony (Dutchâ€German) Mennonite and Hutterite patients in North America. American Journal of Medical Genetics, Part A, 2012, 158A, 1229-1232.	0.7	14

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55	Exome sequencing reveals a novel mutation for autosomal recessive non-syndromic mental retardation in the TECR gene on chromosome 19p13. Human Molecular Genetics, 2011, 20, 1285-1289.	1.4	94
56	A common spinal muscular atrophy deletion mutation is present on a single founder haplotype in the US Hutterites. European Journal of Human Genetics, 2011, 19, 1045-1051.	1.4	15