

# Jessica X Chong

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/3169331/publications.pdf>

Version: 2024-02-01

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	The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2020, 581, 434-443.	13.7	6,140
2	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. <i>American Journal of Human Genetics</i> , 2015, 97, 199-215.	2.6	574
3	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. <i>American Journal of Human Genetics</i> , 2017, 100, 695-705.	2.6	305
4	Solving Glycosylation Disorders: Fundamental Approaches Reveal Complicated Pathways. <i>American Journal of Human Genetics</i> , 2014, 94, 161-175.	2.6	222
5	Estimating the human mutation rate using autozygosity in a founder population. <i>Nature Genetics</i> , 2012, 44, 1277-1281.	9.4	202
6	Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogryposis Type 5. <i>American Journal of Human Genetics</i> , 2014, 94, 734-744.	2.6	171
7	Mendelian Gene Discovery: Fast and Furious with No End in Sight. <i>American Journal of Human Genetics</i> , 2019, 105, 448-455.	2.6	166
8	Insights into genetics, human biology and disease gleaned from family based genomic studies. <i>Genetics in Medicine</i> , 2019, 21, 798-812.	1.1	161
9	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 102, 309-320.	2.6	138
10	De Novo Mutations in NALCN Cause a Syndrome Characterized by Congenital Contractures of the Limbs and Face, Hypotonia, and Developmental Delay. <i>American Journal of Human Genetics</i> , 2015, 96, 462-473.	2.6	124
11	Mosaicism of the UDP-Galactose Transporter SLC35A2 Causes a Congenital Disorder of Glycosylation. <i>American Journal of Human Genetics</i> , 2013, 92, 632-636.	2.6	114
12	Targeted long-read sequencing identifies missing disease-causing variation. <i>American Journal of Human Genetics</i> , 2021, 108, 1436-1449.	2.6	105
13	Recessive TRAPPC11 Mutations Cause a Disease Spectrum of Limb Girdle Muscular Dystrophy and Myopathy with Movement Disorder and Intellectual Disability. <i>American Journal of Human Genetics</i> , 2013, 93, 181-190.	2.6	98
14	Exome sequencing reveals a novel mutation for autosomal recessive non-syndromic mental retardation in the <i>TECR</i> gene on chromosome 19p13. <i>Human Molecular Genetics</i> , 2011, 20, 1285-1289.	1.4	94
15	Mutations in the Epithelial Cadherin-p120-Catenin Complex Cause Mendelian Non-Syndromic Cleft Lip with or without Cleft Palate. <i>American Journal of Human Genetics</i> , 2018, 102, 1143-1157.	2.6	94
16	MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. <i>American Journal of Human Genetics</i> , 2016, 99, 1229-1244.	2.6	91
17	Gene discovery for Mendelian conditions via social networking: de novo variants in <i>KDM1A</i> cause developmental delay and distinctive facial features. <i>Genetics in Medicine</i> , 2016, 18, 788-795.	1.1	88
18	Pathogenic Variants for Mendelian and Complex Traits in Exomes of 6,517 European and African Americans: Implications for the Return of Incidental Results. <i>American Journal of Human Genetics</i> , 2014, 95, 183-193.	2.6	78

#	ARTICLE	IF	CITATIONS
19	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
20	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	2.6	55
21	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
22	Homozygous Founder Mutation in Desmocollin-2 ( DSC2 ) Causes Arrhythmogenic Cardiomyopathy in the Hutterite Population. Circulation: Cardiovascular Genetics, 2013, 6, 327-336.	5.1	47
23	Matchmaker Exchange. Current Protocols in Human Genetics, 2017, 95, 9.31.1-9.31.15.	3.5	47
24	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	13.7	45
25	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
26	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
27	Genetic analysis of CHARGE syndrome identifies overlapping molecular biology. Genetics in Medicine, 2018, 20, 1022-1029.	1.1	43
28	Whole-Genome Sequencing of Individuals from a Founder Population Identifies Candidate Genes for Asthma. PLoS ONE, 2014, 9, e104396.	1.1	42
29	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
30	A postnatal role for embryonic myosin revealed by MYH3 mutations that alter TGF $\beta$ <sup>2</sup> signaling and cause autosomal dominant spondylocarpotarsal synostosis. Scientific Reports, 2017, 7, 41803.	1.6	29
31	Presynaptic congenital myasthenic syndrome with a homozygous sequence variant in <i>LAMA5</i> combines myopia, facial tics, and failure of neuromuscular transmission. American Journal of Medical Genetics, Part A, 2017, 173, 2240-2245.	0.7	29
32	Plain-language medical vocabulary for precision diagnosis. Nature Genetics, 2018, 50, 474-476.	9.4	28
33	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
34	Molecular outcomes, clinical consequences, and genetic diagnosis of Oculocutaneous Albinism in Pakistani population. Scientific Reports, 2017, 7, 44185.	1.6	25
35	Loss of function, missense, and intronic variants in <i>NOTCH1</i> confer different risks for left ventricular outflow tract obstructive heart defects in two European cohorts. Genetic Epidemiology, 2019, 43, 215-226.	0.6	25
36	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

#	ARTICLE	IF	CITATIONS
37	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. <i>Genetics in Medicine</i> , 2020, 22, 538-546.	1.1	24
38	<i>ERCC4</i> variants identified in a cohort of patients with segmental progeroid syndromes. <i>Human Mutation</i> , 2018, 39, 255-265.	1.1	23
39	Mutations in MYLPF Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogryposis. <i>American Journal of Human Genetics</i> , 2020, 107, 293-310.	2.6	21
40	Accurate Imputation of Rare and Common Variants in a Founder Population From a Small Number of Sequenced Individuals. <i>Genetic Epidemiology</i> , 2012, 36, 312-319.	0.6	19
41	<i>DUOX2</i> Gene Mutation Manifesting as Resistance to Thyrotropin Phenotype. <i>Thyroid</i> , 2017, 27, 129-131.	2.4	19
42	Whole genome sequencing of extreme phenotypes identifies variants in CD101 and UBE2V1 associated with increased risk of sexually acquired HIV-1. <i>PLoS Pathogens</i> , 2017, 13, e1006703.	2.1	16
43	A common spinal muscular atrophy deletion mutation is present on a single founder haplotype in the US Hutterites. <i>European Journal of Human Genetics</i> , 2011, 19, 1045-1051.	1.4	15
44	A shared founder mutation underlies restrictive dermopathy in Old Colony (Dutch-German) Mennonite and Hutterite patients in North America. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1229-1232.	0.7	14
45	Variation in Cilia Protein Genes and Progression of Lung Disease in Cystic Fibrosis. <i>Annals of the American Thoracic Society</i> , 2018, 15, 440-448.	1.5	14
46	Exome-wide rare variant analysis in familial essential tremor. <i>Parkinsonism and Related Disorders</i> , 2021, 82, 109-116.	1.1	11
47	Variant-level matching for diagnosis and discovery: Challenges and opportunities. <i>Human Mutation</i> , 2022, , .	1.1	11
48	Survival beyond the perinatal period expands the phenotypes caused by mutations in <i>GLE1</i> . <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3098-3103.	0.7	10
49	Exome sequencing of family trios from the National Birth Defects Prevention Study: Tapping into a rich resource of genetic and environmental data. <i>Birth Defects Research</i> , 2019, 111, 1618-1632.	0.8	9
50	A presynaptic congenital myasthenic syndrome attributed to a homozygous sequence variant in <i>LAMA5</i> . <i>Annals of the New York Academy of Sciences</i> , 2018, 1413, 119-125.	1.8	7
51	Biallelic mutations in LAMA5 disrupts a skeletal noncanonical focal adhesion pathway and produces a distinct bent bone dysplasia. <i>EBioMedicine</i> , 2020, 62, 103075.	2.7	7
52	Disclosure of Genetic Research Results to Members of a Founder Population. <i>Journal of Genetic Counseling</i> , 2014, 23, 984-991.	0.9	5
53	Biallelic variants in MESD, which encodes a WNT-signaling-related protein, in four new families with recessively inherited osteogenesis imperfecta. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100051.	1.0	3
54	Practices and Policies of Clinical Exome Sequencing Providers: Analysis and Implications. , 2013, 161, n/a-n/a.		2

#	ARTICLE	IF	CITATIONS
55	Front Cover, Volume 40, Issue 10. Human Mutation, 2019, 40, i.	1.1	0
56	Response to Hall et al.. American Journal of Human Genetics, 2020, 107, 1188-1189.	2.6	0