Sau Wai Cheung

List of Publications by Year
in descending order

108	9,063	38742 50	42399 92
papers	citations	h-index	g-index

113	113	113	12915
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. Nature Genetics, 2008, 40, 1466-1471.	21.4	535
2	High-resolution genomic profiling of chromosomal aberrations using Infinium whole-genome genotyping. Genome Research, 2006, 16, 1136-1148.	5.5	448
3	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. Journal of Medical Genetics, 2010, 47, 332-341.	3.2	447
4	Chromosome Catastrophes Involve Replication Mechanisms Generating Complex Genomic Rearrangements. Cell, 2011, 146, 889-903.	28.9	391
5	Genomic and Genic Deletions of the FOX Gene Cluster on 16q24.1 and Inactivating Mutations of FOXF1 Cause Alveolar Capillary Dysplasia and Other Malformations. American Journal of Human Genetics, 2009, 84, 780-791.	6.2	389
6	Increased MECP2 gene copy number as the result of genomic duplication in neurodevelopmentally delayed males. Genetics in Medicine, 2006, 8, 784-792.	2.4	245
7	Development and validation of a CGH microarray for clinical cytogenetic diagnosis. Genetics in Medicine, 2005, 7, 422-432.	2.4	241
8	TBX6 Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350.	27.0	239
9	Detection of clinically relevant exonic copy-number changes by array CGH. Human Mutation, 2010, 31, 1326-1342.	2.5	225
10	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	6.2	219
11	Increased LIS1 expression affects human and mouse brain development. Nature Genetics, 2009, 41, 168-177.	21.4	199
12	Clinical Implementation of Chromosomal Microarray Analysis: Summary of 2513 Postnatal Cases. PLoS ONE, 2007, 2, e327.	2.5	191
13	Clinical use of array comparative genomic hybridization (aCGH) for prenatal diagnosis in 300 cases. Prenatal Diagnosis, 2009, 29, 29-39.	2.3	180
14	The array CGH and its clinical applications. Drug Discovery Today, 2008, 13, 760-770.	6.4	171
15	Complex rearrangements in patients with duplications of MECP2 can occur by fork stalling and template switching. Human Molecular Genetics, 2009, 18, 2188-2203.	2.9	165
16	Microarrayâ€based CGH detects chromosomal mosaicism not revealed by conventional cytogenetics. American Journal of Medical Genetics, Part A, 2007, 143A, 1679-1686.	1.2	158
17	Prenatal diagnosis of chromosomal abnormalities using array-based comparative genomic hybridization. Genetics in Medicine, 2006, 8, 719-727.	2.4	154
18	Loss of Î´-catenin function in severe autism. Nature, 2015, 520, 51-56.	27.8	145

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19	Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture. Human Molecular Genetics, 2009, 18, 3579-3593.	2.9	143
20	Positive predictive value estimates for cell-free noninvasive prenatal screening from data of a large referral genetic diagnostic laboratory. American Journal of Obstetrics and Gynecology, 2017, 217, 691.e1-691.e6.	1.3	141
21	Genomic Imbalances in Neonates With Birth Defects: High Detection Rates by Using Chromosomal Microarray Analysis. Pediatrics, 2008, 122, 1310-1318.	2.1	137
22	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	2.4	127
23	NAHR-mediated copy-number variants in a clinical population: Mechanistic insights into both genomic disorders and Mendelizing traits. Genome Research, 2013, 23, 1395-1409.	5.5	120
24	Identification of chromosome abnormalities in subtelomeric regions by microarray analysis: A study of 5,380 cases. American Journal of Medical Genetics, Part A, 2008, 146A, 2242-2251.	1.2	113
25	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. European Journal of Human Genetics, 2014, 22, 79-87.	2.8	112
26	Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. Human Mutation, 2010, 31, 840-850.	2.5	111
27	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	10.2	108
28	Duplications of FOXG1 in 14q12 are associated with developmental epilepsy, mental retardation, and severe speech impairment. European Journal of Human Genetics, 2011, 19, 102-107.	2.8	104
29	Prenatal chromosomal microarray analysis in a diagnostic laboratory; experience with >1000 cases and review of the literature. Prenatal Diagnosis, 2012, 32, 351-361.	2.3	103
30	Phenotypic spectrum and genotypeâ€“phenotype correlations of NRXN1 exon deletions. European Journal of Human Genetics, 2012, 20, 1240-1247.	2.8	99
31	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. American Journal of Human Genetics, 2016, 98, 373-381.	6.2	95
32	Bacterial artificial chromosome-emulation oligonucleotide arrays for targeted clinical array-comparative genomic hybridization analyses. Genetics in Medicine, 2008, 10, 278-289.	2.4	90
33	Insertional translocation detected using FISH confirmation of arrayâ€comparative genomic hybridization (aCGH) results. American Journal of Medical Genetics, Part A, 2010, 152A, 1111-1126.	1.2	85
34	Alu-mediated diverse and complex pathogenic copy-number variants within human chromosome 17 at p13.3. Human Molecular Genetics, 2015, 24, 4061-4077.	2.9	83
35	BAFopathiesâ€™ DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes. Nature Communications, 2018, 9, 4885.	12.8	83
36	Pre- and postnatal genetic testing by array-comparative genomic hybridization: genetic counseling perspectives. Genetics in Medicine, 2008, 10, 13-18.	2.4	77

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37	Syndromic thrombocytopenia and predisposition to acute myelogenous leukemia caused by constitutional microdeletions on chromosome 21q. Blood, 2008, 112, 1042-1047.	1.4	74
38	NUDT21-spanning CNVs lead to neuropsychiatric disease and altered MeCP2 abundance via alternative polyadenylation. ELife, 2015, 4, .	6.0	74
39	Observation and prediction of recurrent human translocations mediated by NAHR between nonhomologous chromosomes. Genome Research, 2011, 21, 33-46.	5.5	72
40	DNA Methylation Profiling of Uniparental Disomy Subjects Provides a Map of Parental Epigenetic Bias in the Human Genome. American Journal of Human Genetics, 2016, 99, 555-566.	6.2	66
41	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	28.9	66
42	Genome Sequencing Explores Complexity of Chromosomal Abnormalities in Recurrent Miscarriage. American Journal of Human Genetics, 2019, 105, 1102-1111.	6.2	66
43	Redefined genomic architecture in 15q24 directed by patient deletion/duplication breakpoint mapping. Human Genetics, 2009, 126, 589-602.	3.8	65
44	Low-pass genome sequencing versus chromosomal microarray analysis: implementation in prenatal diagnosis. Genetics in Medicine, 2020, 22, 500-510.	2.4	64
45	The phenotype of recurrent 10q22q23 deletions and duplications. European Journal of Human Genetics, 2011, 19, 400-408.	2.8	63
46	Comparison of chromosome analysis and chromosomal microarray analysis: what is the value of chromosome analysis in todayâ€™s genomic array era?. Genetics in Medicine, 2013, 15, 450-457.	2.4	63
47	Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. Genome Research, 2013, 23, 1383-1394.	5.5	62
48	Aberrations in Pseudoautosomal Regions (PARs) Found in Infertile Men with Y-Chromosome Microdeletions. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E674-E679.	3.6	58
49	Chromosomal microarray analysis (CMA) detects a large X chromosome deletion including FMR1, FMR2, and IDS in a female patient with mental retardation. American Journal of Medical Genetics, Part A, 2007, 143A, 1358-1365.	1.2	57
50	Rapid prenatal diagnosis using uncultured amniocytes and oligonucleotide array CGH. Prenatal Diagnosis, 2008, 28, 943-949.	2.3	57
51	Detection of copy-number variation in AUTS2 gene by targeted exonic array CGH in patients with developmental delay and autistic spectrum disorders. European Journal of Human Genetics, 2013, 21, 343-346.	2.8	56
52	Identification of balanced chromosomal rearrangements previously unknown among participants in the 1000 Genomes Project: implications for interpretation of structural variation in genomes and the future of clinical cytogenetics. Genetics in Medicine, 2018, 20, 697-707.	2.4	52
53	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	2.4	52
54	Identification of Critical Regions and Candidate Genes for Cardiovascular Malformations and Cardiomyopathy Associated with Deletions of Chromosome 1p36. PLoS ONE, 2014, 9, e85600.	2.5	51

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55	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	8.2	50
56	Increased gene copy number of VAMP7 disrupts human male urogenital development through altered estrogen action. Nature Medicine, 2014, 20, 715-724.	30.7	46
57	Interstitial deletion of 6q25.2â€“q25.3: a novel microdeletion syndrome associated with microcephaly, developmental delay, dysmorphic features and hearing loss. European Journal of Human Genetics, 2009, 17, 573-581.	2.8	45
58	Delineation of candidate genes responsible for structural brain abnormalities in patients with terminal deletions of chromosome 6q27. European Journal of Human Genetics, 2015, 23, 54-60.	2.8	45
59	Mechanisms for Complex Chromosomal Insertions. PLoS Genetics, 2016, 12, e1006446.	3.5	45
60	Accurate Description of DNA-Based Noninvasive Prenatal Screening. New England Journal of Medicine, 2015, 372, 1675-1677.	27.0	44
61	Int22h-1/int22h-2-mediated Xq28 rearrangements: intellectual disability associated with duplications and in utero male lethality with deletions. Journal of Medical Genetics, 2011, 48, 840-850.	3.2	43
62	Branchiootorenal syndrome and oculoauriculovertebral spectrum features associated with duplication ofSIX1,SIX6, andOTX2resulting from a complex chromosomal rearrangement. American Journal of Medical Genetics, Part A, 2008, 146A, 2480-2489.	1.2	42
63	Dosage Changes of a Segment at 17p13.1 Lead to Intellectual Disability and Microcephaly as a Result of Complex Genetic Interaction of Multiple Genes. American Journal of Human Genetics, 2014, 95, 565-578.	6.2	40
64	Genetic diagnosis of autism spectrum disorders: The opportunity and challenge in the genomics era. Critical Reviews in Clinical Laboratory Sciences, 2014, 51, 249-262.	6.1	38
65	CHRNA7 triplication associated with cognitive impairment and neuropsychiatric phenotypes in a three-generation pedigree. European Journal of Human Genetics, 2014, 22, 1071-1076.	2.8	37
66	Clinical characterization of int22h1/int22h2-mediated Xq28 duplication/deletion: new cases and literature review. BMC Medical Genetics, 2015, 16, 12.	2.1	37
67	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
68	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	2.4	36
69	Challenges in clinical interpretation of microduplications detected by array CGH analysis. American Journal of Medical Genetics, Part A, 2010, 152A, 1089-1100.	1.2	35
70	6q22.1 microdeletion and susceptibility to pediatric epilepsy. European Journal of Human Genetics, 2015, 23, 173-179.	2.8	35
71	Nonrecurrent 17p11.2p12 Rearrangement Events that Result in Two Concomitant Genomic Disorders: The PMP22-RAI1 Contiguous Gene Duplication Syndrome. American Journal of Human Genetics, 2015, 97, 691-707.	6.2	33
72	Neurodevelopmental and neurobehavioral characteristics in males and females with CDKL5 duplications. European Journal of Human Genetics, 2015, 23, 915-921.	2.8	32

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73	Deletion and duplication of 15q24: Molecular mechanisms and potential modification by additional copy number variants. Genetics in Medicine, 2010, 12, 573-586.	2.4	31
74	MECP2 duplications in six patients with complex sex chromosome rearrangements. European Journal of Human Genetics, 2011, 19, 409-415.	2.8	30
75	Detection of â‰¥1â€‰Mb microdeletions and microduplications in a single cell using custom oligonucleotide arrays. Prenatal Diagnosis, 2012, 32, 10-20.	2.3	29
76	Genitourinary Defects Associated with Genomic Deletions in 2p15 Encompassing OTX1. PLoS ONE, 2014, 9, e107028.	2.5	29
77	IncreasedSTAG2dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. Human Molecular Genetics, 2015, 24, 7171-7181.	2.9	28
78	Characterization of chromosomal abnormalities in pregnancy losses reveals critical genes and loci for human early development. Human Mutation, 2017, 38, 669-677.	2.5	28
79	Novel applications of array comparative genomic hybridization in molecular diagnostics. Expert Review of Molecular Diagnostics, 2018, 18, 531-542.	3.1	28
80	Prenatal diagnosis of a 9q34.3 microdeletion by arrayâ€CGH in a fetus with an apparently balanced translocation. Prenatal Diagnosis, 2007, 27, 1112-1117.	2.3	26
81	Contribution of genomic copy-number variations in prenatal oral clefts: a multicenter cohort study. Genetics in Medicine, 2016, 18, 1052-1055.	2.4	25
82	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Medicine, 2019, 11, 25.	8.2	22
83	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. Genetics in Medicine, 2019, 21, 1058-1064.	2.4	22
84	Small genomic rearrangements involving FMR1 support the importance of its gene dosage for normal neurocognitive function. Neurogenetics, 2012, 13, 333-339.	1.4	21
85	Universal Prenatal Chromosomal Microarray Analysis: Additive Value and Clinical Dilemmas in Fetuses with a Normal Karyotype. American Journal of Perinatology, 2017, 34, 340-348.	1.4	21
86	Evolution of prenatal genetics: from point mutation testing to chromosomal microarray analysis. Expert Review of Molecular Diagnostics, 2005, 5, 883-892.	3.1	17
87	Xq22 deletions and correlation with distinct neurological disease traits in females: Further evidence for a contiguous gene syndrome. Human Mutation, 2020, 41, 150-168.	2.5	15
88	Deciphering the complexity of simple chromosomal insertions by genome sequencing. Human Genetics, 2021, 140, 361-380.	3.8	15
89	Copy number variants in patients with intellectual disability affect the regulation of ARX transcription factor gene. Human Genetics, 2015, 134, 1163-1182.	3.8	14
90	Xp11.22 deletions encompassing CENPVL1, CENPVL2, MAGED1 and GSPT2 as a cause of syndromic X-linked intellectual disability. PLoS ONE, 2017, 12, e0175962.	2.5	14

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91	Parental somatic mosaicism for CNV deletions â€“ A need for more sensitive and precise detection methods in clinical diagnostics settings. Genomics, 2020, 112, 2937-2941.	2.9	14
92	Noninvasive prenatal screening for fetal sex chromosome aneuploidies. Expert Review of Molecular Diagnostics, 2021, 21, 405-415.	3.1	13
93	Mosaicism for r(X) and der(X)del(X)(p11.23)dup(X)(p11.21p11.22) Provides Insight into the Possible Mechanism of Rearrangement. Molecular Cytogenetics, 2008, 1, 16.	0.9	12
94	Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998.	2.5	12
95	Microarray-Based Comparative Genomic Hybridization Using Sex-Matched Reference DNA Provides Greater Sensitivity for Detection of Sex Chromosome Imbalances than Array-Comparative Genomic Hybridization with Sex-Mismatched Reference DNA. Journal of Molecular Diagnostics, 2009, 11, 226-237.	2.8	11
96	Molecular and clinical analyses of 16q24.1 duplications involving FOXF1 identify an evolutionarily unstable large minisatellite. BMC Medical Genetics, 2014, 15, 128.	2.1	11
97	Application of DNA Microarray to Clinical Diagnostics. Methods in Molecular Biology, 2016, 1368, 111-132.	0.9	10
98	Balanced Chromosomal Rearrangement Detection by Lowâ€Pass Wholeâ€Genome Sequencing. Current Protocols in Human Genetics, 2018, 96, 8.18.1-8.18.16.	3.5	10
99	A de novo 1.58â€‰Mb deletion, including MAP2K6 and mapping 1.28â€‰Mb upstream to SOX9, identified in a patient with Pierre Robin sequence and osteopenia with multiple fractures. American Journal of Medical Genetics, Part A, 2015, 167, 1842-1850.	1.2	9
100	Phenotypic association of 15q11.2 CNVs of the region of breakpoints 1â€“2 (BP1â€“BP2) in a large cohort of samples referred for genetic diagnosis. Journal of Human Genetics, 2019, 64, 253-255.	2.3	9
101	Targeted gene panel sequencing prenatally detects two novel mutations of DYNC2H1 in a fetus with increased biparietal diameter and polyhydramnios. Birth Defects Research, 2018, 110, 364-371.	1.5	6
102	Microarray analysis: <scp>F</scp>irstâ€trimester maternal serum free <scp>Î²â€hCG</scp> and the risk of significant copy number variants. Prenatal Diagnosis, 2018, 38, 971-978.	2.3	5
103	Reply to Amor et al. European Journal of Human Genetics, 2012, 20, 597-597.	2.8	4
104	Identification of complex chromosome 18 rearrangements by FISH and array CGH in two patients with apparent isochromosome 18q. American Journal of Medical Genetics, Part A, 2011, 155, 1465-1468.	1.2	2
105	Prenatal detection of 10q22q23 duplications: dilemmas in phenotype prediction. Prenatal Diagnosis, 2016, 36, 1211-1216.	2.3	1
106	Inside Back Cover, Volume 41, Issue 1. Human Mutation, 2020, 41, ii.	2.5	0
107	Clinical Evaluation of a Custom Genome Wide 44K Oligoarray for Copy Number Changes in Acute Myeloid Leukemia. Blood, 2008, 112, 4869-4869.	1.4	0
108	Clinical Application of aCGH for Prenatal Diagnosis: Experience with >350 Cases. FASEB Journal, 2009, 23, 179.3.	0.5	0