Ana Cristina Krepischi

List of Publications by Year
in descending order

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128 papers	3,307 citations	185998 28 h-index	174990 52 g-index
131 all docs	131 docs citations	131 times ranked	5836 citing authors

#	Article	IF	Citations
1	A Small Supernumerary Xp Marker Chromosome Including Genes <i>NR0B1</i> and <i>MAGEB</i> Causing Partial Gonadal Dysgenesis and Gonadoblastoma. Sexual Development, 2022, 16, 55-63.	1.1	1
2	Family-based whole-exome sequencing identifies rare variants potentially related to cutaneous melanoma predisposition in Brazilian melanoma-prone families. PLoS ONE, 2022, 17, e0262419.	1.1	5
3	Unraveling the Genetic Architecture of Hepatoblastoma Risk: Birth Defects and Increased Burden of Germline Damaging Variants in Gastrointestinal/Renal Cancer Predisposition and DNA Repair Genes. Frontiers in Genetics, 2022, 13, 858396.	1.1	6
4	Genetic investigation of syndromic forms of obesity. International Journal of Obesity, 2022, 46, 1582-1586.	1.6	6
5	Detection of mosaicism for segmental and whole chromosome imbalances by targeted sequencing. Annals of Human Genetics, 2021, 85, 18-26.	0.3	4
6	Germline variants of Brazilian women with breast cancer and detection of a novel pathogenic ATM deletion in early-onset breast cancer. Breast Cancer, 2021, 28, 346-354.	1.3	9
7	Insights from the genetic characterization of central precocious puberty associated with multiple anomalies. Human Reproduction, 2021, 36, 506-518.	0.4	16
8	A novel MYT1L mutation in a boy with syndromic obesity: Case report and literature review. Obesity Research and Clinical Practice, 2021, 15, 124-132.	0.8	9
9	Congenital chromoanagenesis in the routine postnatal chromosomal microarray analyses. American Journal of Medical Genetics, Part A, 2021, 185, 2335-2344.	0.7	1
10	DNA methylation as a key epigenetic player for hepatoblastoma characterization. Clinics and Research in Hepatology and Gastroenterology, 2021, 45, 101684.	0.7	3
11	HDAC1-Dependent Repression of Markers of Hepatocytes and P21 Is Involved in Development of Pediatric Liver Cancer. Cellular and Molecular Gastroenterology and Hepatology, 2021, 12, 1669-1682.	2.3	20
12	Chromoanagenesis Event Underlies a de novo Pericentric and Multiple Paracentric Inversions in a Single Chromosome Causing Coffinâ€“Siris Syndrome. Frontiers in Genetics, 2021, 12, 708348.	1.1	5
13	Molecular and cellular basis of hyperassembly and protein aggregation driven by a rare pathogenic mutation in DDX3X. IScience, 2021, 24, 102841.	1.9	14
14	Congenital limb deficiency: Genetic investigation of 44 individuals presenting mainly longitudinal defects in isolated or syndromic forms. Clinical Genetics, 2021, 100, 615-623.	1.0	4
15	An Apparently Balanced Complex Chromosome Rearrangement Involving Seven Breaks and Four Chromosomes in a Healthy Female and Segregation/Recombination in Her Affected Son. Molecular Syndromology, 2021, 12, 312-320.	0.3	2
16	Molecular and clinical insights into complex genomic rearrangements related to MECP2 duplication syndrome. European Journal of Medical Genetics, 2021, 64, 104367.	0.7	7
17	Two novel pathogenic variants in MED13L: one familial and one isolated case. Journal of Intellectual Disability Research, 2021, 65, 1049-1057.	1.2	4
18	Copy Number Alterations in Hepatoblastoma: Literature Review and a Brazilian Cohort Analysis Highlight New Biological Pathways. Frontiers in Oncology, 2021, 11, 741526.	1.3	5

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19	DNA methylation fingerprint of monozygotic twins and their singleton sibling with intellectual disability carrying a novel KDM5C mutation. European Journal of Medical Genetics, 2020, 63, 103737.	0.7	7
20	Somatic copy number alterations in pleomorphic adenoma and recurrent pleomorphic adenoma. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2020, 129, 59-64.	0.2	4
21	MEG3 and MEG8 aberrant methylation in an infant with neuroblastoma. Pediatric Blood and Cancer, 2020, 67, e28328.	0.8	2
22	Expanding the role of SETD5 haploinsufficiency in neurodevelopment and neuroblastoma. Pediatric Blood and Cancer, 2020, 67, e28376.	0.8	3
23	Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998.	1.1	12
24	Hepatoblastomas exhibit markedNNMTdownregulation driven by promoter DNA hypermethylation. Tumor Biology, 2020, 42, 101042832097712.	0.8	11
25	Insights in Osteosarcoma by Proton Nuclear Magnetic Resonance Serum Metabonomics. Frontiers in Oncology, 2020, 10, 506959.	1.3	9
26	Insights Into the Somatic Mutation Burden of Hepatoblastomas From Brazilian Patients. Frontiers in Oncology, 2020, 10, 556.	1.3	12
27	Understanding the Landscape of X-linked Variants Causing Intellectual Disability in Females Through Extreme X Chromosome Inactivation Skewing. Molecular Neurobiology, 2020, 57, 3671-3684.	1.9	21
28	Genetic investigation of patients with tall stature. European Journal of Endocrinology, 2020, 182, 139-147.	1.9	3
29	Investigating Genetic Factors Contributing to Variable Expressivity of Class I 17p13.3 Microduplication. International Journal of Molecular and Cellular Medicine, 2020, 9, 296-306.	1.1	1
30	A Brazilian cohort of individuals with Phelan-McDermid syndrome: genotype-phenotype correlation and identification of an atypical case. Journal of Neurodevelopmental Disorders, 2019, 11, 13.	1.5	37
31	TET Upregulation Leads to 5-Hydroxymethylation Enrichment in Hepatoblastoma. Frontiers in Genetics, 2019, 10, 553.	1.1	17
32	Insights into the Chemical Biology of Childhood Embryonal Solid Tumors by NMR-Based Metabolomics. Biomolecules, 2019, 9, 843.	1.8	8
33	10q23.31 microduplication encompassing PTEN decreases mTOR signalling activity and is associated with autosomal dominant primary microcephaly. Journal of Medical Genetics, 2019, 56, 543-547.	1.5	9
34	Mechanistic insights revealed by a UBE2A mutation linked to intellectual disability. Nature Chemical Biology, 2019, 15, 62-70.	3.9	19
35	Atypical presentation of a germline APC mutation in a child with supratentorial primitive neuroectodermal tumor. Pediatric Blood and Cancer, 2019, 66, e27566.	0.8	2
36	Deletion of RUNX1 exons 1 and 2 associated with familial platelet disorder with propensity to acute myeloid leukemia. Cancer Genetics, 2018, 222-223, 32-37.	0.2	22

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37	Pathogenic copy number variants in patients with congenital hypopituitarism associated with complex phenotypes. Clinical Endocrinology, 2018, 88, 425-431.	1.2	11
38	KIF11 microdeletion is associated with microcephaly, chorioretinopathy and intellectual disability. Human Genome Variation, 2018, 5, 18010.	0.4	8
39	Genomic profile of a squamous cell carcinoma ex pleomorphic adenoma compared to a head and neck squamous cell carcinoma. Brazilian Journal of Otorhinolaryngology, 2018, 84, 393-397.	0.4	0
40	Recurrent Copy Number Variants Associated with Syndromic Short Stature of Unknown Cause. Hormone Research in Paediatrics, 2018, 89, 13-21.	0.8	29
41	Methylome profiling of healthy and central precocious puberty girls. Clinical Epigenetics, 2018, 10, 146.	1.8	34
42	Utility of trio-based exome sequencing in the elucidation of the genetic basis of isolated syndromic intellectual disability: illustrative cases. The Application of Clinical Genetics, 2018, Volume 11, 93-98.	1.4	34
43	Insight into the mechanisms and consequences of recurrent telomere capture associated with a sub-telomeric deletion. Chromosome Research, 2018, 26, 191-198.	1.0	4
44	Abstract 2072: Genomic studies of Brazilian patients with hepatoblastoma: Insight into somatic mutations using whole-exome sequencing. , 2018, , .		0
45	Epigenetics insights into chronic pain: DNA hypomethylation in fibromyalgiaâ€”a controlled pilot-study. Pain, 2017, 158, 1473-1480.	2.0	65
46	Efficient detection of chromosome imbalances and single nucleotide variants using targeted sequencing in the clinical setting. European Journal of Medical Genetics, 2017, 60, 667-674.	0.7	10
47	The genetic and epigenetic landscapes of hepatoblastomas. Applied Cancer Research, 2017, 37, .	1.0	6
48	Mining Novel Candidate Imprinted Genes Using Genome-Wide Methylation Screening and Literature Review. Epigenomes, 2017, 1, 13.	0.8	2
49	Integrative Variation Analysis Reveals that a Complex Genotype May Specify Phenotype in Siblings with Syndromic Autism Spectrum Disorder. PLoS ONE, 2017, 12, e0170386.	1.1	2
50	Epigenetic signature of differentially methylated genes in cutaneous melanoma. Applied Cancer Research, 2017, 37, .	1.0	1
51	DNA methylation landscape of hepatoblastomas reveals arrest at early stages of liver differentiation and cancer-related alterations. Oncotarget, 2017, 8, 97871-97889.	0.8	32
52	Differential DNA Methylation of MicroRNA Genes in Temporal Cortex from Alzheimerâ€™s Disease Individuals. Neural Plasticity, 2016, 2016, 1-10.	1.0	36
53	Genomic copy number alterations in nonâ€syndromic hearing loss. Clinical Genetics, 2016, 89, 473-477.	1.0	22
54	Genotypeâ€phenotype correlation of 16p13.3 terminal duplication and 22q13.33 deletion: Natural history of a patient and review of the literature. American Journal of Medical Genetics, Part A, 2016, 170, 766-772.	0.7	8

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55	LINE-1 hypomethylation and mutational status in cutaneous melanomas. Journal of Investigative Medicine, 2016, 64, 899-904.	0.7	10
56	Role of rare germline copy number variation in melanoma-prone patients. Future Oncology, 2016, 12, 1345-1357.	1.1	8
57	A genomic case study of desmoplastic small round cell tumor: comprehensive analysis reveals insights into potential therapeutic targets and development of a monitoring tool for a rare and aggressive disease. Human Genomics, 2016, 10, 36.	1.4	28
58	Evaluation of a subset of tumor suppressor gene for copy number and epigenitic changes in pleomorphic adenoma and carcinoma ex-pleomorphic adenoma carcinogenesis. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2016, 122, 322-331.	0.2	8
59	Carcinoma ex-pleomorphic adenoma derived from recurrent pleomorphic adenoma shows important difference by array CGH compared to recurrent pleomorphic adenoma without malignant transformation. Brazilian Journal of Otorhinolaryngology, 2016, 82, 687-694.	0.4	8
60	Genomic imbalances pinpoint potential oncogenes and tumor suppressors in Wilms tumors. Molecular Cytogenetics, 2016, 9, 20.	0.4	36
61	Role of CDKN2A Mutations and Other Relevant Genes in Melanoma Predisposition. , 2016, , 101-117.		0
62	Genomic copy number alterations of primary and secondary metastasizing pleomorphic adenomas. Histopathology, 2015, 67, 410-415.	1.6	6
63	DNA Methylation Levels of Melanoma Risk Genes Are Associated with Clinical Characteristics of Melanoma Patients. BioMed Research International, 2015, 2015, 1-8.	0.9	17
64	Co-expression network of neural-differentiation genes shows specific pattern in schizophrenia. BMC Medical Genomics, 2015, 8, 23.	0.7	45
65	Lymphovascular invasion and histologic grade are associated with specific genomic profiles in invasive carcinomas of the breast. Tumor Biology, 2015, 36, 1835-1848.	0.8	21
66	LINE-1 hypermethylation in peripheral blood of cutaneous melanoma patients is associated with metastasis. Melanoma Research, 2015, 25, 173-177.	0.6	20
67	An Inherited Small Microdeletion at 15q13.3 in a Patient with Early- Onset Obsessive-Compulsive Disorder. PLoS ONE, 2014, 9, e110198.	1.1	14
68	Stability of XIST repression in relation to genomic imprinting following global genome demethylation in a human cell line. Brazilian Journal of Medical and Biological Research, 2014, 47, 1029-1035.	0.7	2
69	Upregulated genes at 2q24 gains as candidate oncogenes in hepatoblastomas. Future Oncology, 2014, 10, 2449-2457.	1.1	29
70	Recurrent somatic mutation in DROSHA induces microRNA profile changes in Wilms tumour. Nature Communications, 2014, 5, 4039.	5.8	159
71	Large germline copy number variations as predisposing factor in childhood neoplasms. Future Oncology, 2014, 10, 1627-1633.	1.1	6
72	Genome-wide DNA methylation profile of leukocytes from melanoma patients with and without CDKN2A mutations. Experimental and Molecular Pathology, 2014, 97, 425-432.	0.9	5

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73	Germline CDKN2A mutations in Brazilian patients of hereditary cutaneous melanoma. Familial Cancer, 2014, 13, 645-649.	0.9	18
74	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.	2.6	40
75	Association of melanoma with intraepithelial neoplasia of the pancreas in three patients. Experimental and Molecular Pathology, 2014, 97, 144-147.	0.9	1
76	Clinical and genetic characterization of basal cell carcinoma and breast cancer in a single patient. SpringerPlus, 2014, 3, 454.	1.2	5
77	Does germ-line deletion of the PIP gene constitute a widespread risk for cancer?. European Journal of Human Genetics, 2014, 22, 307-309.	1.4	4
78	Hereditary breast and ovarian cancer: assessment of point mutations and copy number variations in Brazilian patients. BMC Medical Genetics, 2014, 15, 55.	2.1	57
79	The profile and contribution of rare germline copy number variants to cancer risk in Li-Fraumeni patients negative for TP53 mutations. Orphanet Journal of Rare Diseases, 2014, 9, 63.	1.2	12
80	Abstract 3418: Rare germline copy number variations in hereditary cutaneous melanoma. , 2014, , .		0
81	Mutational spectrum of the APC and MUTYH genes and genotypeâ€“phenotype correlations in Brazilian FAP, AFAP, and MAP patients. Orphanet Journal of Rare Diseases, 2013, 8, 54.	1.2	35
82	Single-nucleotide polymorphism-array improves detection rate of genomic alterations in core-binding factor leukemia. Medical Oncology, 2013, 30, 579.	1.2	6
83	Array-CGH as an adjuvant tool in cytogenetic diagnosis of pediatric MDS and JMML. Medical Oncology, 2013, 30, 734.	1.2	3
84	A microduplication of 5p15.33 reveals CLPTM1L as a candidate gene for cleft lip and palate. European Journal of Medical Genetics, 2013, 56, 222-225.	0.7	11
85	Molecular and clinical delineation of the 17q22 microdeletion phenotype. European Journal of Human Genetics, 2013, 21, 1085-1092.	1.4	11
86	Germline BAX Deletion in a Patient With Melanoma and Gastrointestinal Stromal Tumor. American Journal of Gastroenterology, 2013, 108, 1372-1375.	0.2	3
87	Complex Phenotype Associated with 17q21.31 Microdeletion. Molecular Syndromology, 2013, 4, 297-301.	0.3	11
88	Germline DNA copy number variation in individuals with Argyrophilic grain disease reveals CTNS as a plausible candidate gene. Genetics and Molecular Biology, 2013, 36, 498-501.	0.6	5
89	Comprehensive Analysis of BRCA1, BRCA2 and TP53 Germline Mutation and Tumor Characterization: A Portrait of Early-Onset Breast Cancer in Brazil. PLoS ONE, 2013, 8, e57581.	1.1	70
90	Abstract A025: Screening for genomic rearrangements and germline mutations in BRCA1 and BRCA2 genes in hereditary breast cancer unrelated Brazilian families. , 2013, , .		0

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91	A novel microdeletion syndrome at 3q13.31 characterised by developmental delay, postnatal overgrowth, hypoplastic male genitals, and characteristic facial features. Journal of Medical Genetics, 2012, 49, 104-109.	1.5	46
92	A familial case with interstitial 2q36 deletion: Variable phenotypic expression in full and mosaic state. European Journal of Medical Genetics, 2012, 55, 660-665.	0.7	7
93	Down-regulation of ANAPC13 and CLTCL1: Early Events in the Progression of Preinvasive Ductal Carcinoma of the Breast. Translational Oncology, 2012, 5, 113-IN8.	1.7	13
94	Germline copy number variations and cancer predisposition. Future Oncology, 2012, 8, 441-450.	1.1	73
95	Deletion of the RMGA and CHD2 genes in a child with epilepsy and mental deficiency. European Journal of Medical Genetics, 2012, 55, 132-134.	0.7	42
96	A novel SYBR-based duplex qPCR for the detection of gene dosage: detection of an APC large deletion in a familial adenomatous polyposis patient with an unusual phenotype. BMC Medical Genetics, 2012, 13, 55.	2.1	12
97	Li-Fraumeni-like syndrome associated with a large BRCA1 intragenic deletion. BMC Cancer, 2012, 12, 237.	1.1	25
98	Number of rare germline CNVs and TP53 mutation types. Orphanet Journal of Rare Diseases, 2012, 7, 101.	1.2	9
99	Germline DNA copy number variation in familial and early-onset breast cancer. Breast Cancer Research, 2012, 14, R24.	2.2	76
100	Abstract 1168: Identification of CNA signatures in prostate cancer: Narrowing chromosome regions related with occurrence, prognosis and recurrence after treatment. , 2012, , .		1
101	Breakpoint characterization of a novel large intragenic deletion of MUTYH detected in a MAP patient: Case report. BMC Medical Genetics, 2011, 12, 128.	2.1	18
102	5q12.1 deletion: Delineation of a phenotype including mental retardation and ocular defects. , 2011, 155, 725-731.		20
103	Maternally inherited partial monosomy 9p (pterâ€‰â†’â€‰p24.1) and partial trisomy 20p (pterâ€‰â†’â€‰p12.1) characterized by microarray comparative genomic hybridization. American Journal of Medical Genetics, Part A, 2011, 155, 2754-2761.	0.7	13
104	Abstract PR14: Germline submicroscopic chromosome imbalances in pediatric cancer. , 2011, , .		0
105	Abstract B10: Germline copy number variation in Li-Fraumeni syndrome patients with TP53 mutations. , 2011, , .		0
106	Abstract A53: Genome-wide profile of somatic copy number alterations in Wilms tumor: Comparison between samples derived from patients with and without relapse. , 2011, , .		0
107	A novel de novo microdeletion spanning the SYNGAP1 gene on the short arm of chromosome 6 associated with mental retardation. American Journal of Medical Genetics, Part A, 2010, 152A, 2376-2378.	0.7	26
108	Two distinct regions in 2q24.2â€q24.3 associated with idiopathic epilepsy. Epilepsia, 2010, 51, 2457-2460.	2.6	43

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109	Deletions encompassing 1q41q42.1 and clinical features of autosomal dominant Robinow syndrome. Clinical Genetics, 2010, 77, 404-407.	1.0	14
110	Constitutional Haploinsufficiency of Tumor Suppressor Genes in Mentally Retarded Patients With Microdeletions in 17p13.1. Cytogenetic and Genome Research, 2009, 125, 1-7.	0.6	32
111	Chromosome imbalances in syndromic hearing loss. Clinical Genetics, 2009, 76, 458-464.	1.0	4
112	High frequency of submicroscopic chromosomal imbalances in patients with syndromic craniosynostosis detected by a combined approach of microsatellite segregation analysis, multiplex ligation-dependent probe amplification and array-based comparative genome hybridisation. Journal of Medical Genetics, 2008, 45, 447-450.	1.5	36
113	Clinical and molecular delineation of the 17q21.31 microdeletion syndrome. Journal of Medical Genetics, 2008, 45, 710-720.	1.5	191
114	Array-CGH testing in spontaneous abortions with normal karyotypes. Genetics and Molecular Biology, 2008, 31, 416-422.	0.6	8
115	Genomic imbalances associated with mullerian aplasia. Journal of Medical Genetics, 2007, 45, 228-232.	1.5	110
116	Chromosome abnormalities in two patients with features of autosomal dominant Robinow syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1790-1795.	0.7	11
117	An 11q11â€“q13.3 duplication, including FGF3 and FGF4 genes, in a patient with syndromic multiple craniosynostoses. American Journal of Medical Genetics, Part A, 2007, 143A, 1912-1918.	0.7	25
118	Array CGH identifies reciprocal 16p13.1 duplications and deletions that predispose to autism and/or mental retardation. Human Mutation, 2007, 28, 674-682.	1.1	263
119	Widening the clinical spectrum of Pitt-Rogers-Danks/Wolf-Hirschhorn syndromes. Genetics and Molecular Biology, 2007, 30, 339-342.	0.6	2
120	Whole-genome array-CGH screening in undiagnosed syndromic patients: old syndromes revisited and new alterations. Cytogenetic and Genome Research, 2006, 115, 254-261.	0.6	103
121	Microdeletion encompassing MAPT at chromosome 17q21.3 is associated with developmental delay and learning disability. Nature Genetics, 2006, 38, 1032-1037.	9.4	344
122	Report of a del22q11 in a patient with Mayer-Rokitansky-KÃ¼ster-Hauser (MRKH) anomaly and exclusion ofWNT-4,RAR-gamma, andRXR-alpha as major genes determining MRKH anomaly in a study of 25 affected women. American Journal of Medical Genetics, Part A, 2006, 140A, 1339-1342.	0.7	64
123	A 17q21.31 microdeletion encompassing the MAPT gene in a mentally impaired patient. Cytogenetic and Genome Research, 2006, 114, 89-92.	0.6	45
124	An Xq22.3 duplication detected by comparative genomic hybridization microarray (Array-CGH) defines a new locus (FGS5) for FG syndrome. American Journal of Medical Genetics, Part A, 2005, 139A, 221-226.	0.7	32
125	Array-CGH detection of micro rearrangements in mentally retarded individuals: clinical significance of imbalances present both in affected children and normal parents. Journal of Medical Genetics, 2005, 43, 180-186.	1.5	190
126	Disclosing the mechanisms of origin of de novo short-arm duplications of chromosome 9. American Journal of Medical Genetics Part A, 2003, 117A, 41-46.	2.4	23

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127	Deletion of the factor IX gene as a result of translocation t(X;1) in a girl affected by haemophilia B. British Journal of Haematology, 2001, 113, 616-620.	1.2	10
128	X chromosome-inactivation patterns in patients with Rett syndrome. Human Genetics, 1998, 102, 319-321.	1.8	27