Isabel Marques Carreira

List of Publications by Year
in descending order

117	3,018	159585 30	182427 51
papers	citations	h-index	g-index

117	117	117	4969
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Zinc Prevents DNA Damage in Normal Cells but Shows Genotoxic and Cytotoxic Effects in Acute Myeloid Leukemia Cells. International Journal of Molecular Sciences, 2022, 23, 2567.	4.1	5
2	Recommendations for reporting results of diagnostic genomic testing. European Journal of Human Genetics, 2022, 30, 1011-1016.	2.8	15
3	Prevalence of cytogenetic abnormalities and FMR1 gene premutation in a Portuguese population with premature ovarian insufficiency. Acta Medica Portuguesa, 2021, 34, 580-585.	0.4	2
4	Basal cell carcinomas of the scalp after radiotherapy for tinea capitis in childhood: A genetic and epigenetic study with comparison with basal cell carcinomas evolving in chronically sunâ€exposed areas. Experimental Dermatology, 2021, 30, 1126-1134.	2.9	5
5	Multiple Basal Cell Carcinomas of the Scalp After Radiotherapy: Genomic Study in a Case With Latency Period Over 80 Years. American Journal of Dermatopathology, 2021, 43, 438-442.	0.6	1
6	Cancro da CabeÃ§a e PescoÃ§o: Aspectos Particulares do Cancro Oral. , 2021, , .		0
7	Liquid Biopsies: Applications for Cancer Diagnosis and Monitoring. Genes, 2021, 12, 349.	2.4	93
8	Genomic-Metabolomic Associations Support the Role of LIPC and Glycerophospholipids in Age-Related Macular Degeneration. Ophthalmology Science, 2021, 1, 100017.	2.5	7
9	Genomic characterisation of multiple myeloma: study of a Portuguese cohort. Journal of Clinical Pathology, 2021, , jclinpath-2020-207204.	2.0	1
10	Development of a genomic predictive model for cholangiocarcinoma using copy number alteration data. Journal of Clinical Pathology, 2021, , jclinpath-2020-207346.	2.0	1
11	A seven-gene signature to predict the prognosis of oral squamous cell carcinoma. Oncogene, 2021, 40, 3859-3869.	5.9	11
12	Should sitting time be a treatment target in head and neck cancer patients receiving curative treatment?. Oral Oncology, 2021, 124, 105418.	1.5	1
13	Chromosomal breakpoints in a cohort of head and neck squamous cell carcinoma patients. Genomics, 2020, 112, 297-303.	2.9	9
14	An acquired stable variant of a dicentric dic(9;20) and complex karyotype in a Syrian childhood B-acute lymphoblastic leukemia case. Molecular Cytogenetics, 2020, 13, 29.	0.9	1
15	Intratumoral Heterogeneity in Uveal Melanoma. Ocular Oncology and Pathology, 2020, 7, 1-9.	1.0	0
16	A new childhood ALL case with an extremely complex karyotype and acute spontaneous tumor lysis syndrome. Molecular Cytogenetics, 2020, 13, 44.	0.9	0
17	Tremor is a major feature of 9p13 deletion syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2694-2698.	1.2	1
18	Probability distribution of copy number alterations along the genome: an algorithm to distinguish different tumour profiles. Scientific Reports, 2020, 10, 14868.	3.3	8

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19	Proteomics-based Predictive Model for the Early Detection of Metastasis and Recurrence in Head and Neck Cancer. Cancer Genomics and Proteomics, 2020, 17, 259-269.	2.0	10
20	Complex karyotype with cryptic FUS gene rearrangement and deletion of NR3C1 and VPREB1 genes in childhood Bâ€'cell acute lymphoblastic leukemia: A case report. Oncology Letters, 2020, 19, 2957-2962.	1.8	2
21	Iodineâ€'131 metabolic radiotherapy leads to cell death and genomic alterations through NIS overexpression on cholangiocarcinoma. International Journal of Oncology, 2020, 56, 709-727.	3.3	3
22	Upper aerodigestive tract carcinoma: Development of a (epi)genomic predictive model for recurrence and metastasis. Oncology Letters, 2020, 19, 3459-3468.	1.8	2
23	Head and neck cancer: searching for genomic and epigenetic biomarkers in body fluids â€" the state of art. Molecular Cytogenetics, 2019, 12, 33.	0.9	22
24	(Cyto)genomic and epigenetic characterization of BICR 10 cell line and three new established primary human head and neck squamous cell carcinoma cultures. Genes and Genomics, 2019, 41, 1207-1221.	1.4	2
25	Cytogenetics and Cytogenomics Evaluation in Cancer. International Journal of Molecular Sciences, 2019, 20, 4711.	4.1	14
26	Urine Nuclear Magnetic Resonance (NMR) Metabolomics in Age-Related Macular Degeneration. Journal of Proteome Research, 2019, 18, 1278-1288.	3.7	15
27	A New Complex Karyotype Involving a <i>KMT2A</i>-r Variant Three-Way Translocation in a Rare Clinical Presentation of a Pediatric Patient with Acute Myeloid Leukemia. Cytogenetic and Genome Research, 2019, 157, 213-219.	1.1	0
28	Regarding the rights and duties of Clinical Laboratory Geneticists in genetic healthcare systems; results of a survey in over 50 countries. European Journal of Human Genetics, 2019, 27, 1168-1174.	2.8	12
29	Molecular approaches identify a cryptic MECOM rearrangement in a child with a rapidly progressive myeloid neoplasm. Cancer Genetics, 2018, 221, 25-30.	0.4	7
30	Generation and characterization of a human iPS cell line from a patient-related control to study disease mechanisms associated with DAND5 p.R152H alteration. Stem Cell Research, 2018, 29, 202-206.	0.7	2
31	Cytogenetic, genomic, and epigenetic characterization of the HSC-3 tongue cell line with lymph node metastasis. Journal of Oral Science, 2018, 60, 70-81.	1.7	9
32	Genomic and epigenetic signatures associated with survival rate in oral squamous cell carcinoma patients. Journal of Cancer, 2018, 9, 1885-1895.	2.5	23
33	Stroma-derived IL-6, G-CSF and Activin-A mediated dedifferentiation of lung carcinoma cells into cancer stem cells. Scientific Reports, 2018, 8, 11573.	3.3	26
34	Cryptic NUP214-ABL1 fusion with complex karyotype, episomes and intra-tumor genetic heterogeneity in a T-cell lymphoblastic lymphoma. Journal of Cancer Metastasis and Treatment, 2018, 4, 50.	0.8	0
35	European registration process for Clinical Laboratory Geneticists in genetic healthcare. European Journal of Human Genetics, 2017, 25, 515-519.	2.8	13
36	Genomic predictive model for recurrence and metastasis development in head and neck squamous cell carcinoma patients. Scientific Reports, 2017, 7, 13897.	3.3	38

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37	Genomic profile of oral squamous cell carcinomas with an adjacent leukoplakia or with an erythroleukoplakia that evolved after the treatment of primary tumor: A report of two cases. Molecular Medicine Reports, 2017, 16, 6780-6786.	2.4	11
38	Genomic and epigenetic characterization for the comparison of synchronous bilateral tongue squamous cell carcinomasâ€"A case report. Current Problems in Cancer, 2017, 41, 398-406.	2.0	1
39	Generation of human iPSC line from a patient with laterality defects and associated congenital heart anomalies carrying a DAND5 missense alteration. Stem Cell Research, 2017, 25, 152-156.	0.7	11
40	MLPA analysis in a cohort of patients with autism. Molecular Cytogenetics, 2017, 10, 2.	0.9	1
41	Genetic and epigenetic characterization of the tumors in a patient with a tongue primary tumor, a recurrence and a pharyngoesophageal second primary tumor. Molecular Cytogenetics, 2017, 10, 13.	0.9	0
42	Iodine deficiency a persisting problem: assessment of iodine nutrition and evaluation of thyroid nodular pathology in Portugal. Journal of Endocrinological Investigation, 2017, 40, 185-191.	3.3	9
43	Effects of resistance exercise on endothelial progenitor cell mobilization in women. Scientific Reports, 2017, 7, 17880.	3.3	41
44	Human plasma metabolomics in age-related macular degeneration (AMD) using nuclear magnetic resonance spectroscopy. PLoS ONE, 2017, 12, e0177749.	2.5	51
45	Early detection and personalized treatment in oral cancer: the impact of omics approaches. Molecular Cytogenetics, 2016, 9, 85.	0.9	33
46	BIRC3 alterations in chronic and B-cell acute lymphocytic leukemia patients. Oncology Letters, 2016, 11, 3240-3246.	1.8	13
47	WT1, MSH6, GATA5 and PAX5 as epigenetic oral squamous cell carcinoma biomarkers - a short report. Cellular Oncology (Dordrecht), 2016, 39, 573-582.	4.4	31
48	A novel IGH@ gene rearrangement associated with CDKN2A/B deletion in young adult B-cell acute lymphoblastic leukemia. Oncology Letters, 2016, 11, 2117-2122.	1.8	4
49	Fibroblasts of Machado Joseph Disease patients reveal autophagy impairment. Scientific Reports, 2016, 6, 28220.	3.3	68
50	Metabolic profiling of maternal urine can aid clinical management of gestational diabetes mellitus. Metabolomics, 2016, 12, 1.	3.0	9
51	Newborn Urinary Metabolic Signatures of Prematurity and Other Disorders: A Case Control Study. Journal of Proteome Research, 2016, 15, 311-325.	3.7	24
52	High rates of submicroscopic aberrations in karyotypically normal acute lymphoblastic leukemia. Molecular Cytogenetics, 2015, 8, 45.	0.9	17
53	Copy number variants prioritization after array-CGH analysis â€“ a cohort of 1000 patients. Molecular Cytogenetics, 2015, 8, 103.	0.9	17
54	Isochromosome 17q in Chronic Lymphocytic Leukemia. Leukemia Research and Treatment, 2015, 2015, 1-6.	2.0	1

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55	Following Healthy Pregnancy by NMR Metabolomics of Plasma and Correlation to Urine. Journal of Proteome Research, 2015, 14, 1263-1274.	3.7	72
56	MLLT10 and IL3 rearrangement together with a complex four-way translocation and trisomy 4 in a patient with early T-cell precursor acute lymphoblastic leukemia: A case report. Oncology Reports, 2015, 33, 625-630.	2.6	4
57	Drug transporters play a key role in the complex process of Imatinib resistance in vitro. Leukemia Research, 2015, 39, 355-360.	0.8	18
58	Oculo-auriculo-vertebral spectrum: Clinical and molecular analysis of 51 patients. European Journal of Medical Genetics, 2015, 58, 455-465.	1.3	83
59	Impact of fetal chromosomal disorders on maternal blood metabolome: toward new biomarkers?. American Journal of Obstetrics and Gynecology, 2015, 213, 841.e1-841.e15.	1.3	18
60	Cutis Aplasia as a clinical hallmark for the syndrome associated with 19q13.11 deletion: the possible role for UBA2 gene. Molecular Cytogenetics, 2015, 8, 21.	0.9	20
61	Prediction of Gestational Diabetes through NMR Metabolomics of Maternal Blood. Journal of Proteome Research, 2015, 14, 2696-2706.	3.7	70
62	Novel Cryptic Rearrangements in Adult B-Cell Precursor Acute Lymphoblastic Leukemia Involving the MLL Gene. Journal of Histochemistry and Cytochemistry, 2015, 63, 384-390.	2.5	7
63	12q21.2q22 deletion: A new patient. American Journal of Medical Genetics, Part A, 2015, 167, 1877-1883.	1.2	7
64	Iodine deficiency and thyroid nodular pathology - epidemiological and cancer characteristics in different populations: Portugal and South Africa. BMC Research Notes, 2015, 8, 284.	1.4	8
65	A unique phenotype in a patient with a rare triplication of the 22q11.2 region and new clinical insights of the 22q11.2 microduplication syndrome: a report of two cases. BMC Pediatrics, 2015, 15, 95.	1.7	12
66	Senescent bronchial fibroblasts induced to senescence by Cr(VI) promote epithelialâ€“mesenchymal transition when co-cultured with bronchial epithelial cells in the presence of Cr(VI). Mutagenesis, 2015, 30, 277-286.	2.6	9
67	NMR metabolomics of human lung tumours reveals distinct metabolic signatures for adenocarcinoma and squamous cell carcinoma. Carcinogenesis, 2015, 36, 68-75.	2.8	75
68	Interstitial 287Âkb deletion of 4p16.3 including FGFRL1 gene associated with language impairment and overgrowth. Molecular Cytogenetics, 2014, 7, 87.	0.9	5
69	Screening of copy number variants in the 22q11.2 region of congenital heart disease patients from the SÃ£o Miguel Island, Azores, revealed the second patient with a triplication. BMC Genetics, 2014, 15, 115.	2.7	13
70	A Novel Cryptic Three-Way Translocation t(2;9;18)(p23.2;p21.3;q21.33) with Deletion of Tumor Suppressor Genes in 9p21.3 and 13q14 in a T-Cell Acute Lymphoblastic Leukemia. Leukemia Research and Treatment, 2014, 2014, 1-7.	2.0	7
71	Genetic gains and losses in oral squamous cell carcinoma: impact on clinical management. Cellular Oncology (Dordrecht), 2014, 37, 29-39.	4.4	46
72	Genetic imbalances detected by multiplex ligation-dependent probe amplification in a cohort of patients with oral squamous cell carcinomaâ€”the first step towards clinical personalized medicine. Tumor Biology, 2014, 35, 4687-95.	1.8	22

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73	Genomic characterization of three urinary bladder cancer cell lines: understanding genomic types of urinary bladder cancer. Tumor Biology, 2014, 35, 4599-4617.	1.8	33
74	Human plasma stability during handling and storage: impact on NMR metabolomics. Analyst, The, 2014, 139, 1168-1177.	3.5	139
75	Neurodevelopmental disorders associated with dosage imbalance ofZBTB20correlate with the morbidity spectrum of ZBTB20 candidate target genes. Journal of Medical Genetics, 2014, 51, 605-613.	3.2	26
76	Maternal plasma phospholipids are altered in trisomy 21 cases and prior to preeclampsia and preterm outcomes. Rapid Communications in Mass Spectrometry, 2014, 28, 1635-1638.	1.5	14
77	Potential Markers of Cisplatin Treatment Response Unveiled by NMR Metabolomics of Human Lung Cells. Molecular Pharmaceutics, 2013, 10, 4242-4251.	4.6	39
78	Mosaicism for FMR1 gene full mutation and intermediate allele in a female foetus: A postzygotic retraction event. Gene, 2013, 527, 421-425.	2.2	11
79	Following Healthy Pregnancy by Nuclear Magnetic Resonance (NMR) Metabolic Profiling of Human Urine. Journal of Proteome Research, 2013, 12, 969-979.	3.7	50
80	Insertional translocation leading to a 4q13 duplication including the EPHA5 gene in two siblings with attentionâ€deficit hyperactivity disorder. American Journal of Medical Genetics, Part A, 2013, 161, 1923-1928.	1.2	14
81	Mid-infrared (MIR) metabolic fingerprinting of amniotic fluid: A possible avenue for early diagnosis of prenatal disorders?. Analytica Chimica Acta, 2013, 764, 24-31.	5.4	26
82	Cryptic 7q36.2q36.3 deletion causes multiple congenital eye anomalies and craniofacial dysmorphism. American Journal of Medical Genetics, Part A, 2013, 161, 589-593.	1.2	9
83	Second Trimester Maternal Urine for the Diagnosis of Trisomy 21 and Prediction of Poor Pregnancy Outcomes. Journal of Proteome Research, 2013, 12, 2946-2957.	3.7	68
84	Inv21p12q22del21q22 and intellectual disability. Gene, 2013, 517, 120-124.	2.2	0
85	Â thalassemia major due to acquired uniparental disomy in a previously healthy adolescent. Haematologica, 2013, 98, e4-e6.	3.5	8
86	High resolution melting: improvements in the genetic diagnosis of hypertrophic cardiomyopathy in a Portuguese cohort. BMC Medical Genetics, 2012, 13, 17.	2.1	30
87	Critical region in 2q31.2q32.3 deletion syndrome: Report of two phenotypically distinct patients, one with an additional deletion in Alagille syndrome region. Molecular Cytogenetics, 2012, 5, 25.	0.9	8
88	Can Biofluids Metabolic Profiling Help to Improve Healthcare during Pregnancy?. Spectroscopy, 2012, 27, 515-523.	0.8	10
89	UPLC-MS metabolic profiling of second trimester amniotic fluid and maternal urine and comparison with NMR spectral profiling for the identification of pregnancy disorder biomarkers. Molecular BioSystems, 2012, 8, 1243.	2.9	94
90	Metabolic Signatures of Lung Cancer in Biofluids: NMR-Based Metabonomics of Urine. Journal of Proteome Research, 2011, 10, 221-230.	3.7	205

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91	Metabolic Signatures of Lung Cancer in Biofluids: NMR-Based Metabonomics of Blood Plasma. Journal of Proteome Research, 2011, 10, 4314-4324.	3.7	154
92	Metabolic Biomarkers of Prenatal Disorders: An Exploratory NMR Metabonomics Study of Second Trimester Maternal Urine and Blood Plasma. Journal of Proteome Research, 2011, 10, 3732-3742.	3.7	144
93	Chromosome 5 derived small supernumerary marker: towards a genotype/phenotype correlation of proximal chromosome 5 imbalances. Journal of Applied Genetics, 2011, 52, 193-200.	1.9	13
94	NMR metabonomic study of lung cancer: metabolic profiling of tissues. BMC Proceedings, 2010, 4, .	1.6	0
95	Can nuclear magnetic resonance (NMR) spectroscopy reveal different metabolic signatures for lung tumours?. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2010, 457, 715-725.	2.8	34
96	X-chromosome terminal deletion in a female with premature ovarian failure: Haploinsufficiency of X-linked genes as a possible explanation. Molecular Cytogenetics, 2010, 3, 14.	0.9	24
97	Lateâ€onset hyperpigmentation: a case with multiâ€systemic involvement and recombinant X chromosome. Journal of the European Academy of Dermatology and Venereology, 2010, 24, 84-85.	2.4	1
98	Metabolic Profiling of Human Lung Cancer Tissue by 1H High Resolution Magic Angle Spinning (HRMAS) NMR Spectroscopy. Journal of Proteome Research, 2010, 9, 319-332.	3.7	136
99	Impact of Prenatal Disorders on the Metabolic Profile of Second Trimester Amniotic Fluid: A Nuclear Magnetic Resonance Metabonomic Study. Journal of Proteome Research, 2010, 9, 6016-6024.	3.7	94
100	Molecular Cytogenetic Characterization of Two Cases with de novo Small Mosaic Supernumerary Marker Chromosomes Derived from Chromosome 16: Towards a Genotype/Phenotype Correlation. Cytogenetic and Genome Research, 2009, 125, 109-114.	1.1	5
101	Human bronchial epithelial cells malignantly transformed by hexavalent chromium exhibit an aneuploid phenotype but no microsatellite instability. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2009, 670, 42-52.	1.0	45
102	Molecular cytogenetic characterisation of a mosaic add(12)(p13.3) with an inv dup(3)(q26.31 â†' qter) detected in an autistic boy. Molecular Cytogenetics, 2009, 2, 16.	0.9	6
103	¹H NMR Based Metabonomics of Human Amniotic Fluid for the Metabolic Characterization of Fetus Malformations. Journal of Proteome Research, 2009, 8, 4144-4150.	3.7	62
104	Analytical Approaches toward Successful Human Cell Metabolome Studies by NMR Spectroscopy. Analytical Chemistry, 2009, 81, 5023-5032.	6.5	61
105	Tetraâ€amelia and lung hypo/aplasia syndrome: New case report and review. American Journal of Medical Genetics, Part A, 2008, 146A, 2799-2803.	1.2	18
106	Mowatâ€“Wilson syndrome: an underdiagnosed syndrome?. Clinical Genetics, 2008, 73, 579-584.	2.0	15
107	Metabolite Profiling of Human Amniotic Fluid by Hyphenated Nuclear Magnetic Resonance Spectroscopy. Analytical Chemistry, 2008, 80, 6085-6092.	6.5	46
108	First prenatally detected small supernumerary neocentromeric derivative chromosome 13 resulting in a non-mosaic partial tetrasomy 13q. Cytogenetic and Genome Research, 2008, 121, 293-297.	1.1	8

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109	Three Unusual but Cytogenetically Similar Cases With up to Five Different Cell Lines Involving Structural and Numerical Abnormalities of Chromosome 18. Journal of Histochemistry and Cytochemistry, 2007, 55, 1123-1128.	2.5	13
110	Null mutations and lethal congenital form of glycogen storage disease type IV. Biochemical and Biophysical Research Communications, 2007, 361, 445-450.	2.1	29
111	Potential of NMR Spectroscopy for the Study of Human Amniotic Fluid. Analytical Chemistry, 2007, 79, 8367-8375.	6.5	35
112	Two new cases ofde novo small supernumerary marker chromosomes (sSMC) detected at prenatal diagnosis. Prenatal Diagnosis, 2007, 27, 380-381.	2.3	7
113	Cytogenetically invisible microdeletions involving PITX2 in Rieger syndrome. Clinical Genetics, 2007, 72, 464-470.	2.0	15
114	Mitochondrial DNA Variants in a Portuguese Population of Patients with Alzheimerâ€™s Disease. European Neurology, 2005, 53, 121-124.	1.4	9
115	Partial tetrasomy of chromosome 3q and mosaicism in a child with autism. Journal of Autism and Developmental Disorders, 2003, 33, 177-185.	2.7	12
116	Novel mutations and polymorphisms in the Fanconi anemia group C gene. Human Mutation, 1996, 8, 140-148.	2.5	42
117	Presence of Y chromosome sequences and their effect on the phenotype of six patients with Y chromosome anomalies. American Journal of Medical Genetics Part A, 1995, 55, 269-275.	2.4	7