

Isabel Marques Carreira

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/881409/publications.pdf>

Version: 2024-02-01

117
papers

3,018
citations

159525

30
h-index

182361

51
g-index

117
all docs

117
docs citations

117
times ranked

4969
citing authors

#	ARTICLE	IF	CITATIONS
1	Zinc Prevents DNA Damage in Normal Cells but Shows Genotoxic and Cytotoxic Effects in Acute Myeloid Leukemia Cells. <i>International Journal of Molecular Sciences</i> , 2022, 23, 2567.	1.8	5
2	Recommendations for reporting results of diagnostic genomic testing. <i>European Journal of Human Genetics</i> , 2022, 30, 1011-1016.	1.4	15
3	Prevalence of cytogenetic abnormalities and FMR1 gene premutation in a Portuguese population with premature ovarian insufficiency. <i>Acta Medica Portuguesa</i> , 2021, 34, 580-585.	0.2	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 sun-exposed areas. <i>Experimental Dermatology</i> , 2021, 30, 1126-1134.	1.4	5
5	Multiple Basal Cell Carcinomas of the Scalp After Radiotherapy: Genomic Study in a Case With Latency Period Over 80 Years. <i>American Journal of Dermatopathology</i> , 2021, 43, 438-442.	0.3	1
6	Cancro da Cabeça e Pescoço: Aspectos Particulares do Cancro Oral. , 2021, , .		0
7	Liquid Biopsies: Applications for Cancer Diagnosis and Monitoring. <i>Genes</i> , 2021, 12, 349.	1.0	93
8	Genomic-Metabolomic Associations Support the Role of LPC and Glycerophospholipids in Age-Related Macular Degeneration. <i>Ophthalmology Science</i> , 2021, 1, 100017.	1.0	7
9	Genomic characterisation of multiple myeloma: study of a Portuguese cohort. <i>Journal of Clinical Pathology</i> , 2021, , jclinpath-2020-207204.	1.0	1
10	Development of a genomic predictive model for cholangiocarcinoma using copy number alteration data. <i>Journal of Clinical Pathology</i> , 2021, , jclinpath-2020-207346.	1.0	1
11	A seven-gene signature to predict the prognosis of oral squamous cell carcinoma. <i>Oncogene</i> , 2021, 40, 3859-3869.	2.6	11
12	Should sitting time be a treatment target in head and neck cancer patients receiving curative treatment?. <i>Oral Oncology</i> , 2021, 124, 105418.	0.8	1
13	Chromosomal breakpoints in a cohort of head and neck squamous cell carcinoma patients. <i>Genomics</i> , 2020, 112, 297-303.	1.3	9
14	An acquired stable variant of a dicentric dic(9;20) and complex karyotype in a Syrian childhood B-acute lymphoblastic leukemia case. <i>Molecular Cytogenetics</i> , 2020, 13, 29.	0.4	1
15	Intratumoral Heterogeneity in Uveal Melanoma. <i>Ocular Oncology and Pathology</i> , 2020, 7, 1-9.	0.5	0
16	A new childhood ALL case with an extremely complex karyotype and acute spontaneous tumor lysis syndrome. <i>Molecular Cytogenetics</i> , 2020, 13, 44.	0.4	0
17	Tremor is a major feature of 9p13 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2694-2698.	0.7	1
18	Probability distribution of copy number alterations along the genome: an algorithm to distinguish different tumour profiles. <i>Scientific Reports</i> , 2020, 10, 14868.	1.6	8

#	ARTICLE	IF	CITATIONS
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. <i>Molecular Medicine Reports</i> , 2017, 16, 6780-6786.	1.1	11
38	Genomic and epigenetic characterization for the comparison of synchronous bilateral tongue squamous cell carcinomas – A case report. <i>Current Problems in Cancer</i> , 2017, 41, 398-406.	1.0	1
39	Generation of human iPSC line from a patient with laterality defects and associated congenital heart anomalies carrying a DAND5 missense alteration. <i>Stem Cell Research</i> , 2017, 25, 152-156.	0.3	11
40	MLPA analysis in a cohort of patients with autism. <i>Molecular Cytogenetics</i> , 2017, 10, 2.	0.4	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. <i>Molecular Cytogenetics</i> , 2017, 10, 13.	0.4	0
42	Iodine deficiency a persisting problem: assessment of iodine nutrition and evaluation of thyroid nodular pathology in Portugal. <i>Journal of Endocrinological Investigation</i> , 2017, 40, 185-191.	1.8	9
43	Effects of resistance exercise on endothelial progenitor cell mobilization in women. <i>Scientific Reports</i> , 2017, 7, 17880.	1.6	41
44	Human plasma metabolomics in age-related macular degeneration (AMD) using nuclear magnetic resonance spectroscopy. <i>PLoS ONE</i> , 2017, 12, e0177749.	1.1	51
45	Early detection and personalized treatment in oral cancer: the impact of omics approaches. <i>Molecular Cytogenetics</i> , 2016, 9, 85.	0.4	33
46	BIRC3 alterations in chronic and B-cell acute lymphocytic leukemia patients. <i>Oncology Letters</i> , 2016, 11, 3240-3246.	0.8	13
47	WT1, MSH6, GATA5 and PAX5 as epigenetic oral squamous cell carcinoma biomarkers - a short report. <i>Cellular Oncology (Dordrecht)</i> , 2016, 39, 573-582.	2.1	31
48	A novel IGH@ gene rearrangement associated with CDKN2A/B deletion in young adult B-cell acute lymphoblastic leukemia. <i>Oncology Letters</i> , 2016, 11, 2117-2122.	0.8	4
49	Fibroblasts of Machado Joseph Disease patients reveal autophagy impairment. <i>Scientific Reports</i> , 2016, 6, 28220.	1.6	68
50	Metabolic profiling of maternal urine can aid clinical management of gestational diabetes mellitus. <i>Metabolomics</i> , 2016, 12, 1.	1.4	9
51	Newborn Urinary Metabolic Signatures of Prematurity and Other Disorders: A Case Control Study. <i>Journal of Proteome Research</i> , 2016, 15, 311-325.	1.8	24
52	High rates of submicroscopic aberrations in karyotypically normal acute lymphoblastic leukemia. <i>Molecular Cytogenetics</i> , 2015, 8, 45.	0.4	17
53	Copy number variants prioritization after array-CGH analysis – a cohort of 1000 patients. <i>Molecular Cytogenetics</i> , 2015, 8, 103.	0.4	17
54	Isochromosome 17q in Chronic Lymphocytic Leukemia. <i>Leukemia Research and Treatment</i> , 2015, 2015, 1-6.	2.0	1

#	ARTICLE	IF	CITATIONS
55	Following Healthy Pregnancy by NMR Metabolomics of Plasma and Correlation to Urine. <i>Journal of Proteome Research</i> , 2015, 14, 1263-1274.	1.8	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. <i>Oncology Reports</i> , 2015, 33, 625-630.	1.2	4
57	Drug transporters play a key role in the complex process of Imatinib resistance in vitro. <i>Leukemia Research</i> , 2015, 39, 355-360.	0.4	18
58	Oculo-auriculo-vertebral spectrum: Clinical and molecular analysis of 51 patients. <i>European Journal of Medical Genetics</i> , 2015, 58, 455-465.	0.7	83
59	Impact of fetal chromosomal disorders on maternal blood metabolome: toward new biomarkers?. <i>American Journal of Obstetrics and Gynecology</i> , 2015, 213, 841.e1-841.e15.	0.7	18
60	Cutis Aplasia as a clinical hallmark for the syndrome associated with 19q13.11 deletion: the possible role for UBA2 gene. <i>Molecular Cytogenetics</i> , 2015, 8, 21.	0.4	20
61	Prediction of Gestational Diabetes through NMR Metabolomics of Maternal Blood. <i>Journal of Proteome Research</i> , 2015, 14, 2696-2706.	1.8	70
62	Novel Cryptic Rearrangements in Adult B-Cell Precursor Acute Lymphoblastic Leukemia Involving the MLL Gene. <i>Journal of Histochemistry and Cytochemistry</i> , 2015, 63, 384-390.	1.3	7
63	12q21.2q22 deletion: A new patient. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1877-1883.	0.7	7
64	Iodine deficiency and thyroid nodular pathology - epidemiological and cancer characteristics in different populations: Portugal and South Africa. <i>BMC Research Notes</i> , 2015, 8, 284.	0.6	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. <i>BMC Pediatrics</i> , 2015, 15, 95.	0.7	12
66	Senescent bronchial fibroblasts induced to senescence by Cr(VI) promote epithelial to mesenchymal transition when co-cultured with bronchial epithelial cells in the presence of Cr(VI). <i>Mutagenesis</i> , 2015, 30, 277-286.	1.0	9
67	NMR metabolomics of human lung tumours reveals distinct metabolic signatures for adenocarcinoma and squamous cell carcinoma. <i>Carcinogenesis</i> , 2015, 36, 68-75.	1.3	75
68	Interstitial 287 kb deletion of 4p16.3 including FGFRL1 gene associated with language impairment and overgrowth. <i>Molecular Cytogenetics</i> , 2014, 7, 87.	0.4	5
69	Screening of copy number variants in the 22q11.2 region of congenital heart disease patients from the S�o Miguel Island, Azores, revealed the second patient with a triplication. <i>BMC Genetics</i> , 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. <i>Leukemia Research and Treatment</i> , 2014, 2014, 1-7.	2.0	7
71	Genetic gains and losses in oral squamous cell carcinoma: impact on clinical management. <i>Cellular Oncology (Dordrecht)</i> , 2014, 37, 29-39.	2.1	46
72	Genetic imbalances detected by multiplex ligation-dependent probe amplification in a cohort of patients with oral squamous cell carcinoma – the first step towards clinical personalized medicine. <i>Tumor Biology</i> , 2014, 35, 4687-95.	0.8	22

#	ARTICLE	IF	CITATIONS
73	Genomic characterization of three urinary bladder cancer cell lines: understanding genomic types of urinary bladder cancer. <i>Tumor Biology</i> , 2014, 35, 4599-4617.	0.8	33
74	Human plasma stability during handling and storage: impact on NMR metabolomics. <i>Analyst, The</i> , 2014, 139, 1168-1177.	1.7	139
75	Neurodevelopmental disorders associated with dosage imbalance of <i>ZBTB20</i> correlate with the morbidity spectrum of ZBTB20 candidate target genes. <i>Journal of Medical Genetics</i> , 2014, 51, 605-613.	1.5	26
76	Maternal plasma phospholipids are altered in trisomy 21 cases and prior to preeclampsia and preterm outcomes. <i>Rapid Communications in Mass Spectrometry</i> , 2014, 28, 1635-1638.	0.7	14
77	Potential Markers of Cisplatin Treatment Response Unveiled by NMR Metabolomics of Human Lung Cells. <i>Molecular Pharmaceutics</i> , 2013, 10, 4242-4251.	2.3	39
78	Mosaicism for FMR1 gene full mutation and intermediate allele in a female foetus: A postzygotic retraction event. <i>Gene</i> , 2013, 527, 421-425.	1.0	11
79	Following Healthy Pregnancy by Nuclear Magnetic Resonance (NMR) Metabolic Profiling of Human Urine. <i>Journal of Proteome Research</i> , 2013, 12, 969-979.	1.8	50
80	Insertional translocation leading to a 4q13 duplication including the <i>EPHA5</i> gene in two siblings with attention deficit hyperactivity disorder. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1923-1928.	0.7	14
81	Mid-infrared (MIR) metabolic fingerprinting of amniotic fluid: A possible avenue for early diagnosis of prenatal disorders?. <i>Analytica Chimica Acta</i> , 2013, 764, 24-31.	2.6	26
82	Cryptic 7q36.2q36.3 deletion causes multiple congenital eye anomalies and craniofacial dysmorphism. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 589-593.	0.7	9
83	Second Trimester Maternal Urine for the Diagnosis of Trisomy 21 and Prediction of Poor Pregnancy Outcomes. <i>Journal of Proteome Research</i> , 2013, 12, 2946-2957.	1.8	68
84	Inv21p12q22del21q22 and intellectual disability. <i>Gene</i> , 2013, 517, 120-124.	1.0	0
85	Â thalassemia major due to acquired uniparental disomy in a previously healthy adolescent. <i>Haematologica</i> , 2013, 98, e4-e6.	1.7	8
86	High resolution melting: improvements in the genetic diagnosis of hypertrophic cardiomyopathy in a Portuguese cohort. <i>BMC Medical Genetics</i> , 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. <i>Molecular Cytogenetics</i> , 2012, 5, 25.	0.4	8
88	Can Biofluids Metabolic Profiling Help to Improve Healthcare during Pregnancy?. <i>Spectroscopy</i> , 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. <i>Molecular BioSystems</i> , 2012, 8, 1243.	2.9	94
90	Metabolic Signatures of Lung Cancer in Biofluids: NMR-Based Metabonomics of Urine. <i>Journal of Proteome Research</i> , 2011, 10, 221-230.	1.8	205

#	ARTICLE	IF	CITATIONS
91	Metabolic Signatures of Lung Cancer in Biofluids: NMR-Based Metabonomics of Blood Plasma. <i>Journal of Proteome Research</i> , 2011, 10, 4314-4324.	1.8	154
92	Metabolic Biomarkers of Prenatal Disorders: An Exploratory NMR Metabonomics Study of Second Trimester Maternal Urine and Blood Plasma. <i>Journal of Proteome Research</i> , 2011, 10, 3732-3742.	1.8	144
93	Chromosome 5 derived small supernumerary marker: towards a genotype/phenotype correlation of proximal chromosome 5 imbalances. <i>Journal of Applied Genetics</i> , 2011, 52, 193-200.	1.0	13
94	NMR metabonomic study of lung cancer: metabolic profiling of tissues. <i>BMC Proceedings</i> , 2010, 4, .	1.8	0
95	Can nuclear magnetic resonance (NMR) spectroscopy reveal different metabolic signatures for lung tumours?. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2010, 457, 715-725.	1.4	34
96	X-chromosome terminal deletion in a female with premature ovarian failure: Haploinsufficiency of X-linked genes as a possible explanation. <i>Molecular Cytogenetics</i> , 2010, 3, 14.	0.4	24
97	Late-onset hyperpigmentation: a case with multi-systemic involvement and recombinant X chromosome. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2010, 24, 84-85.	1.3	1
98	Metabolic Profiling of Human Lung Cancer Tissue by 1H High Resolution Magic Angle Spinning (HRMAS) NMR Spectroscopy. <i>Journal of Proteome Research</i> , 2010, 9, 319-332.	1.8	136
99	Impact of Prenatal Disorders on the Metabolic Profile of Second Trimester Amniotic Fluid: A Nuclear Magnetic Resonance Metabonomic Study. <i>Journal of Proteome Research</i> , 2010, 9, 6016-6024.	1.8	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. <i>Cytogenetic and Genome Research</i> , 2009, 125, 109-114.	0.6	5
101	Human bronchial epithelial cells malignantly transformed by hexavalent chromium exhibit an aneuploid phenotype but no microsatellite instability. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2009, 670, 42-52.	0.4	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. <i>Molecular Cytogenetics</i> , 2009, 2, 16.	0.4	6
103	¹ H NMR Based Metabonomics of Human Amniotic Fluid for the Metabolic Characterization of Fetus Malformations. <i>Journal of Proteome Research</i> , 2009, 8, 4144-4150.	1.8	62
104	Analytical Approaches toward Successful Human Cell Metabolome Studies by NMR Spectroscopy. <i>Analytical Chemistry</i> , 2009, 81, 5023-5032.	3.2	61
105	Tetraamelia and lung hypo/aplasia syndrome: New case report and review. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2799-2803.	0.7	18
106	Mowat-Wilson syndrome: an underdiagnosed syndrome?. <i>Clinical Genetics</i> , 2008, 73, 579-584.	1.0	15
107	Metabolite Profiling of Human Amniotic Fluid by Hyphenated Nuclear Magnetic Resonance Spectroscopy. <i>Analytical Chemistry</i> , 2008, 80, 6085-6092.	3.2	46
108	First prenatally detected small supernumerary neocentromeric derivative chromosome 13 resulting in a non-mosaic partial tetrasomy 13q. <i>Cytogenetic and Genome Research</i> , 2008, 121, 293-297.	0.6	8

#	ARTICLE	IF	CITATIONS
109	Three Unusual but Cytogenetically Similar Cases With up to Five Different Cell Lines Involving Structural and Numerical Abnormalities of Chromosome 18. <i>Journal of Histochemistry and Cytochemistry</i> , 2007, 55, 1123-1128.	1.3	13
110	Null mutations and lethal congenital form of glycogen storage disease type IV. <i>Biochemical and Biophysical Research Communications</i> , 2007, 361, 445-450.	1.0	29
111	Potential of NMR Spectroscopy for the Study of Human Amniotic Fluid. <i>Analytical Chemistry</i> , 2007, 79, 8367-8375.	3.2	35
112	Two new cases of de novo small supernumerary marker chromosomes (sSMC) detected at prenatal diagnosis. <i>Prenatal Diagnosis</i> , 2007, 27, 380-381.	1.1	7
113	Cytogenetically invisible microdeletions involving <i>PITX2</i> in Rieger syndrome. <i>Clinical Genetics</i> , 2007, 72, 464-470.	1.0	15
114	Mitochondrial DNA Variants in a Portuguese Population of Patients with Alzheimer's Disease. <i>European Neurology</i> , 2005, 53, 121-124.	0.6	9
115	Partial tetrasomy of chromosome 3q and mosaicism in a child with autism. <i>Journal of Autism and Developmental Disorders</i> , 2003, 33, 177-185.	1.7	12
116	Novel mutations and polymorphisms in the Fanconi anemia group C gene. <i>Human Mutation</i> , 1996, 8, 140-148.	1.1	42
117	Presence of Y chromosome sequences and their effect on the phenotype of six patients with Y chromosome anomalies. <i>American Journal of Medical Genetics Part A</i> , 1995, 55, 269-275.	2.4	7