Sofia DÃ³ria

List of Publications by Year
in descending order

34 papers	597 citations	759233 12 h-index	610901 24 g-index
36 all docs	36 docs citations	36 times ranked	1076 citing authors

#	Article	IF	Citations
1	Clinical Findings on Chromosome 1 Copy Number Variations. Neuropediatrics, 2022, 53, 265-273.	0.6	1
2	Deregulation of imprinted genes expression and epigenetic regulators in placental tissue from intrauterine growth restriction. Journal of Assisted Reproduction and Genetics, 2021, 38, 791-801.	2.5	11
3	X-chromosome inactivation: implications in human disease. Journal of Genetics, 2021, 100, 1.	0.7	16
4	Clinical outcomes of 77 TESE treatment cycles in non-mosaic Klinefelter syndrome patients. Jornal Brasileiro De Reproducao Assistida, 2021, , .	0.7	3
5	X-chromosome inactivation: implications in human disease. Journal of Genetics, 2021, 100, .	0.7	0
6	12q14 microduplication: a new clinical entity reciprocal to the microdeletion syndrome?. BMC Medical Genomics, 2020, 13, 2.	1.5	6
7	Premature ovarian insufficiency: clinical orientations for genetic testing and genetic counseling. Porto Biomedical Journal, 2020, 5, e62.	1.0	20
8	46,XX male disorder of sexual development. Clinical Pediatric Endocrinology, 2020, 29, 43-45.	0.8	8
9	Identification of Copy Number Variation by Array-CGH in Portuguese Children and Adolescents Diagnosed with Autism Spectrum Disorders. Neuropediatrics, 2019, 50, 367-377.	0.6	6
10	Altered expression of epigenetic regulators and imprinted genes in human placenta and fetal tissues from second trimester spontaneous pregnancy losses. Epigenetics, 2019, 14, 1234-1244.	2.7	19
11	Reproductive success of assisted reproductive technology in couples with chromosomal abnormalities. Journal of Assisted Reproduction and Genetics, 2019, 36, 1471-1479.	2.5	9
12	Prenatal diagnosis: the clinical usefulness of array comparative genomic hybridization. Porto Biomedical Journal, 2018, 3, e13.	1.0	2
13	Neonatal dilated cardiomyopathy. Revista Portuguesa De Cardiologia, 2017, 36, 201-214.	0.5	18
14	Intellectual disability and overgrowthâ€"A new case of 19p13.13 microdeletion syndrome with digital abnormalities. American Journal of Medical Genetics, Part A, 2015, 167, 2839-2843.	1.2	7
15	Chromosome mis-segregation and cytokinesis failure in trisomic human cells. ELife, 2015, 4, .	6.0	87
16	Estudo da PrevalÃªncia de Anomalias CromossÃ³micas em Abortamentos EspontÃ¢neos ou Mortes Fetais. Acta Medica Portuguesa, 2014, 27, 42-48.	0.4	3
17	Treatment by testicular sperm extraction and intracytoplasmic sperm injection of 65 azoospermic patients with nonâ€mosaic Klinefelter syndrome with birth of 17 healthy children. Andrology, 2014, 2, 623-631.	3.5	68
18	Relevance of genomic imprinting in intrauterine human growth expression of CDKN1C, H19, IGF2, KCNQ1 and PHLDA2 imprinted genes. Journal of Assisted Reproduction and Genetics, 2014, 31, 1361-1368.	2.5	44

#	Article	IF	Citations
19	Inv21p12q22del21q22 and intellectual disability. Gene, 2013, 517, 120-124.	2.2	0
20	Lateâ€onset Lennoxâ€Gastaut syndrome as a phenotype of 15q11.1q13.3 duplication. Epileptic Disorders, 2012, 14, 159-162.	1.3	11
21	Normal sperm in a 2;2 homologous male translocation carrier. Journal of Assisted Reproduction and Genetics, 2012, 29, 665-668.	2.5	4
22	Aneuploidies Detection in Miscarriages and Fetal Deaths Using Multiplex Ligation-Dependent Probe Amplification: An Alternative for Speeding up Results?. Obstetrical and Gynecological Survey, 2011, 66, 139-141.	0.4	0
23	Application of touch FISH in the study of mosaic tetraploidy and maternal cell contamination in pregnancy losses. Journal of Assisted Reproduction and Genetics, 2010, 27, 657-662.	2.5	11
24	Aneuploidies detection in miscarriages and fetal deaths using multiplex ligation-dependent probe amplification: an alternative for speeding up results?. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2010, 153, 151-155.	1.1	25
25	Gene expression pattern ofIGF2,PHLDA2,PEG10andCDKN1Cimprinted genes in spontaneous miscarriages or fetal deaths. Epigenetics, 2010, 5, 444-450.	2.7	51
26	Phenotypic Expression in the First Case of Complete Trisomy 12: Combination of Prenatal Ultrasound and Necropsic Examination. Fetal Diagnosis and Therapy, 2009, 25, 234-238.	1.4	3
27	An efficient protocol for the detection of chromosomal abnormalities in spontaneous miscarriages or foetal deaths. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2009, 147, 144-150.	1.1	33
28	Clinicopathological features of 45,X/46,Xidic(Y) mosaicism and therapeutic implications: case report. Sao Paulo Medical Journal, 2008, 126, 297-299.	0.9	6
29	New findings in partial trisomy 16q: clinical report. Acta Paediatrica, International Journal of Paediatrics, 2004, 93, 852-854.	1.5	8
30	New findings in partial trisomy 16q: clinical report. Acta Paediatrica, International Journal of Paediatrics, 2004, 93, 852-854.	1.5	0
31	Frequency of NUP98-NSD1 fusion transcript in childhood acute myeloid leukaemia. Leukemia, 2003, 17, 2244-2247.	7.2	61
32	Chronic Eosinophilic Leukaemia Presenting with Erythroderma, Mild Eosinophilia and Hyper-IgE: Clinical, Immunological and Cytogenetic Features and Therapeutic Approach. Acta Haematologica, 2002, 107, 108-112.	1.4	27
33	Expression and function of sodium transporters in two opossum kidney cell clonal sublines. American Journal of Physiology - Renal Physiology, 2002, 283, F73-F85.	2.7	12
34	Detection of prognostic significant translocations in childhood acute lymphoblastic leukaemia by one-step multiplex reverse transcription polymerase chain reaction. British Journal of Haematology, 2000, 109, 638-640.	2.5	13