Sofia DÃ³ria

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2024506/publications.pdf

Version: 2024-02-01

33 papers	597 citations	758635 12 h-index	610482 24 g-index
36 all docs	36 docs citations	36 times ranked	1076 citing authors

#	Article	IF	Citations
1	Chromosome mis-segregation and cytokinesis failure in trisomic human cells. ELife, 2015, 4, .	2.8	87
2	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.	1.9	68
3	Frequency of NUP98-NSD1 fusion transcript in childhood acute myeloid leukaemia. Leukemia, 2003, 17, 2244-2247.	3.3	61
4	Gene expression pattern ofIGF2,PHLDA2,PEG10andCDKN1Cimprinted genes in spontaneous miscarriages or fetal deaths. Epigenetics, 2010, 5, 444-450.	1.3	51
5	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.	1.2	44
6	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.	0.5	33
7	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.	0.7	27
8	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.	0.5	25
9	Premature ovarian insufficiency: clinical orientations for genetic testing and genetic counseling. Porto Biomedical Journal, 2020, 5, e62.	0.4	20
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.	1.3	19
11	Neonatal dilated cardiomyopathy. Revista Portuguesa De Cardiologia, 2017, 36, 201-214.	0.2	18
12	X-chromosome inactivation: implications in human disease. Journal of Genetics, 2021, 100, 1.	0.4	16
13	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.	1.2	13
14	Expression and function of sodium transporters in two opossum kidney cell clonal sublines. American Journal of Physiology - Renal Physiology, 2002, 283, F73-F85.	1.3	12
15	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.	1.2	11
16	Lateâ€onset Lennoxâ€Gastaut syndrome as a phenotype of 15q11.1q13.3 duplication. Epileptic Disorders, 2012, 14, 159-162.	0.7	11
17	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.	1.2	11
18	Reproductive success of assisted reproductive technology in couples with chromosomal abnormalities. Journal of Assisted Reproduction and Genetics, 2019, 36, 1471-1479.	1.2	9

#	Article	IF	Citations
19	New findings in partial trisomy 16q: clinical report. Acta Paediatrica, International Journal of Paediatrics, 2004, 93, 852-854.	0.7	8
20	46,XX male disorder of sexual development. Clinical Pediatric Endocrinology, 2020, 29, 43-45.	0.4	8
21	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.	0.7	7
22	Clinicopathological features of 45,X/46,Xidic(Y) mosaicism and therapeutic implications: case report. Sao Paulo Medical Journal, 2008, 126, 297-299.	0.4	6
23	Identification of Copy Number Variation by Array-CGH in Portuguese Children and Adolescents Diagnosed with Autism Spectrum Disorders. Neuropediatrics, 2019, 50, 367-377.	0.3	6
24	12q14 microduplication: a new clinical entity reciprocal to the microdeletion syndrome?. BMC Medical Genomics, 2020, 13, 2.	0.7	6
25	Normal sperm in a 2;2 homologous male translocation carrier. Journal of Assisted Reproduction and Genetics, 2012, 29, 665-668.	1.2	4
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.	0.6	3
27	Estudo da PrevalÃªncia de Anomalias CromossÃ³micas em Abortamentos EspontÃ¢neos ou Mortes Fetais. Acta Medica Portuguesa, 2014, 27, 42-48.	0.2	3
28	Clinical outcomes of 77 TESE treatment cycles in non-mosaic Klinefelter syndrome patients. Jornal Brasileiro De Reproducao Assistida, 2021, , .	0.3	3
29	Prenatal diagnosis: the clinical usefulness of array comparative genomic hybridization. Porto Biomedical Journal, 2018, 3, e13.	0.4	2
30	Clinical Findings on Chromosome 1 Copy Number Variations. Neuropediatrics, 2022, 53, 265-273.	0.3	1
31	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.	0.2	0
32	Inv21p12q22del21q22 and intellectual disability. Gene, 2013, 517, 120-124.	1.0	0
33	X-chromosome inactivation: implications in human disease. Journal of Genetics, 2021, 100, .	0.4	0