

# Sofia DÃ³ria

## List of Publications by Year in descending order

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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	Clinical Findings on Chromosome 1 Copy Number Variations. <i>Neuropediatrics</i> , 2022, 53, 265-273.	0.3	1
2	Deregulation of imprinted genes expression and epigenetic regulators in placental tissue from intrauterine growth restriction. <i>Journal of Assisted Reproduction and Genetics</i> , 2021, 38, 791-801.	1.2	11
3	X-chromosome inactivation: implications in human disease. <i>Journal of Genetics</i> , 2021, 100, 1.	0.4	16
4	Clinical outcomes of 77 TESE treatment cycles in non-mosaic Klinefelter syndrome patients. <i>Jornal Brasileiro De Reproducao Assistida</i> , 2021, , .	0.3	3
5	X-chromosome inactivation: implications in human disease. <i>Journal of Genetics</i> , 2021, 100, .	0.4	0
6	12q14 microduplication: a new clinical entity reciprocal to the microdeletion syndrome?. <i>BMC Medical Genomics</i> , 2020, 13, 2.	0.7	6
7	Premature ovarian insufficiency: clinical orientations for genetic testing and genetic counseling. <i>Porto Biomedical Journal</i> , 2020, 5, e62.	0.4	20
8	46,XX male disorder of sexual development. <i>Clinical Pediatric Endocrinology</i> , 2020, 29, 43-45.	0.4	8
9	Identification of Copy Number Variation by Array-CGH in Portuguese Children and Adolescents Diagnosed with Autism Spectrum Disorders. <i>Neuropediatrics</i> , 2019, 50, 367-377.	0.3	6
10	Altered expression of epigenetic regulators and imprinted genes in human placenta and fetal tissues from second trimester spontaneous pregnancy losses. <i>Epigenetics</i> , 2019, 14, 1234-1244.	1.3	19
11	Reproductive success of assisted reproductive technology in couples with chromosomal abnormalities. <i>Journal of Assisted Reproduction and Genetics</i> , 2019, 36, 1471-1479.	1.2	9
12	Prenatal diagnosis: the clinical usefulness of array comparative genomic hybridization. <i>Porto Biomedical Journal</i> , 2018, 3, e13.	0.4	2
13	Neonatal dilated cardiomyopathy. <i>Revista Portuguesa De Cardiologia</i> , 2017, 36, 201-214.	0.2	18
14	Intellectual disability and overgrowthâ€”A new case of 19p13.13 microdeletion syndrome with digital abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2839-2843.	0.7	7
15	Chromosome mis-segregation and cytokinesis failure in trisomic human cells. <i>ELife</i> , 2015, 4, .	2.8	87
16	Estudo da PrevalÃªncia de Anomalias CromossÃ3micas em Abortamentos EspontÃ¢neos ou Mortes Fetais. <i>Acta Medica Portuguesa</i> , 2014, 27, 42-48.	0.2	3
17	Treatment by testicular sperm extraction and intracytoplasmic sperm injection of 65 azoospermic patients with nonÃ¢mosaic Klinefelter syndrome with birth of 17 healthy children. <i>Andrology</i> , 2014, 2, 623-631.	1.9	68
18	Relevance of genomic imprinting in intrauterine human growth expression of CDKN1C, H19, IGF2, KCNQ1 and PHLDA2 imprinted genes. <i>Journal of Assisted Reproduction and Genetics</i> , 2014, 31, 1361-1368.	1.2	44

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19	Inv21p12q22del21q22 and intellectual disability. <i>Gene</i> , 2013, 517, 120-124.	1.0	0
20	Late-onset Lennox-Gastaut syndrome as a phenotype of 15q11.1q13.3 duplication. <i>Epileptic Disorders</i> , 2012, 14, 159-162.	0.7	11
21	Normal sperm in a 2;2 homologous male translocation carrier. <i>Journal of Assisted Reproduction and Genetics</i> , 2012, 29, 665-668.	1.2	4
22	Aneuploidies Detection in Miscarriages and Fetal Deaths Using Multiplex Ligation-Dependent Probe Amplification: An Alternative for Speeding up Results?. <i>Obstetrical and Gynecological Survey</i> , 2011, 66, 139-141.	0.2	0
23	Application of touch FISH in the study of mosaic tetraploidy and maternal cell contamination in pregnancy losses. <i>Journal of Assisted Reproduction and Genetics</i> , 2010, 27, 657-662.	1.2	11
24	Aneuploidies detection in miscarriages and fetal deaths using multiplex ligation-dependent probe amplification: an alternative for speeding up results?. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2010, 153, 151-155.	0.5	25
25	Gene expression pattern of <i>IGF2</i> , <i>PHLDA2</i> , <i>PEG10</i> and <i>CDKN1C</i> imprinted genes in spontaneous miscarriages or fetal deaths. <i>Epigenetics</i> , 2010, 5, 444-450.	1.3	51
26	Phenotypic Expression in the First Case of Complete Trisomy 12: Combination of Prenatal Ultrasound and Necropsic Examination. <i>Fetal Diagnosis and Therapy</i> , 2009, 25, 234-238.	0.6	3
27	An efficient protocol for the detection of chromosomal abnormalities in spontaneous miscarriages or foetal deaths. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2009, 147, 144-150.	0.5	33
28	Clinicopathological features of 45,X/46,Xidic(Y) mosaicism and therapeutic implications: case report. <i>Sao Paulo Medical Journal</i> , 2008, 126, 297-299.	0.4	6
29	New findings in partial trisomy 16q: clinical report. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2004, 93, 852-854.	0.7	8
30	Frequency of NUP98-NSD1 fusion transcript in childhood acute myeloid leukaemia. <i>Leukemia</i> , 2003, 17, 2244-2247.	3.3	61
31	Chronic Eosinophilic Leukaemia Presenting with Erythroderma, Mild Eosinophilia and Hyper-IgE: Clinical, Immunological and Cytogenetic Features and Therapeutic Approach. <i>Acta Haematologica</i> , 2002, 107, 108-112.	0.7	27
32	Expression and function of sodium transporters in two opossum kidney cell clonal sublines. <i>American Journal of Physiology - Renal Physiology</i> , 2002, 283, F73-F85.	1.3	12
33	Detection of prognostic significant translocations in childhood acute lymphoblastic leukaemia by one-step multiplex reverse transcription polymerase chain reaction. <i>British Journal of Haematology</i> , 2000, 109, 638-640.	1.2	13