Irene Madrigal

List of Publications by Year
in descending order

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Version: 2024-02-01

51 papers	1,002 citations	567281 15 h-index	454955 30 g-index
54 all docs	54 docs citations	54 times ranked	1590 citing authors

#	Article	IF	Citations
1	Diagnostic yield of next-generation sequencing in 87 families with neurodevelopmental disorders. Orphanet Journal of Rare Diseases, 2022, 17, 60.	2.7	21
2	Lethal Congenital Contracture Syndrome 11: A Case Report and Literature Review. Journal of Clinical Medicine, 2022, 11, 3570.	2.4	0
3	Cell-Free DNA Testing: What Is the Reason Why High-Risk Women Choose It?. Fetal Diagnosis and Therapy, 2021, 48, 9-14.	1.4	0
4	Novel Compound Heterozygous Mutation in TRAPPC9 Gene: The Relevance of Whole Genome Sequencing. Genes, 2021, 12, 557.	2.4	14
5	The Contribution of QF-PCR and Pathology Studies in the Diagnosis of Diandric Triploidy/Partial Mole. Diagnostics, 2021, 11, 1811.	2.6	2
6	Significance of Low Maternal Serum Î’-hCG Levels in the Assessment of the Risk of Atypical Chromosomal Abnormalities. Fetal Diagnosis and Therapy, 2021, 48, 849-856.	1.4	3
7	Chromosome microarray analysis should be offered to all invasive prenatal diagnostic testing following a normal rapid aneuploidy test result. Clinical Genetics, 2020, 98, 379-383.	2.0	5
8	Cytogenetic Investigation in 136 Consecutive Stillbirths: Does the Tissue Type Affect the Success Rate of Chromosomal Microarray Analysis and Karyotype?. Fetal Diagnosis and Therapy, 2020, 47, 315-320.	1.4	3
9	Role of mitochondrial DNA variants in the development of fragile X-associated tremor/ataxia syndrome. Mitochondrion, 2020, 52, 157-162.	3.4	4
10	Fragile Xâ€associated tremor/ataxia syndrome: Regional decrease of mitochondrial DNA copy number relates to clinical manifestations. Genes, Brain and Behavior, 2019, 18, e12565.	2.2	11
11	Genetic linkage analysis of a large family identifies FIGN as a candidate modulator of reduced penetrance in heritable pulmonary arterial hypertension. Journal of Medical Genetics, 2019, 56, 481-490.	3.2	3
12	Spectrum of clinical heterogeneity of Î²-tubulin TUBB5 gene mutations. Gene, 2019, 695, 12-17.	2.2	11
13	Severe neurocognitive and growth disorders due to variation in THOC2 , an essential component of nuclear mRNA export machinery. Human Mutation, 2018, 39, 1126-1138.	2.5	28
14	New insights into the regulatory function of CYFIP1 in the context of WAVE- and FMRP-containing complexes. DMM Disease Models and Mechanisms, 2017, 10, 463-474.	2.4	49
15	Paternal transmission of a FMR1 full mutation allele. American Journal of Medical Genetics, Part A, 2017, 173, 2795-2797.	1.2	12
16	A 92,XXXY Miscarriage Consecutive to a Digynic Triploid Pregnancy. Cytogenetic and Genome Research, 2016, 149, 258-261.	1.1	1
17	Carriage of One or Two FMR1 Premutation Alleles Seems to Have No Effect on Illness Severity in a FXTAS Female with an Autozygous FMR1 Premutation Allele. Cerebellum, 2016, 15, 570-577.	2.5	2
18	A novel splicing mutation in the IQSEC2 gene that modulates the phenotype severity in a family with intellectual disability. European Journal of Human Genetics, 2016, 24, 1117-1123.	2.8	9

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19	Comprehensive molecular testing in patients with high functioning autism spectrum disorder. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2016, 784-785, 46-52.	1.0	24
20	Skewed X Inactivation in Women Carrying the <i>FMR1</i> Premutation and Its Relation with Fragile-X-Associated Tremor/Ataxia Syndrome. Neurodegenerative Diseases, 2016, 16, 290-292.	1.4	14
21	Blood expression profiles of fragile X premutation carriers identify candidate genes involved in neurodegenerative and infertility phenotypes. Neurobiology of Disease, 2014, 65, 43-54.	4.4	23
22	Efficient application of next-generation sequencing for the diagnosis of rare genetic syndromes. Journal of Clinical Pathology, 2014, 67, 1099-1103.	2.0	13
23	Prevalence Studies on Fragile X Alleles in Autism. , 2014, , 2755-2771.		1
24	A parallel study of different array-CGH platforms in a set of Spanish patients with developmental delay and intellectual disability. Gene, 2013, 521, 82-86.	2.2	15
25	Screening for the presence of FMR1 premutation alleles in women with fibromyalgia. Gene, 2013, 512, 305-308.	2.2	13
26	High apolipoprotein E4 allele frequency in FXTAS patients. Genetics in Medicine, 2013, 15, 639-642.	2.4	13
27	A170P mutation in‹i›SHOX‹/i›gene in a patient not presenting with Madelung deformity: Figure 1. Journal of Clinical Pathology, 2012, 65, 844-846.	2.0	3
28	15q11.2 microdeletion and FMR1 premutation in a family with intellectual disabilities and autism. Gene, 2012, 508, 92-95.	2.2	21
29	12p13 rearrangements: 6â€‰Mb deletion responsible for ID/MCA and reciprocal duplication without clinical responsibility. American Journal of Medical Genetics, Part A, 2012, 158A, 1071-1076.	1.2	10
30	Reply to He et al. European Journal of Human Genetics, 2011, 19, 124-124.	2.8	2
31	Intermediate FMR1 alleles and cognitive and/or behavioural phenotypes. European Journal of Human Genetics, 2011, 19, 921-923.	2.8	17
32	Novel MLH1 duplication identified in Colombian families with Lynch syndrome. Genetics in Medicine, 2011, 13, 155-160.	2.4	14
33	Subtelomeric MLPA: is it really useful in prenatal diagnosis?. Prenatal Diagnosis, 2010, 30, 1165-1169.	2.3	6
34	X-inactivation of HSD17B10 revealed by cDNA analysis in two female patients with 17Î²-hydroxysteroid dehydrogenase 10 deficiency. European Journal of Human Genetics, 2010, 18, 1353-1355.	2.8	12
35	Xq26.2-q26.3 microduplication in two brothers with intellectual disabilities: clinical and molecular characterization. Journal of Human Genetics, 2010, 55, 822-826.	2.3	11
36	Study of the BMPR2 Gene in Patients with Pulmonary Arterial Hypertension. Archivos De Bronconeumologia, 2010, 46, 129-134.	0.8	2

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37	Identification of human specific gene duplications relative to other primates by array CGH and quantitative PCR. Genomics, 2010, 95, 203-209.	2.9	7
38	Protocol proposal for Friedreich ataxia molecular diagnosis using fluorescent and triplet repeat primed polymerase chain reaction. Translational Research, 2010, 156, 309-314.	5.0	8
39	Prenatal diagnosis of two different unbalanced forms of an inherited (Y;12) translocation. American Journal of Medical Genetics, Part A, 2009, 149A, 2820-2823.	1.2	7
40	Molecular analysis of the APC and MUTYH genes in Galician and Catalonian FAP families: a different spectrum of mutations?. BMC Medical Genetics, 2009, 10, 57.	2.1	48
41	Penetrance of FMR1 premutation associated pathologies in fragile X syndrome families. European Journal of Human Genetics, 2009, 17, 1359-1362.	2.8	254
42	Dosage-Dependent Severity of the Phenotype in Patients with Mental Retardation Due to a Recurrent Copy-Number Gain at Xq28 Mediated by an Unusual Recombination. American Journal of Human Genetics, 2009, 85, 809-822.	6.2	70
43	Premature ovarian failure and fragile X female premutation carriers. Menopause, 2009, 16, 944-949.	2.0	27
44	Atypical XX male with theSRY gene located at the long arm of chromosome 1 and a 1qter microdeletion. American Journal of Medical Genetics, Part A, 2008, 146A, 1335-1340.	1.2	27
45	Screening for FXTAS in 95 Spanish Patients Negative for Huntington Disease. Genetic Testing and Molecular Biomarkers, 2008, 12, 135-138.	1.7	13
46	Evidence of depressive symptoms in fragile-X syndrome premutated females. Psychiatric Genetics, 2008, 18, 153-155.	1.1	61
47	Study of the genotypeâ€“phenotype relationship in four cases of congenital erythropoietic porphyria. Blood Cells, Molecules, and Diseases, 2007, 38, 242-246.	1.4	15
48	MLPA as first screening method for the detection of microduplications and microdeletions in patients with X-linked mental retardation. Genetics in Medicine, 2007, 9, 117-122.	2.4	34
49	Trisomy of 19.4 Mb region of chromosome 22 and subtelomeric 17p identified in a male without clinical affectation. American Journal of Medical Genetics, Part A, 2007, 143A, 2423-2429.	1.2	2
50	Molecular characterization of a t(9;12)(p21;q13) balanced chromosome translocation in combination with integrative genomics analysis identifiesC9orf14as a candidate tumor-suppressor. Genes Chromosomes and Cancer, 2007, 46, 155-162.	2.8	10
51	Fluorescence in situ hybridization studies using BAC clones of the EVI1 locus in hematological malignancies with 3q rearrangements. Cancer Genetics and Cytogenetics, 2006, 170, 115-120.	1.0	12