Irene Madrigal

List of Publications by Year
in descending order

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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	Penetrance of FMR1 premutation associated pathologies in fragile X syndrome families. European Journal of Human Genetics, 2009, 17, 1359-1362.	2.8	254
2	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
3	Evidence of depressive symptoms in fragile-X syndrome premutated females. Psychiatric Genetics, 2008, 18, 153-155.	1.1	61
4	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
5	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
6	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
7	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
8	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
9	Premature ovarian failure and fragile X female premutation carriers. Menopause, 2009, 16, 944-949.	2.0	27
10	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
11	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
12	15q11.2 microdeletion and FMR1 premutation in a family with intellectual disabilities and autism. Gene, 2012, 508, 92-95.	2.2	21
13	Diagnostic yield of next-generation sequencing in 87 families with neurodevelopmental disorders. Orphanet Journal of Rare Diseases, 2022, 17, 60.	2.7	21
14	Intermediate FMR1 alleles and cognitive and/or behavioural phenotypes. European Journal of Human Genetics, 2011, 19, 921-923.	2.8	17
15	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
16	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
17	Novel MLH1 duplication identified in Colombian families with Lynch syndrome. Genetics in Medicine, 2011, 13, 155-160.	2.4	14
18	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

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19	Novel Compound Heterozygous Mutation in TRAPPC9 Gene: The Relevance of Whole Genome Sequencing. Genes, 2021, 12, 557.	2.4	14
20	Screening for FXTAS in 95 Spanish Patients Negative for Huntington Disease. Genetic Testing and Molecular Biomarkers, 2008, 12, 135-138.	1.7	13
21	Screening for the presence of FMR1 premutation alleles in women with fibromyalgia. Gene, 2013, 512, 305-308.	2.2	13
22	High apolipoprotein E4 allele frequency in FXTAS patients. Genetics in Medicine, 2013, 15, 639-642.	2.4	13
23	Efficient application of next-generation sequencing for the diagnosis of rare genetic syndromes. Journal of Clinical Pathology, 2014, 67, 1099-1103.	2.0	13
24	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
25	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
26	Paternal transmission of a FMR1 full mutation allele. American Journal of Medical Genetics, Part A, 2017, 173, 2795-2797.	1.2	12
27	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
28	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
29	Spectrum of clinical heterogeneity of Î²-tubulin TUBB5 gene mutations. Gene, 2019, 695, 12-17.	2.2	11
30	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
31	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
32	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
33	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
34	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
35	Identification of human specific gene duplications relative to other primates by array CGH and quantitative PCR. Genomics, 2010, 95, 203-209.	2.9	7
36	Subtelomeric MLPA: is it really useful in prenatal diagnosis?. Prenatal Diagnosis, 2010, 30, 1165-1169.	2.3	6

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37	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
38	Role of mitochondrial DNA variants in the development of fragile X-associated tremor/ataxia syndrome. Mitochondrion, 2020, 52, 157-162.	3.4	4
39	A170P mutation inSHOXgene in a patient not presenting with Madelung deformity: Figure 1. Journal of Clinical Pathology, 2012, 65, 844-846.	2.0	3
40	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
41	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
42	Significance of Low Maternal Serum Î²-hCG Levels in the Assessment of the Risk of Atypical Chromosomal Abnormalities. Fetal Diagnosis and Therapy, 2021, 48, 849-856.	1.4	3
43	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
44	Study of the BMPR2 Gene in Patients with Pulmonary Arterial Hypertension. Archivos De Bronconeumologia, 2010, 46, 129-134.	0.8	2
45	Reply to He et al. European Journal of Human Genetics, 2011, 19, 124-124.	2.8	2
46	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
47	The Contribution of QF-PCR and Pathology Studies in the Diagnosis of Diandric Triploidy/Partial Mole. Diagnostics, 2021, 11, 1811.	2.6	2
48	A 92,XXXY Miscarriage Consecutive to a Digynic Triploid Pregnancy. Cytogenetic and Genome Research, 2016, 149, 258-261.	1.1	1
49	Prevalence Studies on Fragile X Alleles in Autism. , 2014, , 2755-2771.		1
50	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
51	Lethal Congenital Contracture Syndrome 11: A Case Report and Literature Review. Journal of Clinical Medicine, 2022, 11, 3570.	2.4	0