Mariluce Riegel

List of Publications by Year
in descending order

79 papers	1,785 citations	361413 20 h-index	302126 39 g-index
84 all docs	84 docs citations	84 times ranked	2588 citing authors

#	Article	IF	Citations
1	Cryptic deletions are a common finding in "balanced" reciprocal and complex chromosome rearrangements: a study of 59 patients. Journal of Medical Genetics, 2007, 44, 750-762.	3.2	244
2	Multipotent mesenchymal stem cells from human placenta: critical parameters for isolation and maintenance of stemness after isolation. American Journal of Obstetrics and Gynecology, 2010, 202, 193.e1-193.e13.	1.3	96
3	Survival with trisomy 18â€"data from Switzerland. American Journal of Medical Genetics, Part A, 2006, 140A, 952-959.	1.2	88
4	Submicroscopic terminal deletions and duplications in retarded patients with unclassified malformation syndromes. Human Genetics, 2001, 109, 286-294.	3.8	86
5	Duplications in addition to terminal deletions are present in a proportion of ring chromosomes: clues to the mechanisms of formation. Journal of Medical Genetics, 2007, 45, 147-154.	3.2	85
6	New microdeletion and microduplication syndromes: a comprehensive review. Genetics and Molecular Biology, 2014, 37, 210-219.	1.3	84
7	European Cytogeneticists Association Register ofÂUnbalanced Chromosome Aberrations (ECARUCA); anÂonline database forÂrare chromosome abnormalities. European Journal of Medical Genetics, 2006, 49, 279-291.	1.3	74
8	Terminal deletion, del(1)(p36.3), detected through screening for terminal deletions in patients with unclassified malformation syndromes. American Journal of Medical Genetics Part A, 1999, 82, 249-253.	2.4	68
9	Human molecular cytogenetics: from cells to nucleotides. Genetics and Molecular Biology, 2014, 37, 194-209.	1.3	55
10	Monochorionicâ€diamniotic twins discordant in gender from a naturally conceived pregnancy through postzygotic sex chromosome loss in a 47,XXY zygote. Prenatal Diagnosis, 2008, 28, 759-763.	2.3	43
11	Genetic causes of intellectual disability in a birth cohort: A populationâ€based study. American Journal of Medical Genetics, Part A, 2015, 167, 1204-1214.	1.2	39
12	The Fate of Children with Microdeletion 22q11.2 Syndrome and Congenital Heart Defect: Clinical Course and Cardiac Outcome. Pediatric Cardiology, 2008, 29, 76-83.	1.3	38
13	Relative frequency and estimated minimal frequency of Lysosomal Storage Diseases in Brazil: Report from a Reference Laboratory. Genetics and Molecular Biology, 2017, 40, 31-39.	1.3	35
14	Neurological manifestations of lysosomal disorders and emerging therapies targeting the CNS. The Lancet Child and Adolescent Health, 2018, 2, 56-68.	5.6	32
15	Diagnostic and treatment strategies in mucopolysaccharidosis VI. The Application of Clinical Genetics, 2015, 8, 245.	3.0	31
16	An unexpected finding: younger fathers have a higher risk for offspring with chromosomal aneuploidies. European Journal of Human Genetics, 2015, 23, 466-472.	2.8	27
17	Unbalanced 18q/21q translocation in a patient previously reported as monosomy 21. European Journal of Medical Genetics, 2005, 48, 167-174.	1.3	26
18	Intellectual Disability in a Birth Cohort: Prevalence, Etiology, and Determinants at the Age of 4 Years. Public Health Genomics, 2016, 19, 290-297.	1.0	25

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19	Persistent low thymic activity and non-cardiac mortality in children with chromosome 22q11Â·2 microdeletion and partial DiGeorge syndrome. Clinical and Experimental Immunology, 2009, 155, 189-198.	2.6	22
20	Non-random asynchronous replication at 22q11.2 favours unequal meiotic crossovers leading to the human 22q11.2 deletion. Journal of Medical Genetics, 2004, 41, 413-420.	3.2	21
21	Tetrasomy 12pter-12p13.31 in a girl with partial Pallisterâ€“Killian syndrome phenotype. European Journal of Medical Genetics, 2005, 48, 319-327.	1.3	21
22	Pre- and postnatal findings in trisomy 17 mosaicism. American Journal of Medical Genetics, Part A, 2006, 140A, 1628-1636.	1.2	21
23	Mosaic imprinting defect in a patient with an almost typical expression of the Praderâ€“Willi syndrome. European Journal of Human Genetics, 2005, 13, 273-277.	2.8	20
24	Atypical macrocephaly-cutis marmorata telangiectatica congenita with retinoblastoma. Clinical Dysmorphology, 2002, 11, 199-202.	0.3	19
25	Prenatal diagnosis of mosaicism for a del(22)(q13). Prenatal Diagnosis, 2000, 20, 76-79.	2.3	18
26	Opitz â€œCâ€trigonocephaly-like syndrome in a patient with terminal deletion of 2p and partial duplication of 17q. , 2004, 131A, 310-312.		18
27	Juvenile myoclonic epilepsy with photosensitivity in a female with Velocardiofacial syndrome (del(22)(q11.2))â€”Causal relationship or coincidence?. Seizure: the Journal of the British Epilepsy Association, 2009, 18, 660-663.	2.0	17
28	Optimization of the culturing conditions of human umbilical cord bloodâ€derived endothelial colonyâ€forming cells under xenoâ€free conditions applying a transcriptomic approach. Genes To Cells, 2010, 15, 671-687.	1.2	17
29	Longâ€term followâ€up of four patients with langerâ€“giedion syndrome: Clinical course and complications. American Journal of Medical Genetics, Part A, 2013, 161, 2216-2225.	1.2	17
30	Microarray-based comparative genomic hybridization analysis in neonates with congenital anomalies: detection of chromosomal imbalances. Jornal De Pediatria, 2015, 91, 59-67.	2.0	17
31	Cri-Du-Chat Syndrome: Clinical Profile and Chromosomal Microarray Analysis in Six Patients. BioMed Research International, 2016, 2016, 1-9.	1.9	17
32	Long-term follow-up of a 26-year-old male with duplication of 16p: Clinical report and review. American Journal of Medical Genetics, Part A, 2007, 143A, 399-408.	1.2	16
33	Blepharophimosis and mental retardation (BMR) phenotypes caused by chromosomal rearrangements: Description in a boy with partial trisomy 10q and monosomy 4q and review of the literature. European Journal of Medical Genetics, 2008, 51, 113-123.	1.3	16
34	Severe phenotype in MPS II patients associated with a large deletion including contiguous genes. American Journal of Medical Genetics, Part A, 2012, 158A, 1055-1059.	1.2	16
35	Cytogenomic Evaluation of Subjects with Syndromic and Nonsyndromic Conotruncal Heart Defects. BioMed Research International, 2015, 2015, 1-12.	1.9	16
36	Rare disease landscape in Brazil: report of a successful experience in inborn errors of metabolism. Orphanet Journal of Rare Diseases, 2016, 11, 76.	2.7	16

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37	Integrated analysis of the critical region 5p15.3â€“p15.2 associated with cri-du-chat syndrome. Genetics and Molecular Biology, 2019, 42, 186-196.	1.3	16
38	Trisomy 18: Changes in sex ratio during intrauterine life. American Journal of Medical Genetics, Part A, 2006, 140A, 2365-2367.	1.2	15
39	Inv dup del(4)(:p13â€‰â†’â€‰p16.3::p16.3â€‰â†’â€‰qter) in a girl without typical manifestations of Wolfâ€“Hirschhorn syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 1302-1307.	1.2	14
40	Molecular Screening for 22Q11.2 Deletion Syndrome in Patients With Congenital Heart Disease. Pediatric Cardiology, 2014, 35, 1356-1362.	1.3	14
41	Current molecular genetics strategies for the diagnosis of lysosomal storage disorders. Expert Review of Molecular Diagnostics, 2016, 16, 113-123.	3.1	13
42	Intrathecal/Intracerebroventricular enzyme replacement therapy for the mucopolysaccharidoses: efficacy, safety, and prospects. Expert Opinion on Orphan Drugs, 2018, 6, 403-411.	0.8	13
43	Maternal uniparental isodisomy 10 and mosaicism for an additional marker chromosome derived from the paternal chromosome 10 in a fetus. Prenatal Diagnosis, 2002, 22, 418-421.	2.3	12
44	Isolated central form of tetrahydrobiopterin deficiency associated with hemizygosity on chromosome 11q and a mutant allele of PTPS. Human Mutation, 2000, 16, 54-60.	2.5	11
45	New case of non-mosaic tetrasomy 9p in a severely polymalformed newborn girl. Birth Defects Research Part A: Clinical and Molecular Teratology, 2003, 67, 985-988.	1.6	11
46	Postzygotic isochromosome formation as a cause for false-negative results from chorionic villus chromosome examinations. Prenatal Diagnosis, 2006, 26, 221-225.	2.3	11
47	Velofacial hypoplasia (Sedlackova syndrome): a variant of velocardiofacial (Shprintzen) syndrome and part of the phenotypical spectrum of del 22q11.2. European Journal of Pediatrics, 2001, 160, 54-57.	2.7	10
48	Patient with rheumatoid arthritis and MCA/MR syndrome due to unbalanced der(18) transmission of a paternal translocation t(18;20)(p11.1;p11.1). American Journal of Medical Genetics Part A, 2002, 108, 226-228.	2.4	10
49	Novel duplication on chromosome 16 (q12.1-q21) associated with behavioral disorder, mild cognitive impairment, speech delay, and dysmorphic features: case report. Croatian Medical Journal, 2011, 52, 415-422.	0.7	10
50	Clinical findings in Brazilian patients with adult GM1 gangliosidosis. JIMD Reports, 2019, 49, 96-106.	1.5	10
51	Longitudinal observation of a patient with Rieger syndrome and interstitial deletion 4 (q25â€“q31.1). American Journal of Medical Genetics, Part A, 2010, 152A, 977-981.	1.2	9
52	Information and Diagnosis Networks â€“ tools to improve diagnosis and treatment for patients with rare genetic diseases. Genetics and Molecular Biology, 2019, 42, 155-164.	1.3	9
53	A further case of a Prader-Willi syndrome phenotype in a patient with Angelman syndrome molecular defect. Arquivos De Neuro-Psiquiatria, 2002, 60, 1011-1014.	0.8	8
54	Population medical genetics: translating science to the community. Genetics and Molecular Biology, 2019, 42, 312-320.	1.3	8

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55	Distal deletion, del(2)(q33.3q33.3), in a patient with severe growth deficiency and minor anomalies. American Journal of Medical Genetics Part A, 2001, 102, 227-230.	2.4	7
56	Duplication of (2)(q11.1-q13.2) in a boy with mental retardation and cleft lip and palate: Another clefting gene locus on proximal 2q?. American Journal of Medical Genetics Part A, 2002, 111, 76-80.	2.4	7
57	Newborn with malformations and a combined duplication of 9pter-q22 and 16q22-qter resulting from unbalanced segregation of a complex maternal translocation. American Journal of Medical Genetics Part A, 2003, 120A, 247-252.	2.4	7
58	Analysis of a Protein Network Related to Copy Number Variations in Autism Spectrum Disorder. Journal of Molecular Neuroscience, 2019, 69, 140-149.	2.3	7
59	Duplication of (12)(pter-q13.3) combined with deletion of (22)(pter-q11.2) in a patient with features of both chromosome aberrations. European Journal of Medical Genetics, 2007, 50, 128-132.	1.3	6
60	Interstitial 14q24.3 to q31.3 deletion in a 6-year-old boy with a non-specific dysmorphic phenotype. Molecular Cytogenetics, 2014, 7, 77.	0.9	6
61	Cytogenomic Integrative Network Analysis of the Critical Region Associated with Wolf-Hirschhorn Syndrome. BioMed Research International, 2018, 2018, 1-10.	1.9	6
62	An unusual reciprocal translocation detected by subtelomeric FISH: Interstitial and not terminal. , 2005, 135A, 86-90.		5
63	Candidate Genes Associated with Delayed Neuropsychomotor Development and Seizures in a Patient with Ring Chromosome 20. Case Reports in Genetics, 2020, 2020, 1-6.	0.2	5
64	Mosaic supernumerary ring chromosome 1 in a three-generational family: 10-year follow-up report. European Journal of Medical Genetics, 2011, 54, 152-156.	1.3	4
65	Candidate Genes Associated With Neurological Findings in a Patient With Trisomy 4p16.3 and Monosomy 5p15.2. Frontiers in Genetics, 2020, 11, 561.	2.3	4
66	Towards New Approaches to Evaluate Dynamic Mosaicism in Ring Chromosome 13 Syndrome. Case Reports in Genetics, 2019, 2019, 1-10.	0.2	3
67	Wolf-Hirschhorn syndrome due to a 3:1 segregation of a maternal balanced t(4;15)(p16.3;q11) translocation. Prenatal Diagnosis, 2000, 20, 847-850.	2.3	2
68	Networkâ€based analysis using chromosomal microdeletion syndromes as a model. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 337-348.	1.6	2
69	A child with cat-eye syndrome and oculo-auriculo-vertebral spectrum phenotype: A discussion around molecular cytogenetic findings. European Journal of Medical Genetics, 2021, 64, 104319.	1.3	2
70	Interstitial deletion of 7q31.32Âât'Âq33 secondary to a paracentric inversion of a maternal chromosome 7. European Journal of Medical Genetics, 2011, 54, 181-185.	1.3	1
71	A patient presenting a 22q13 deletion associated with an apparently balanced translocation t(16;22): an illustrative case in the investigation of patients with low ARSA activity. Genetics and Molecular Biology, 2012, 35, 424-427.	1.3	1
72	Spontaneous Pubertal Onset in a Male Patient With Mixed Gonadal Dysgenesis With Mosaicism 45,X/ 46, X, mar (Y)/ 47,X,mar(Y),+mar(Y) - Pediatric Case Report. Urology, 2021, , .	1.0	1

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73	Molecular cytogenetic evaluation of chromosomal microdeletions: the experience of a public hospital in Southern Brazil. Clinical and Biomedical Research, 2014, 34, 357-365.	0.1	1
74	Short stature, myopia, severe developmental delay, and peculiar facial appearance in two brothers: A new syndrome?. , 1999, 86, 486-491.		0
75	The Contribution of Molecular Techniques in Prenatal Diagnosis and Post mortem Fetus with Multiple Malformation. , 0, , .		0
76	Shared Neurodevelopmental Perturbations Can Lead to Intellectual Disability in Individuals with Distinct Rare Chromosome Duplications. Genes, 2021, 12, 632.	2.4	0
77	AnÃ¡lise comparativa entre as metodologias de PCR metilaÃ§Ã£o-especÃfica (MSP), Southern blot (SB) e FISH utilizadas no diagnÃ³stico genÃ©tico molecular de pacientes com suspeita clÃnica das sÃndromes de Prader-Willi ou Angelman. Clinical and Biomedical Research, 2016, 36, 71-79.	0.1	0
78	Diabetes insipidus como manifestaÃ§Ã£o inicial de leucemia mieloide aguda em paciente com monossomia do cromossomo 7. Clinical and Biomedical Research, 2017, 37, 55-58.	0.1	0
79	Desenvolvimento na sÃndrome Cri-du-chat: Estudo de caso com acompanhamento longitudinal durante 20 anos com relevÃ¢ncia na interaÃ§Ã£o com a famÃlia e tratamento continuado. Brazilian Journal of Health Review, 2019, 2, 4436-4444.	0.1	0