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	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
2	Shared Neurodevelopmental Perturbations Can Lead to Intellectual Disability in Individuals with Distinct Rare Chromosome Duplications. Genes, 2021, 12, 632.	2.4	0
3	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
4	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
5	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
6	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
7	Clinical findings in Brazilian patients with adult GM1 gangliosidosis. JIMD Reports, 2019, 49, 96-106.	1.5	10
8	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
9	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
10	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
11	Population medical genetics: translating science to the community. Genetics and Molecular Biology, 2019, 42, 312-320.	1.3	8
12	Desenvolvimento na sÃ¬ndrome Cri-du-chat: Estudo de caso com acompanhamento longitudinal durante 20 anos com relevÃ¢ncia na interaÃ§Ã£o com a famÃ¼lia e tratamento continuado. Brazilian Journal of Health Review, 2019, 2, 4436-4444.	0.1	0
13	Towards New Approaches to Evaluate Dynamic Mosaicism in Ring Chromosome 13 Syndrome. Case Reports in Genetics, 2019, 2019, 1-10.	0.2	3
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	Cytogenomic Integrative Network Analysis of the Critical Region Associated with Wolf-Hirschhorn Syndrome. BioMed Research International, 2018, 2018, 1-10.	1.9	6
16	Intrathecal/Intracerebroventricular enzyme replacement therapy for the mucopolysaccharidoses: efficacy, safety, and prospects. Expert Opinion on Orphan Drugs, 2018, 6, 403-411.	0.8	13
17	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
18	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

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19	Cri-Du-Chat Syndrome: Clinical Profile and Chromosomal Microarray Analysis in Six Patients. BioMed Research International, 2016, 2016, 1-9.	1.9	17
20	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
21	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
22	Current molecular genetics strategies for the diagnosis of lysosomal storage disorders. Expert Review of Molecular Diagnostics, 2016, 16, 113-123.	3.1	13
23	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
24	Diagnostic and treatment strategies in mucopolysaccharidosis VI. The Application of Clinical Genetics, 2015, 8, 245.	3.0	31
25	Cytogenomic Evaluation of Subjects with Syndromic and Nonsyndromic Conotruncal Heart Defects. BioMed Research International, 2015, 2015, 1-12.	1.9	16
26	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
27	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
28	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
29	New microdeletion and microduplication syndromes: a comprehensive review. Genetics and Molecular Biology, 2014, 37, 210-219.	1.3	84
30	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
31	Molecular Screening for 22Q11.2 Deletion Syndrome in Patients With Congenital Heart Disease. Pediatric Cardiology, 2014, 35, 1356-1362.	1.3	14
32	Human molecular cytogenetics: from cells to nucleotides. Genetics and Molecular Biology, 2014, 37, 194-209.	1.3	55
33	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
34	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
35	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
36	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

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37	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
38	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
39	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
40	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
41	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
42	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
43	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
44	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
45	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
46	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
47	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
48	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
49	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
50	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
51	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
52	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
53	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
54	Postzygotic isochromosome formation as a cause for false-negative results from chorionic villus chromosome examinations. Prenatal Diagnosis, 2006, 26, 221-225.	2.3	11

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55	Survival with trisomy 18â€"data from Switzerland. American Journal of Medical Genetics, Part A, 2006, 140A, 952-959.	1.2	88
56	Pre- and postnatal findings in trisomy 17 mosaicism. American Journal of Medical Genetics, Part A, 2006, 140A, 1628-1636.	1.2	21
57	Trisomy 18: Changes in sex ratio during intrauterine life. American Journal of Medical Genetics, Part A, 2006, 140A, 2365-2367.	1.2	15
58	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
59	An unusual reciprocal translocation detected by subtelomeric FISH: Interstitial and not terminal. , 2005, 135A, 86-90.		5
60	Unbalanced 18q/21q translocation in a patient previously reported as monosomy 21. European Journal of Medical Genetics, 2005, 48, 167-174.	1.3	26
61	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
62	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
63	Opitz â€œCâ€•trigonocephaly-like syndrome in a patient with terminal deletion of 2p and partial duplication of 17q. , 2004, 131A, 310-312.		18
64	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
65	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
66	Atypical macrocephaly-cutis marmorata telangiectatica congenita with retinoblastoma. Clinical Dysmorphology, 2002, 11, 199-202.	0.3	19
67	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
68	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
69	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
70	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
71	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
72	Submicroscopic terminal deletions and duplications in retarded patients with unclassified malformation syndromes. Human Genetics, 2001, 109, 286-294.	3.8	86

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73	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
74	Prenatal diagnosis of mosaicism for a del(22)(q13). Prenatal Diagnosis, 2000, 20, 76-79.	2.3	18
75	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
76	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
77	Short stature, myopia, severe developmental delay, and peculiar facial appearance in two brothers: A new syndrome?. , 1999, 86, 486-491.		0
78	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
79	The Contribution of Molecular Techniques in Prenatal Diagnosis and Post mortem Fetus with Multiple Malformation. , 0, , .		0