Vera LÃºcia Gil-da-Silva-Lopes

List of Publications by Year
in descending order

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80 papers	1,407 citations	516215 16 h-index	395343 33 g-index
88 all docs	88 docs citations	88 times ranked	2529 citing authors

#	Article	IF	Citations
1	Deficiency of UBR1, a ubiquitin ligase of the N-end rule pathway, causes pancreatic dysfunction, malformations and mental retardation (Johanson-Blizzard syndrome). Nature Genetics, 2005, 37, 1345-1350.	9.4	252
2	Recessive and Dominant De Novo ITPR1 Mutations Cause Gillespie Syndrome. American Journal of Human Genetics, 2016, 98, 971-980.	2.6	113
3	22q11.2 deletion syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 879-888.	0.7	103
4	Defining new guidelines for screening the 22q11.2 deletion based on a clinical and dysmorphologic evaluation of 194 individuals and review of the literature. European Journal of Pediatrics, 2013, 172, 927-945.	1.3	53
5	A multicentric association study between 39 genes and nonsyndromic cleft lip and palate in a Brazilian population. Journal of Cranio-Maxillo-Facial Surgery, 2016, 44, 16-20.	0.7	48
6	Neonatal Care of Infants with Cleft Lip and/or Palate: Feeding Orientation and Evolution of Weight Gain in a Nonspecialized Brazilian Hospital. Cleft Palate-Craniofacial Journal, 2007, 44, 329-334.	0.5	43
7	DLX4is associated with orofacial clefting and abnormal jaw development. Human Molecular Genetics, 2015, 24, 4340-4352.	1.4	36
8	Variants in members of the cadherinâ€“catenin complex, CDH1 and CTNND1, cause blepharocheilodontic syndrome. European Journal of Human Genetics, 2018, 26, 210-219.	1.4	34
9	Investigation of genetic factors underlying typical orofacial clefts: mutational screening and copy number variation. Journal of Human Genetics, 2015, 60, 17-25.	1.1	30
10	Genomic imbalances in syndromic congenital heart disease. Jornal De Pediatria, 2017, 93, 497-507.	0.9	27
11	Distal deletion at 22q11.2 as differential diagnosis in Craniofacial Microsomia: Case report and literature review. European Journal of Medical Genetics, 2018, 61, 262-268.	0.7	23
12	Feeding Infants with Cleft Lip and/or Palate in Brazil: Suggestions to Improve Health Policy and Research. Cleft Palate-Craniofacial Journal, 2013, 50, 577-590.	0.5	22
13	Insertional translocation of 15q25â€q26 into 11p13 and duplication at 8p23.1 characterized by high resolution arrays in a boy with congenital malformations and aniridia. American Journal of Medical Genetics, Part A, 2012, 158A, 2905-2910.	0.7	18
14	Brazil's Craniofacial Project: Genetic Evaluation and Counseling in the Reference Network for Craniofacial Treatment. Cleft Palate-Craniofacial Journal, 2006, 43, 577-579.	0.5	17
15	Atypical copy number abnormalities in 22q11.2 region: Report of three cases. European Journal of Medical Genetics, 2013, 56, 515-520.	0.7	17
16	Clinical, cytogenetic, and molecular characterization of six patients with ring chromosomes 22, including one with concomitant 22q11.2 deletion. American Journal of Medical Genetics, Part A, 2014, 164, 1659-1665.	0.7	16
17	Clinical Features in Patients with 22q11.2 Deletion Syndrome Ascertained by Palatal Abnormalities. Cleft Palate-Craniofacial Journal, 2015, 52, 411-416.	0.5	16
18	Ancestry Informative Marker Panel to Estimate Population Stratification Using Genomeâ€wide Human Array. Annals of Human Genetics, 2017, 81, 225-233.	0.3	16

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19	Q289P Mutation in FGFR2 Gene Causes Saethre-Chotzen Syndrome: Some Considerations About Familial Heterogeneity. Cleft Palate-Craniofacial Journal, 2006, 43, 142-147.	0.5	15
20	Genetics and public health: the experience of a reference center for diagnosis of 22q11.2 deletion in Brazil and suggestions for implementing genetic testing. Journal of Community Genetics, 2013, 4, 99-106.	0.5	15
21	Risk factors and the prevention of oral clefts. Brazilian Oral Research, 2014, 28, 1-5.	0.6	15
22	17p13.3 Microdeletion: Insights on Genotype-Phenotype Correlation. Molecular Syndromology, 2017, 8, 36-41.	0.3	15
23	Maternally inherited partial monosomy 9p (pterâ€‰â†'â€‰p24.1) and partial trisomy 20p (pterâ€‰â†'â€‰p12.1) characterized by microarray comparative genomic hybridization. American Journal of Medical Genetics, Part A, 2011, 155, 2754-2761.	0.7	13
24	Preliminary Analysis of the Nonsynonymous Polymorphism rs17563 in BMP4 Gene in Brazilian Population Suggests Protection for Nonsyndromic Cleft Lip and Palate. Plastic Surgery International, 2012, 2012, 1-5.	0.7	13
25	Study of <scp>IRF</scp>6 and 8q24 region in nonâ€syndromic oral clefts in the Brazilian population. Oral Diseases, 2016, 22, 241-245.	1.5	13
26	Blepharocheilodontic (BCD) syndrome: Expanding the phenotype?. American Journal of Medical Genetics Part A, 2003, 121A, 266-270.	2.4	12
27	Diagnostic implications of associated defects in patients with typical orofacial clefts. Jornal De Pediatria, 2015, 91, 485-492.	0.9	12
28	Application of highâ€resolution array platform for genomeâ€wide copy number variation analysis in patients with nonsyndromic cleft lip and palate. Journal of Clinical Laboratory Analysis, 2018, 32, e22428.	0.9	12
29	Evaluation of Craniofacial Care outside the Brazilian Reference Network for Craniofacial Treatment. Cleft Palate-Craniofacial Journal, 2009, 46, 204-211.	0.5	11
30	Testing criteria for 22q11.2 deletion syndrome: preliminary results of a low cost strategy for public health. Orphanet Journal of Rare Diseases, 2019, 14, 123.	1.2	11
31	Frontonasal dysplasia, Poland anomaly and unilateral hypoplasia of lower limb: report on a male patient. Clinical Dysmorphology, 2003, 12, 233-236.	0.1	10
32	8p23.1 Interstitial Deletion in a Patient with Congenital Cardiopathy, Neurobehavioral Disorders, and Minor Signs Suggesting 22q11.2 Deletion Syndrome. Journal of Developmental and Behavioral Pediatrics, 2015, 36, 544-548.	0.6	10
33	A boy with partial dup(18q)/del(18p) due to a maternal pericentric inversion: Genotypeâ€"phenotype correlation and risk of recombinant chromosomes based on systematic review of the literature. American Journal of Medical Genetics, Part A, 2017, 173, 143-150.	0.7	10
34	CranFlow: An Application for Recordâ€Taking and Management Through the Brazilian Database on Craniofacial Anomalies. Birth Defects Research, 2018, 110, 72-80.	0.8	10
35	A recognizable phenotype related to 19p13.12 microdeletion. American Journal of Medical Genetics, Part A, 2018, 176, 1753-1759.	0.7	10
36	Diagnostic Approach to Microdeletion Syndromes Based on 22q11.2 Investigation: Challenges in Four Cases. Molecular Syndromology, 2017, 8, 244-252.	0.3	10

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37	Gillespie Syndrome: Additional Findings and Parental Consanguinity. Ophthalmic Genetics, 2007, 28, 89-93.	0.5	9
38	Preliminary molecular studies on blepharocheilodontic syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 2757-2759.	0.7	9
39	Anthropometric and body-mass composition suggests an intrinsic feature in Williams-Beuren syndrome. Revista Da AssociaÃ§Ã£o MÃ©dica Brasileira, 2011, 57, 681-685.	0.3	9
40	22q11.2 Deletion Syndrome: Laboratory Diagnosis and TBX1 and FGF8 Mutation Screening. Journal of Pediatric Genetics, 2015, 04, 017-022.	0.3	9
41	Copy number variation in the susceptibility to systemic lupus erythematosus. PLoS ONE, 2018, 13, e0206683.	1.1	9
42	Genetic comparison of sickle cell anaemia cohorts from Brazil andÂthe United States reveals high levels of divergence. Scientific Reports, 2019, 9, 10896.	1.6	9
43	IRF6 polymorphisms in Brazilian patients with non-syndromic cleft lip with or without palate. Brazilian Journal of Otorhinolaryngology, 2020, 86, 696-702.	0.4	9
44	Local Strategies to Address Health Needs of Individuals with Orofacial Clefts in Alagoas, Brazil. Cleft Palate-Craniofacial Journal, 2013, 50, 424-431.	0.5	8
45	Genotypeâ€phenotype correlation of 16p13.3 terminal duplication and 22q13.33 deletion: Natural history of a patient and review of the literature. American Journal of Medical Genetics, Part A, 2016, 170, 766-772.	0.7	8
46	Report of two unrelated families with Jalili syndrome and a novel nonsense heterozygous mutation in CNNM4 gene. European Journal of Medical Genetics, 2018, 61, 384-387.	0.7	8
47	Genomic imbalances in craniofacial microsomia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 970-985.	0.7	8
48	Clinical Findings in Four Brazilian Families Affected by Saethre-Chotzen Syndrome without TWIST Mutations. Cleft Palate-Craniofacial Journal, 2004, 41, 250-255.	0.5	7
49	Congenital Temporomandibular Joint Ankylosis: Clinical Characterization and Natural History of Four Unrelated Affected Individuals. Cleft Palate-Craniofacial Journal, 2005, 42, 694-698.	0.5	7
50	A clinical study of 31 individuals with midline facial defects with hypertelorism and a guideline for follow-up. Arquivos De Neuro-Psiquiatria, 2007, 65, 396-401.	0.3	7
51	A familial case with interstitial 2q36 deletion: Variable phenotypic expression in full and mosaic state. European Journal of Medical Genetics, 2012, 55, 660-665.	0.7	7
52	A new case of partial 14q31.3-qter trisomy due to maternal pericentric inversion. Gene, 2013, 523, 192-194.	1.0	7
53	Brazil's Craniofacial Project: Different approaches on orofacial clefts and 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 912-927.	0.7	7
54	Distal 22q11.2 microduplication combined with typical 22q11.2 proximal deletion: A case report. American Journal of Medical Genetics, Part A, 2015, 167, 215-220.	0.7	6

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55	Trisomy X in a patient with childhood-onset systemic lupus erythematosus. Journal of Translational Autoimmunity, 2020, 3, 100043.	2.0	6
56	Identification of genomic imbalances in oral clefts. Jornal De Pediatria, 2021, 97, 321-328.	0.9	6
57	De novo double reciprocal translocations in addition to partial monosomy at another chromosome: A very rare case. Gene, 2015, 573, 166-170.	1.0	5
58	Genomic Investigation of Balanced Chromosomal Rearrangements in Patients with Abnormal Phenotypes. Molecular Syndromology, 2017, 8, 187-194.	0.3	5
59	A New Case of the Rare 10q22.3q23.2 Microdeletion Flanked by Low-Copy Repeats 3/4. Molecular Syndromology, 2017, 8, 161-167.	0.3	5
60	An overview of the trajectory of Brazilian individuals with 22q11.2 deletion syndrome until diagnosis. Orphanet Journal of Rare Diseases, 2022, 17, 67.	1.2	5
61	Cerebellar Involvement in Midline Facial Defects with Ocular Hypertelorism. Cleft Palate-Craniofacial Journal, 2006, 43, 466-470.	0.5	4
62	Central nervous system abnormalities on midline facial defects with hypertelorism detected by magnetic resonance image and computed tomography. Arquivos De Neuro-Psiquiatria, 2006, 64, 916-920.	0.3	3
63	Midline Facial Defects with Hypertelorism and Low-Grade Astrocytoma: A Previously Undescribed Association. Cleft Palate-Craniofacial Journal, 2006, 43, 748-751.	0.5	3
64	Partial monosomy 21 (q11.2â†'q21.3) combined with 3p25.3â†'pter monosomy due to an unbalanced translocation in a patient presenting dysmorphic features and developmental delay. Gene, 2013, 513, 301-304.	1.0	3
65	Pure 21q22.3 deletion identified in a patient with mild phenotypic features. Congenital Anomalies (discontinued), 2018, 58, 178-180.	0.3	3
66	Syndromic Oral Clefts: Challenges of Genetic Assessment in Brazil and Suggestions to Improve Health Policies. Public Health Genomics, 2019, 22, 69-76.	0.6	3
67	Malformations of Cortical Development in Patients with Midline Facial Defects and Ocular Hypertelorism. Cleft Palate-Craniofacial Journal, 2010, 47, 343-351.	0.5	2
68	Investigation of the 22q11.2 candidate region in patients with midline facial defects with hypertelorism. Journal of Applied Genetics, 2010, 51, 219-221.	1.0	2
69	Tetrasomy 3q26.32-q29 due to a supernumerary marker chromosome in a child with pigmentary mosaicism of Ito. Genetics and Molecular Biology, 2016, 39, 35-39.	0.6	2
70	A Rare Case of Concomitant Deletions in 15q11.2 and 19p13.3. Cytogenetic and Genome Research, 2018, 156, 80-86.	0.6	2
71	A Pure 2-Mb 3q26.2 Duplication Proximal to the Critical Region of 3q Duplication Syndrome. Molecular Syndromology, 2018, 9, 197-204.	0.3	2
72	Craniofacial microsomia: Reflections on diagnosis and severity assessment based on a series of cases. Congenital Anomalies (discontinued), 2021, 61, 148-158.	0.3	2

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73	Cerebellar Involvement in Midline Facial Defects With Ocular Hypertelorism. Cleft Palate-Craniofacial Journal, 2006, 43, 466.	0.5	2
74	Time of diagnosis of oral clefts: a multicenter study. Jornal De Pediatria, 2011, 87, 225-30.	0.9	2
75	Infertility and marker chromosomes: Application of molecular cytogenetic techniques in a case of inv dup(15). Journal of Applied Genetics, 2006, 47, 89-91.	1.0	1
76	A Cytogenomic Approach in a Case of Syndromic XY Gonadal Dysgenesis. Sexual Development, 2016, 10, 23-27.	1.1	1
77	ADULT Phenotype and rs16864880 in the TP63 Gene: Two New Cases and Review of the Literature. Molecular Syndromology, 2017, 8, 201-205.	0.3	1
78	Cardiovascular abnormalities in patients with oral cleft: a clinical-electrocardiographic-echocardiographic study. Clinics, 2018, 73, e108.	0.6	1
79	Cover Image, Volume 173A, Number 4, April 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
80	Family care practitioners experience with individuals with orofacial clefts in Brazil. Cadernos Saude Coletiva, 2013, 21, 237-244.	0.2	0