# 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 | 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 |
| 2 | Identification of genomic imbalances in oral clefts. Jornal De Pediatria, 2021, 97, 321-328. | 0.9 | 6 |
| 3 | Craniofacial microsomia: Reflections on diagnosis and severity assessment based on a series of cases. Congenital Anomalies (discontinued), 2021, 61, 148-158. | 0.3 | 2 |
| 4 | 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 |
| 5 | Genomic imbalances in craniofacial microsomia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 970-985. | 0.7 | 8 |
| 6 | 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 |
| 7 | Trisomy X in a patient with childhood-onset systemic lupus erythematosus. Journal of Translational Autoimmunity, 2020, 3, 100043. | 2.0 | 6 |
| 8 | 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 |
| 9 | 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 |
| 10 | 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 |
| 11 | 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 |
| 12 | Pure 21q22.3 deletion identified in a patient with mild phenotypic features. Congenital Anomalies (discontinued), 2018, 58, 178-180. | 0.3 | 3 |
| 13 | 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 |
| 14 | 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 |
| 15 | 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 |
| 16 | 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 |
| 17 | Copy number variation in the susceptibility to systemic lupus erythematosus. PLoS ONE, 2018, 13, e0206683. | 1.1 | 9 |
| 18 | A Rare Case of Concomitant Deletions in 15q11.2 and 19p13.3. Cytogenetic and Genome Research, 2018, 156, 80-86. | 0.6 | 2 |

| # | ARTICLE | IF | CITATIONS |
|---|---|---|---|
| 19 | 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 |
| 20 | A recognizable phenotype related to 19p13.12 microdeletion. American Journal of Medical Genetics, Part A, 2018, 176, 1753-1759. | 0.7 | 10 |
| 21 | Cardiovascular abnormalities in patients with oral cleft: a clinical-electrocardiographic-echocardiographic study. Clinics, 2018, 73, e108. | 0.6 | 1 |
| 22 | 17p13.3 Microdeletion: Insights on Genotype-Phenotype Correlation. Molecular Syndromology, 2017, 8, 36-41. | 0.3 | 15 |
| 23 | 22q11.2 deletion syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 879-888. | 0.7 | 103 |
| 24 | Cover Image, Volume 173A, Number 4, April 2017. American Journal of Medical Genetics, Part A, 2017, 173, i. | 0.7 | 0 |
| 25 | Genomic imbalances in syndromic congenital heart disease. Jornal De Pediatria, 2017, 93, 497-507. | 0.9 | 27 |
| 26 | Genomic Investigation of Balanced Chromosomal Rearrangements in Patients with Abnormal Phenotypes. Molecular Syndromology, 2017, 8, 187-194. | 0.3 | 5 |
| 27 | Ancestry Informative Marker Panel to Estimate Population Stratification Using Genomeâ€wide Human Array. Annals of Human Genetics, 2017, 81, 225-233. | 0.3 | 16 |
| 28 | 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 |
| 29 | 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 |
| 30 | 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 |
| 31 | Diagnostic Approach to Microdeletion Syndromes Based on 22q11.2 Investigation: Challenges in Four Cases. Molecular Syndromology, 2017, 8, 244-252. | 0.3 | 10 |
| 32 | 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 |
| 33 | 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 |
| 34 | 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 |
| 35 | Recessive and Dominant De Novo ITPR1 Mutations Cause Gillespie Syndrome. American Journal of Human Genetics, 2016, 98, 971-980. | 2.6 | 113 |
| 36 | A Cytogenomic Approach in a Case of Syndromic XY Gonadal Dysgenesis. Sexual Development, 2016, 10, 23-27. | 1.1 | 1 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | 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 |
| 38 | 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 |
| 39 | Clinical Features in Patients with 22q11.2 Deletion Syndrome Ascertained by Palatal Abnormalities. Cleft Palate-Craniofacial Journal, 2015, 52, 411-416. | 0.5 | 16 |
| 40 | Diagnostic implications of associated defects in patients with typical orofacial clefts. Jornal De Pediatria, 2015, 91, 485-492. | 0.9 | 12 |
| 41 | DLX4is associated with orofacial clefting and abnormal jaw development. Human Molecular Genetics, 2015, 24, 4340-4352. | 1.4 | 36 |
| 42 | 22q11.2 Deletion Syndrome: Laboratory Diagnosis and TBX1 and FGF8 Mutation Screening. Journal of Pediatric Genetics, 2015, 04, 017-022. | 0.3 | 9 |
| 43 | 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 |
| 44 | 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 |
| 45 | 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 |
| 46 | Risk factors and the prevention of oral clefts. Brazilian Oral Research, 2014, 28, 1-5. | 0.6 | 15 |
| 47 | 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 |
| 48 | 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 |
| 49 | A new case of partial 14q31.3-qter trisomy due to maternal pericentric inversion. Gene, 2013, 523, 192-194. | 1.0 | 7 |
| 50 | 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 |
| 51 | Atypical copy number abnormalities in 22q11.2 region: Report of three cases. European Journal of Medical Genetics, 2013, 56, 515-520. | 0.7 | 17 |
| 52 | 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 |
| 53 | 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 |
| 54 | 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 |

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|---|---|---|---|
| 55 | Family care practitioners experience with individuals with orofacial clefts in Brazil. Cadernos Saude Coletiva, 2013, 21, 237-244. | 0.2 | 0 |
| 56 | 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 |
| 57 | 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 |
| 58 | 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 |
| 59 | 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 |
| 60 | 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 |
| 61 | Time of diagnosis of oral clefts: a multicenter study. Jornal De Pediatria, 2011, 87, 225-30. | 0.9 | 2 |
| 62 | Malformations of Cortical Development in Patients with Midline Facial Defects and Ocular Hypertelorism. Cleft Palate-Craniofacial Journal, 2010, 47, 343-351. | 0.5 | 2 |
| 63 | 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 |
| 64 | Evaluation of Craniofacial Care outside the Brazilian Reference Network for Craniofacial Treatment. Cleft Palate-Craniofacial Journal, 2009, 46, 204-211. | 0.5 | 11 |
| 65 | 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 |
| 66 | Gillespie Syndrome: Additional Findings and Parental Consanguinity. Ophthalmic Genetics, 2007, 28, 89-93. | 0.5 | 9 |
| 67 | 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 |
| 68 | Preliminary molecular studies on blepharocheilodontic syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 2757-2759. | 0.7 | 9 |
| 69 | 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 |
| 70 | 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 |
| 71 | 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 |
| 72 | Cerebellar Involvement in Midline Facial Defects with Ocular Hypertelorism. Cleft Palate-Craniofacial Journal, 2006, 43, 466-470. | 0.5 | 4 |

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|---|---|---|---|
| 73 | Midline Facial Defects with Hypertelorism and Low-Grade Astrocytoma: A Previously Undescribed Association. Cleft Palate-Craniofacial Journal, 2006, 43, 748-751. | 0.5 | 3 |
| 74 | 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 |
| 75 | Cerebellar Involvement in Midline Facial Defects With Ocular Hypertelorism. Cleft Palate-Craniofacial Journal, 2006, 43, 466. | 0.5 | 2 |
| 76 | 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 |
| 77 | 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 |
| 78 | 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 |
| 79 | Blepharocheilodontic (BCD) syndrome: Expanding the phenotype?. American Journal of Medical Genetics Part A, 2003, 121A, 266-270. | 2.4 | 12 |
| 80 | Frontonasal dysplasia, Poland anomaly and unilateral hypoplasia of lower limb: report on a male patient. Clinical Dysmorphology, 2003, 12, 233-236. | 0.1 | 10 |