# Nara Lygia Sobreira

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4898802/publications.pdf

Version: 2024-02-01

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|---|---|---|---|
| 64<br>papers | 3,534<br>citations | 257450<br>24<br>h-index | 155660<br>55<br>g-index |
| 67<br>all docs | 67<br>docs citations | 67<br>times ranked | 9438<br>citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Expansion of the phenotypic and mutational spectrum of Carpenter syndrome. European Journal of Medical Genetics, 2022, 65, 104377. | 1.3 | 3 |
| 2 | Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797. | 2.4 | 44 |
| 3 | Variantâ€level matching for diagnosis and discovery: Challenges and opportunities. Human Mutation, 2022, , . | 2.5 | 11 |
| 4 | The impact of GeneMatcher on international data sharing and collaboration. Human Mutation, 2022, , . | 2.5 | 7 |
| 5 | Genomeâ€wide <scp>DNA</scp> methylation profiling confirms a case of lowâ€level mosaic Kabuki syndrome 1. American Journal of Medical Genetics, Part A, 2022, 188, 2217-2225. | 1.2 | 14 |
| 6 | Progressive liver, kidney, and heart degeneration in children and adults affected by TULP3 mutations. American Journal of Human Genetics, 2022, 109, 928-943. | 6.2 | 22 |
| 7 | Teaching NeuroImages: Trigeminal Ganglia Hypoplasia as Imaging Clue for the Diagnosis of GÃ³mez-LÃ³pez-HernÃ¡ndez Syndrome. Neurology, 2021, 96, e1593-e1594. | 1.1 | 1 |
| 8 | Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133. | 1.2 | 17 |
| 9 | Clinical and molecular characterization of patients with hereditary hemorrhagic telangiectasia: Experience from an HHT Center of Excellence. American Journal of Medical Genetics, Part A, 2021, 185, 1981-1990. | 1.2 | 4 |
| 10 | Pathogenic alleles in microtubule, secretory granule and extracellular matrix-related genes in familial keratoconus. Human Molecular Genetics, 2021, 30, 658-671. | 2.9 | 12 |
| 11 | Nextâ€generation sequencing and the evolution of data sharing. American Journal of Medical Genetics, Part A, 2021, 185, 2633-2635. | 1.2 | 4 |
| 12 | UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. Genetics in Medicine, 2021, 23, 1624-1635. | 2.4 | 7 |
| 13 | Germline ERBB2/HER2 Coding Variants Are Associated with Increased Risk of Myeloproliferative Neoplasms. Cancers, 2021, 13, 3246. | 3.7 | 5 |
| 14 | IFIH1 loss-of-function variants contribute to very early-onset inflammatory bowel disease. Human Genetics, 2021, 140, 1299-1312. | 3.8 | 17 |
| 15 | Variants of human CLDN9 cause mild to profound hearing loss. Human Mutation, 2021, 42, 1321-1335. | 2.5 | 5 |
| 16 | PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. Orphanet Journal of Rare Diseases, 2021, 16, 365. | 2.7 | 24 |
| 17 | Using Online Mendelian Inheritance in Man in lowâ€and middleâ€income countries. American Journal of Medical Genetics, Part A, 2021, 185, 3284-3286. | 1.2 | 1 |
| 18 | Clinical and molecular evaluation of 13 Brazilian patients with Gomezâ€LÃ³pezâ€HernÃ¡ndez syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1047-1058. | 1.2 | 8 |

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|---|---|---|---|
| 19 | Embryonic lethal genetic variants and chromosomally normal pregnancy loss. Fertility and Sterility, 2021, 116, 1351-1358. | 1.0 | 5 |
| 20 | Spectrum of genetic variants in moderate to severe sporadic hearing loss in Pakistan. Scientific Reports, 2020, 10, 11902. | 3.3 | 9 |
| 21 | Biallelic ZNF407 mutations in a neurodevelopmental disorder with ID, short stature and variable microcephaly, hypotonia, ocular anomalies and facial dysmorphism. Journal of Human Genetics, 2020, 65, 1115-1123. | 2.3 | 5 |
| 22 | Exome sequencing in patients with microphthalmia, anophthalmia, and coloboma ( MAC ) from a consanguineous population. Clinical Genetics, 2020, 98, 499-506. | 2.0 | 7 |
| 23 | De novo missense variants in LMBRD2 are associated with developmental and motor delays, brain structure abnormalities and dysmorphic features. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-107137. | 3.2 | 3 |
| 24 | Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226. | 2.4 | 22 |
| 25 | <scp>KIAA1217</scp>: A novel candidate gene associated with isolated and syndromic vertebral malformations. American Journal of Medical Genetics, Part A, 2020, 182, 1664-1672. | 1.2 | 15 |
| 26 | Deficiency of Adenosine Deaminase 2 (DADA2): Hidden Variants, Reduced Penetrance, and Unusual Inheritance. Journal of Clinical Immunology, 2020, 40, 917-926. | 3.8 | 32 |
| 27 | The utility of exome sequencing for fetal pleural effusions. Prenatal Diagnosis, 2020, 40, 590-595. | 2.3 | 9 |
| 28 | Gomezâ€“LÃ³pezâ€“HernÃ¡ndez syndrome: A case report with clinical and molecular evaluation and literature review. American Journal of Medical Genetics, Part A, 2020, 182, 1761-1766. | 1.2 | 9 |
| 29 | Role of telomere shortening in anticipation of inflammatory bowel disease. World Journal of Gastrointestinal Pharmacology and Therapeutics, 2020, 11, 69-78. | 1.1 | 5 |
| 30 | Missense Pathogenic variants in KIF4A Affect Dental Morphogenesis Resulting in X-linked Taurodontism, Microdontia and Dens-Invaginatus. Frontiers in Genetics, 2019, 10, 800. | 2.3 | 7 |
| 31 | Pathogenic Variants in NUP214 Cause â€œPluggedâ€•Nuclear Pore Channels and Acute Febrile Encephalopathy. American Journal of Human Genetics, 2019, 105, 48-64. | 6.2 | 29 |
| 32 | Bi-allelic Pro291Leu variant in KCNQ4 leads to early onset non-syndromic hearing loss. Gene, 2019, 705, 109-112. | 2.2 | 7 |
| 33 | ROBO4 variants predispose individuals to bicuspid aortic valve and thoracic aortic aneurysm. Nature Genetics, 2019, 51, 42-50. | 21.4 | 101 |
| 34 | Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812. | 2.4 | 161 |
| 35 | Homozygous deletion in MYL9 expands the molecular basis of megacystisâ€“microcolonâ€“intestinal hypoperistalsis syndrome. European Journal of Human Genetics, 2018, 26, 669-675. | 2.8 | 41 |
| 36 | Peripheral Cone Dystrophy: Expanded Clinical Spectrum, Multimodal and Ultrawide-Field Imaging, and Genomic Analysis. Journal of Ophthalmology, 2018, 2018, 1-13. | 1.3 | 3 |

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|---|---|---|---|
| 37 | No major role for rare plectin variants in arrhythmogenic right ventricular cardiomyopathy. PLoS ONE, 2018, 13, e0203078. | 2.5 | 2 |
| 38 | SLC13A5is the second gene associated with KohlschÃ¼tterâ€“TÃ¶nz syndrome. Journal of Medical Genetics, 2017, 54, 54-62. | 3.2 | 45 |
| 39 | A novel mutation in GMPPA in siblings with apparent intellectual disability, epilepsy, dysmorphism, and autonomic dysfunction. American Journal of Medical Genetics, Part A, 2017, 173, 2246-2250. | 1.2 | 9 |
| 40 | Mutations in RARS cause a hypomyelination disorder akin to Pelizaeusâ€“Merzbacher disease. European Journal of Human Genetics, 2017, 25, 1134-1141. | 2.8 | 26 |
| 41 | Patients with a Kabuki syndrome phenotype demonstrate DNA methylation abnormalities. European Journal of Human Genetics, 2017, 25, 1335-1344. | 2.8 | 52 |
| 42 | Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with â€œCorner Fracturesâ€. American Journal of Human Genetics, 2017, 101, 815-823. | 6.2 | 37 |
| 43 | A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome. Nature Communications, 2017, 8, 16077. | 12.8 | 72 |
| 44 | Whole-exome sequencing identifies novel variants in PNPT1 causing oxidative phosphorylation defects and severe multisystem disease. European Journal of Human Genetics, 2017, 25, 79-84. | 2.8 | 33 |
| 45 | Family Based Whole Exome Sequencing Reveals the Multifaceted Role of Notch Signaling in Congenital Heart Disease. PLoS Genetics, 2016, 12, e1006335. | 3.5 | 59 |
| 46 | Exome sequencing identifies a de novo frameshift mutation in the imprinted gene ZDBF2 in a sporadic patient with Nasopalpebral Lipomaâ€coloboma syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1934-1937. | 1.2 | 4 |
| 47 | A standardized, evidence-based protocol to assess clinical actionability of genetic disorders associated with genomic variation. Genetics in Medicine, 2016, 18, 1258-1268. | 2.4 | 89 |
| 48 | Gonadal mosaicism in ARID1B gene causes intellectual disability and dysmorphic features in three siblings. American Journal of Medical Genetics, Part A, 2016, 170, 156-161. | 1.2 | 16 |
| 49 | Visceral myopathy: Clinical and molecular survey of a cohort of seven new patients and state of the art of overlapping phenotypes. American Journal of Medical Genetics, Part A, 2016, 170, 2965-2974. | 1.2 | 31 |
| 50 | Two novel germline DDX41 mutations in a family with inherited myelodysplasia/acute myeloid leukemia. Haematologica, 2016, 101, e228-e231. | 3.5 | 47 |
| 51 | GeneMatcher Aids in the Identification of a New Malformation Syndrome with Intellectual Disability, Unique Facial Dysmorphisms, and Skeletal and Connective Tissue Abnormalities Caused by De Novo Variants inHNRNPK. Human Mutation, 2015, 36, 1009-1014. | 2.5 | 56 |
| 52 | NovelCOL2A1Variant (c.619G>A, p.Gly207Arg) Manifesting as a Phenotype Similar to Progressive Pseudorheumatoid Dysplasia and Spondyloepiphyseal Dysplasia, Stanescu Type. Human Mutation, 2015, 36, 1004-1008. | 2.5 | 17 |
| 53 | GeneMatcher: A Matching Tool for Connecting Investigators with an Interest in the Same Gene. Human Mutation, 2015, 36, 928-930. | 2.5 | 1,153 |
| 54 | The Matchmaker Exchange API: Automating Patient Matching Through the Exchange of Structured Phenotypic and Genotypic Profiles. Human Mutation, 2015, 36, 922-927. | 2.5 | 50 |

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|---|---|---|---|
| 55 | Identification and functional characterization of natural human melanocortin 1 receptor mutant alleles in Pakistani population. Pigment Cell and Melanoma Research, 2015, 28, 730-735. | 3.3 | 4 |
| 56 | An anadysplasiaâ€like, spontaneously remitting spondylometaphyseal dysplasia secondary to lamin B receptor (LBR) gene mutations: Further definition of the phenotypic heterogeneity of LBRâ€bone dysplasias. American Journal of Medical Genetics, Part A, 2015, 167, 159-163. | 1.2 | 15 |
| 57 | Assessment of incidental findings in 232 whole-exome sequences from the Baylorâ€“Hopkins Center for Mendelian Genomics. Genetics in Medicine, 2015, 17, 782-788. | 2.4 | 41 |
| 58 | New Tools for Mendelian Disease Gene Identification: PhenoDB Variant Analysis Module; and GeneMatcher, a Web-Based Tool for Linking Investigators with an Interest in the Same Gene. Human Mutation, 2015, 36, 425-431. | 2.5 | 141 |
| 59 | The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215. | 6.2 | 574 |
| 60 | Mutations in SPATA5 Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. American Journal of Human Genetics, 2015, 97, 457-464. | 6.2 | 134 |
| 61 | Novel Deletion of <b><i>SERPINF1</i></b> Causes Autosomal Recessive Osteogenesis Imperfecta Type VI in Two Brazilian Families. Molecular Syndromology, 2014, 5, 268-275. | 0.8 | 18 |
| 62 | Mutations in PCYT1A, Encoding a Key Regulator of Phosphatidylcholine Metabolism, Cause Spondylometaphyseal Dysplasia with Cone-Rod Dystrophy. American Journal of Human Genetics, 2014, 94, 105-112. | 6.2 | 53 |
| 63 | <scp>P</scp> heno <scp>DB</scp> : A New Webâ€Based Tool for the Collection, Storage, and Analysis of Phenotypic Features. Human Mutation, 2013, 34, 566-571. | 2.5 | 64 |
| 64 | Interstitial deletion 5q14.3â€q21 associated with iris coloboma, hearing loss, dental anomaly, moderate intellectual disability, and attention deficit and hyperactivity disorder. American Journal of Medical Genetics, Part A, 2009, 149A, 2581-2583. | 1.2 | 16 |