Nara Lygia Sobreira

List of Publications by Year
in descending order

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

#	Article	IF	Citations
1	GeneMatcher: A Matching Tool for Connecting Investigators with an Interest in the Same Gene. Human Mutation, 2015, 36, 928-930.	2.5	1,153
2	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	6.2	574
3	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	2.4	161
4	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
5	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
6	ROBO4 variants predispose individuals to bicuspid aortic valve and thoracic aortic aneurysm. Nature Genetics, 2019, 51, 42-50.	21.4	101
7	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
8	A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome. Nature Communications, 2017, 8, 16077.	12.8	72
9	<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
10	Family Based Whole Exome Sequencing Reveals the Multifaceted Role of Notch Signaling in Congenital Heart Disease. PLoS Genetics, 2016, 12, e1006335.	3.5	59
11	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
12	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
13	Patients with a Kabuki syndrome phenotype demonstrate DNA methylation abnormalities. European Journal of Human Genetics, 2017, 25, 1335-1344.	2.8	52
14	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
15	Two novel germline DDX41 mutations in a family with inherited myelodysplasia/acute myeloid leukemia. Haematologica, 2016, 101, e228-e231.	3.5	47
16	SLC13A5is the second gene associated with KohlschÃ¼tterâ€“TÃ¶nz syndrome. Journal of Medical Genetics, 2017, 54, 54-62.	3.2	45
17	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
18	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

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19	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
20	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
21	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
22	Deficiency of Adenosine Deaminase 2 (DADA2): Hidden Variants, Reduced Penetrance, and Unusual Inheritance. Journal of Clinical Immunology, 2020, 40, 917-926.	3.8	32
23	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
24	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
25	Mutations in RARS cause a hypomyelination disorder akin to Pelizaeusâ€“Merzbacher disease. European Journal of Human Genetics, 2017, 25, 1134-1141.	2.8	26
26	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. Orphanet Journal of Rare Diseases, 2021, 16, 365.	2.7	24
27	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
28	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
29	Novel Deletion of <i>SERPINF1</i> Causes Autosomal Recessive Osteogenesis Imperfecta Type VI in Two Brazilian Families. Molecular Syndromology, 2014, 5, 268-275.	0.8	18
30	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
31	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
32	IFIH1 loss-of-function variants contribute to very early-onset inflammatory bowel disease. Human Genetics, 2021, 140, 1299-1312.	3.8	17
33	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
34	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
35	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
36	<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

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37	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
38	Pathogenic alleles in microtubule, secretory granule and extracellular matrix-related genes in familial keratoconus. Human Molecular Genetics, 2021, 30, 658-671.	2.9	12
39	Variantâ€level matching for diagnosis and discovery: Challenges and opportunities. Human Mutation, 2022, , .	2.5	11
40	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
41	Spectrum of genetic variants in moderate to severe sporadic hearing loss in Pakistan. Scientific Reports, 2020, 10, 11902.	3.3	9
42	The utility of exome sequencing for fetal pleural effusions. Prenatal Diagnosis, 2020, 40, 590-595.	2.3	9
43	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
44	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
45	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
46	Bi-allelic Pro291Leu variant in KCNQ4 leads to early onset non-syndromic hearing loss. Gene, 2019, 705, 109-112.	2.2	7
47	Exome sequencing in patients with microphthalmia, anophthalmia, and coloboma (MAC) from a consanguineous population. Clinical Genetics, 2020, 98, 499-506.	2.0	7
48	UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. Genetics in Medicine, 2021, 23, 1624-1635.	2.4	7
49	The impact of GeneMatcher on international data sharing and collaboration. Human Mutation, 2022, , .	2.5	7
50	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
51	Germline ERBB2/HER2 Coding Variants Are Associated with Increased Risk of Myeloproliferative Neoplasms. Cancers, 2021, 13, 3246.	3.7	5
52	Variants of human CLDN9 cause mild to profound hearing loss. Human Mutation, 2021, 42, 1321-1335.	2.5	5
53	Role of telomere shortening in anticipation of inflammatory bowel disease. World Journal of Gastrointestinal Pharmacology and Therapeutics, 2020, 11, 69-78.	1.1	5
54	Embryonic lethal genetic variants and chromosomally normal pregnancy loss. Fertility and Sterility, 2021, 116, 1351-1358.	1.0	5

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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	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
57	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
58	Nextâ€generation sequencing and the evolution of data sharing. American Journal of Medical Genetics, Part A, 2021, 185, 2633-2635.	1.2	4
59	Peripheral Cone Dystrophy: Expanded Clinical Spectrum, Multimodal and Ultrawide-Field Imaging, and Genomic Analysis. Journal of Ophthalmology, 2018, 2018, 1-13.	1.3	3
60	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
61	Expansion of the phenotypic and mutational spectrum of Carpenter syndrome. European Journal of Medical Genetics, 2022, 65, 104377.	1.3	3
62	No major role for rare plectin variants in arrhythmogenic right ventricular cardiomyopathy. PLoS ONE, 2018, 13, e0203078.	2.5	2
63	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
64	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