Nara Sobreira

List of Publications by Year
in descending order

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43 papers	3,032 citations	430874 18 h-index	265206 42 g-index
45 all docs	45 docs citations	45 times ranked	8524 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	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
6	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
7	Pathogenic alleles in microtubule, secretory granule and extracellular matrix-related genes in familial keratoconus. Human Molecular Genetics, 2021, 30, 658-671.	2.9	12
8	Germline ERBB2/HER2 Coding Variants Are Associated with Increased Risk of Myeloproliferative Neoplasms. Cancers, 2021, 13, 3246.	3.7	5
9	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. Orphanet Journal of Rare Diseases, 2021, 16, 365.	2.7	24
10	Spectrum of genetic variants in moderate to severe sporadic hearing loss in Pakistan. Scientific Reports, 2020, 10, 11902.	3.3	9
11	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
12	Exome sequencing in patients with microphthalmia, anophthalmia, and coloboma (MAC) from a consanguineous population. Clinical Genetics, 2020, 98, 499-506.	2.0	7
13	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
14	The utility of exome sequencing for fetal pleural effusions. Prenatal Diagnosis, 2020, 40, 590-595.	2.3	9
15	Role of telomere shortening in anticipation of inflammatory bowel disease. World Journal of Gastrointestinal Pharmacology and Therapeutics, 2020, 11, 69-78.	1.1	5
16	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
17	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
18	Bi-allelic Pro291Leu variant in KCNQ4 leads to early onset non-syndromic hearing loss. Gene, 2019, 705, 109-112.	2.2	7

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19	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	2.4	161
20	A novel PRRT2 pathogenic variant in a family with paroxysmal kinesigenic dyskinesia and benign familial infantile seizures. Journal of Physical Education and Sports Management, 2018, 4, a002287.	1.2	9
21	Peripheral Cone Dystrophy: Expanded Clinical Spectrum, Multimodal and Ultrawide-Field Imaging, and Genomic Analysis. Journal of Ophthalmology, 2018, 2018, 1-13.	1.3	3
22	Apparent Acetaminophen Toxicity in a Patient with Transaldolase Deficiency. JIMD Reports, 2018, 44, 9-15.	1.5	12
23	Patients with a Kabuki syndrome phenotype demonstrate DNA methylation abnormalities. European Journal of Human Genetics, 2017, 25, 1335-1344.	2.8	52
24	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
25	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
26	Family Based Whole Exome Sequencing Reveals the Multifaceted Role of Notch Signaling in Congenital Heart Disease. PLoS Genetics, 2016, 12, e1006335.	3.5	59
27	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
28	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
29	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
30	Two novel germline DDX41 mutations in a family with inherited myelodysplasia/acute myeloid leukemia. Haematologica, 2016, 101, e228-e231.	3.5	47
31	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
32	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
33	GeneMatcher: A Matching Tool for Connecting Investigators with an Interest in the Same Gene. Human Mutation, 2015, 36, 928-930.	2.5	1,153
34	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
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	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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37	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
38	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	6.2	574
39	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
40	A Germline Mutation in ERBB3 Predisposes to Inherited Erythroid Myelodysplasia/Erythroleukemia. Blood, 2015, 126, 4105-4105.	1.4	1
41	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
42	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
43	<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