Regie Lyn P Santos-Cortez

List of Publications by Year
in descending order

130 papers	5,823 citations	94269 37 h-index	88477 70 g-index
134 all docs	134 docs citations	134 times ranked	12555 citing authors

#	Article	IF	Citations
1	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
2	Excess of rare, inherited truncating mutations in autism. Nature Genetics, 2015, 47, 582-588.	9.4	531
3	Discovery of common and rare genetic risk variants for colorectal cancer. Nature Genetics, 2019, 51, 76-87.	9.4	377
4	COPA mutations impair ER-Golgi transport and cause hereditary autoimmune-mediated lung disease and arthritis. Nature Genetics, 2015, 47, 654-660.	9.4	302
5	Genetic linkage analysis in the age of whole-genome sequencing. Nature Reviews Genetics, 2015, 16, 275-284.	7.7	225
6	Alterations of the CIB2 calcium- and integrin-binding protein cause Usher syndrome type 1J and nonsyndromic deafness DFNB48. Nature Genetics, 2012, 44, 1265-1271.	9.4	217
7	Novel somatic and germline mutations in intracranial germ cell tumours. Nature, 2014, 511, 241-245.	13.7	181
8	LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. Circulation Research, 2016, 118, 928-934.	2.0	180
9	RNF213 Rare Variants in an Ethnically Diverse Population With Moyamoya Disease. Stroke, 2014, 45, 3200-3207.	1.0	129
10	Mutations of ESRRB Encoding Estrogen-Related Receptor Beta Cause Autosomal-Recessive Nonsyndromic Hearing Impairment DFNB35. American Journal of Human Genetics, 2008, 82, 125-138.	2.6	127
11	Noncoding Mutations of HGF Are Associated with Nonsyndromic Hearing Loss, DFNB39. American Journal of Human Genetics, 2009, 85, 25-39.	2.6	119
12	Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. American Journal of Human Genetics, 2011, 88, 127-137.	2.6	108
13	Functional Null Mutations of MSRB3 Encoding Methionine Sulfoxide Reductase Are Associated with Human Deafness DFNB74. American Journal of Human Genetics, 2011, 88, 19-29.	2.6	107
14	Detection of genotyping errors and pseudo-SNPs via deviations from Hardy-Weinberg equilibrium. Genetic Epidemiology, 2005, 29, 204-214.	0.6	105
15	MAT2A Mutations Predispose Individuals to Thoracic Aortic Aneurysms. American Journal of Human Genetics, 2015, 96, 170-177.	2.6	92
16	Mutations in KARS, Encoding Lysyl-tRNA Synthetase, Cause Autosomal-Recessive Nonsyndromic Hearing Impairment DFNB89. American Journal of Human Genetics, 2013, 93, 132-140.	2.6	90
17	Frequency and Complexity of De Novo Structural Mutation in Autism. American Journal of Human Genetics, 2016, 98, 667-679.	2.6	88
18	Guidelines for Large-Scale Sequence-Based Complex Trait Association Studies: Lessons Learned from the NHLBI Exome Sequencing Project. American Journal of Human Genetics, 2016, 99, 791-801.	2.6	79

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19	Mutations in TBC1D24, a Gene Associated With Epilepsy, Also Cause Nonsyndromic Deafness DFNB86. American Journal of Human Genetics, 2014, 94, 144-152.	2.6	72
20	Variant Association Tools for Quality Control and Analysis of Large-Scale Sequence and Genotyping Array Data. American Journal of Human Genetics, 2014, 94, 770-783.	2.6	71
21	Rare-Variant Extensions of the Transmission Disequilibrium Test: Application to Autism Exome Sequence Data. American Journal of Human Genetics, 2014, 94, 33-46.	2.6	69
22	Mutational Spectrum ofMYO15Aand the Molecular Mechanisms of DFNB3 Human Deafness. Human Mutation, 2016, 37, 991-1003.	1.1	67
23	SimPed: A Simulation Program to Generate Haplotype and Genotype Data for Pedigree Structures. Human Heredity, 2005, 60, 119-122.	0.4	64
24	Homozygosity Mapping Reveals Mutations of GRXCR1 as a Cause of Autosomal-Recessive Nonsyndromic Hearing Impairment. American Journal of Human Genetics, 2010, 86, 138-147.	2.6	58
25	Mutations in the Wolfram Syndrome Type 1 Gene (WFS1) Define a Clinical Entity of Dominant Low-Frequency Sensorineural Hearing Loss. JAMA Otolaryngology, 2003, 129, 411.	1.5	57
26	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. American Journal of Human Genetics, 2020, 106, 234-245.	2.6	56
27	Novel sequence variants in theTMC1 gene in Pakistani families with autosomal recessive hearing impairment. Human Mutation, 2005, 26, 396-396.	1.1	52
28	LTBP3 Pathogenic Variants Predispose Individuals to Thoracic Aortic Aneurysms and Dissections. American Journal of Human Genetics, 2018, 102, 706-712.	2.6	51
29	Genome-wide association study of platelet aggregation in African Americans. BMC Genetics, 2015, 16, 58.	2.7	50
30	Mitochondrial DNA variant interactions modify breast cancer risk. Journal of Human Genetics, 2008, 53, 924-928.	1.1	49
31	Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. Human Mutation, 2019, 40, 53-72.	1.1	48
32	Splice-site mutations in the TRIC gene underlie autosomal recessive nonsyndromic hearing impairment in Pakistani families. Journal of Human Genetics, 2008, 53, 101-105.	1.1	45
33	Shared Genetic Risk Factors of Intracranial, Abdominal, and Thoracic Aneurysms. Journal of the American Heart Association, 2016, 5, .	1.6	45
34	Mutations of GIPC3 cause nonsyndromic hearing loss DFNB72 but not DFNB81 that also maps to chromosome 19p. Human Genetics, 2011, 130, 759-765.	1.8	44
35	Mutational and phenotypic spectra of KCNE1 deficiency in Jervell and Langeâ€Nielsen Syndrome and Romanoâ€Ward Syndrome. Human Mutation, 2019, 40, 162-176.	1.1	44
36	Autosomal-Recessive Hearing Impairment Due to Rare Missense Variants within S1PR2. American Journal of Human Genetics, 2016, 98, 331-338.	2.6	43

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37	SMAD3pathogenic variants: risk for thoracic aortic disease and associated complications from the Montalcino Aortic Consortium. Journal of Medical Genetics, 2019, 56, 252-260.	1.5	43
38	Novel candidate genes and variants underlying autosomal recessive neurodevelopmental disorders with intellectual disability. Human Genetics, 2018, 137, 735-752.	1.8	42
39	FUT2 Variants Confer Susceptibility to Familial Otitis Media. American Journal of Human Genetics, 2018, 103, 679-690.	2.6	40
40	The effect of phenotypic outliers and non-normality on rare-variant association testing. European Journal of Human Genetics, 2016, 24, 1188-1194.	1.4	39
41	Rare Variation Facilitates Inferences of Fine-Scale Population Structure in Humans. Molecular Biology and Evolution, 2015, 32, 653-660.	3.5	38
42	Rare A2ML1 variants confer susceptibility to otitis media. Nature Genetics, 2015, 47, 917-920.	9.4	38
43	MYLK pathogenic variants aortic disease presentation, pregnancy risk, and characterization of pathogenic missense variants. Genetics in Medicine, 2019, 21, 144-151.	1.1	36
44	Challenges and solutions for gene identification in the presence of familial locus heterogeneity. European Journal of Human Genetics, 2015, 23, 1207-1215.	1.4	35
45	Identification of CACNA1D variants associated with sinoatrial node dysfunction and deafness in additional Pakistani families reveals a clinical significance. Journal of Human Genetics, 2019, 64, 153-160.	1.1	32
46	Novel CLDN14 mutations in Pakistani families with autosomal recessive nonâ€syndromic hearing loss. American Journal of Medical Genetics, Part A, 2012, 158A, 315-321.	0.7	28
47	FAM92A Underlies Nonsyndromic Postaxial Polydactyly in Humans and an Abnormal Limb and Digit Skeletal Phenotype in Mice. Journal of Bone and Mineral Research, 2019, 34, 375-386.	3.1	27
48	Collapsed haplotype pattern method for linkage analysis of next-generation sequence data. European Journal of Human Genetics, 2015, 23, 1739-1743.	1.4	26
49	The Rare-Variant Generalized Disequilibrium Test for Association Analysis of Nuclear and Extended Pedigrees with Application to Alzheimer Disease WGS Data. American Journal of Human Genetics, 2017, 100, 193-204.	2.6	26
50	Middle ear microbiome differences in indigenous Filipinos with chronic otitis media due to a duplication in the A2ML1 gene. Infectious Diseases of Poverty, 2016, 5, 97.	1.5	24
51	A homozygous missense variant in type I keratin KRT25 causes autosomal recessive woolly hair. Journal of Medical Genetics, 2015, 52, 676-680.	1.5	23
52	Autosomal Dominantly Inherited GREB1L Variants in Individuals with Profound Sensorineural Hearing Impairment. Genes, 2020, 11, 687.	1.0	23
53	Exome sequencing reveals predominantly de novo variants in disorders with intellectual disability (ID) in the founder population of Finland. Human Genetics, 2021, 140, 1011-1029.	1.8	23
54	Longâ€read wholeâ€genome sequencing for the genetic diagnosis of dystrophinopathies. Annals of Clinical and Translational Neurology, 2020, 7, 2041-2046.	1.7	22

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55	A novel autosomal recessive nonsyndromic hearing impairment locus (DFNB42) maps to chromosome 3q13.31-q22.3. American Journal of Medical Genetics, Part A, 2005, 133A, 18-22.	0.7	20
56	Expansion of the spectrum of ITGB6-related disorders to adolescent alopecia, dentogingival abnormalities and intellectual disability. European Journal of Human Genetics, 2016, 24, 1223-1227.	1.4	20
57	Practical approach to the genetic diagnosis of unsolved dystrophinopathies: a stepwise strategy in the genomic era. Journal of Medical Genetics, 2021, 58, 743-751.	1.5	20
58	Novel sequence variants in the TMIE gene in families with autosomal recessive nonsyndromic hearing impairment. Journal of Molecular Medicine, 2006, 84, 226-231.	1.7	19
59	Genetic and Environmental Determinants of Otitis Media in an Indigenous Filipino Population. Otolaryngology - Head and Neck Surgery, 2016, 155, 856-862.	1.1	19
60	Identification of ASAH1 as a susceptibility gene for familial keloids. European Journal of Human Genetics, 2017, 25, 1155-1161.	1.4	19
61	Disparities in discovery of pathogenic variants for autosomal recessive non-syndromic hearing impairment by ancestry. European Journal of Human Genetics, 2019, 27, 1456-1465.	1.4	19
62	A Novel Locus Harbouring a Functional CD164 Nonsense Mutation Identified in a Large Danish Family with Nonsyndromic Hearing Impairment. PLoS Genetics, 2015, 11, e1005386.	1.5	18
63	A variant in LMX1A causes autosomal recessive severe-to-profound hearing impairment. Human Genetics, 2018, 137, 471-478.	1.8	18
64	Novel digenic inheritance of PCDH15 and USH1G underlies profound non-syndromic hearing impairment. BMC Medical Genetics, 2018, 19, 122.	2.1	18
65	Genetic maps of microsatellite and single-nucleotide polymorphism markers: Are the distances accurate?. Genetic Epidemiology, 2003, 24, 243-252.	0.6	17
66	Regulatory variants in TCF7L2 are associated with thoracic aortic aneurysm. American Journal of Human Genetics, 2021, 108, 1578-1589.	2.6	17
67	DFNB68, a novel autosomal recessive non-syndromic hearing impairment locus at chromosomal region 19p13.2. Human Genetics, 2006, 120, 85-92.	1.8	16
68	Confirmation of the Role ofDHX38in the Etiology of Early-Onset Retinitis Pigmentosa. , 2018, 59, 4552.		16
69	A Rare Variant Nonparametric Linkage Method for Nuclear and Extended Pedigrees with Application to Late-Onset Alzheimer Disease via WGS Data. American Journal of Human Genetics, 2019, 105, 822-835.	2.6	16
70	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. Human Genetics, 2019, 138, 593-600.	1.8	16
71	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	1.1	16
72	DFNB39, a recessive form of sensorineural hearing impairment, maps to chromosome 7q11.22â€“q21.12. European Journal of Human Genetics, 2003, 11, 812-815.	1.4	14

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73	Otitis media susceptibility and shifts in the head and neck microbiome due to SPINK5 variants. Journal of Medical Genetics, 2021, 58, 442-452.	1.5	14
74	Localization of A Novel Autosomal Recessive Non-Syndromic Hearing Impairment Locus (DFNB38) to 6q26-q27 in a Consanguineous Kindred from Pakistan. Human Heredity, 2003, 55, 71-74.	0.4	13
75	A novel homozygous variant in BMPR1B underlies acromesomelic dysplasia Hunterâ€“Thompson type. Annals of Human Genetics, 2018, 82, 129-134.	0.3	13
76	Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. American Journal of Human Genetics, 2021, 108, 2368-2384.	2.6	12
77	DFNB44, a Novel Autosomal Recessive Non-Syndromic Hearing Impairment Locus, Maps to Chromosome 7p14.1-q11.22. Human Heredity, 2004, 57, 195-199.	0.4	11
78	DFNB89, a novel autosomal recessive nonsyndromic hearing impairment locus on chromosome 16q21-q23.2. Human Genetics, 2011, 129, 379-385.	1.8	11
79	Generation of sequence-based data for pedigree-segregating Mendelian or Complex traits. Bioinformatics, 2015, 31, 3706-3708.	1.8	10
80	A2ML1and otitis media: novel variants, differential expression, and relevant pathways. Human Mutation, 2019, 40, 1156-1171.	1.1	10
81	Hearing impairment locus heterogeneity and identification of PLS1 as a new autosomal dominant gene in Hungarian Roma. European Journal of Human Genetics, 2019, 27, 869-878.	1.4	10
82	SEQSpark: A Complete Analysis Tool for Large-Scale Rare Variant Association Studies Using Whole-Genome and Exome Sequence Data. American Journal of Human Genetics, 2017, 101, 115-122.	2.6	9
83	A disease-causing novel missense mutation in the ST14 gene underlies autosomal recessive ichthyosis with hypotrichosis syndrome in a consanguineous family. European Journal of Dermatology, 2018, 28, 209-216.	0.3	9
84	Sleepwalking and Sleep Paralysis: Prevalence in Colombian Families With Genetic Generalized Epilepsy. Journal of Child Neurology, 2019, 34, 491-498.	0.7	9
85	Exome sequencing reveals novel variants and unique allelic spectrum for hearing impairment in Filipino cochlear implantees. Clinical Genetics, 2019, 95, 634-636.	1.0	9
86	Splicing Characteristics of Dystrophin Pseudoexons and Identification of a Novel Pathogenic Intronic Variant in the DMD Gene. Genes, 2020, 11, 1180.	1.0	9
87	Identification of Novel Genes and Biological Pathways That Overlap in Infectious and Nonallergic Diseases of the Upper and Lower Airways Using Network Analyses. Frontiers in Genetics, 2019, 10, 1352.	1.1	9
88	Fine-Mapping of Common Genetic Variants Associated with Colorectal Tumor Risk Identified Potential Functional Variants. PLoS ONE, 2016, 11, e0157521.	1.1	8
89	Further confirmation of the association of SLC12A2 with non-syndromic autosomal-dominant hearing impairment. Journal of Human Genetics, 2021, 66, 1169-1175.	1.1	8
90	Genomic analysis of childhood hearing loss in the Yoruba population of Nigeria. European Journal of Human Genetics, 2022, 30, 42-52.	1.4	7

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91	Novel mutations in the genes <scp>TGM</scp>1 and <scp>ALOXE</scp>3 underlying autosomal recessive congenital ichthyosis. International Journal of Dermatology, 2016, 55, 524-530.	0.5	6
92	Sequence variants in nine different genes underlying rare skin disorders in 10 consanguineous families. International Journal of Dermatology, 2017, 56, 1406-1413.	0.5	6
93	The SLC26A4 c.706C>G (p.Leu236Val) Variant is a Frequent Cause of Hearing Impairment in Filipino Cochlear Implantees. Otology and Neurotology, 2018, 39, e726-e730.	0.7	6
94	A de novo SIX1 variant in a patient with a rare nonsyndromic cochleovestibular nerve abnormality, cochlear hypoplasia, and bilateral sensorineural hearing loss. Molecular Genetics & Genomic Medicine, 2019, 7, e995.	0.6	6
95	Further evidence of involvement of TMEM132E in autosomal recessive nonsyndromic hearing impairment. Journal of Human Genetics, 2020, 65, 187-192.	1.1	6
96	Genes Implicated in Rare Congenital Inner Ear and Cochleovestibular Nerve Malformations. Ear and Hearing, 2020, 41, 983-989.	1.0	6
97	ADAMTS1, MPDZ, MVD, and SEZ6: candidate genes for autosomal recessive nonsyndromic hearing impairment. European Journal of Human Genetics, 2021, , .	1.4	6
98	The FUT2 Variant c.461G>A (p.Trp154*) Is Associated With Differentially Expressed Genes and Nasopharyngeal Microbiota Shifts in Patients With Otitis Media. Frontiers in Cellular and Infection Microbiology, 2021, 11, 798246.	1.8	6
99	Mitral regurgitation as a phenotypic manifestation of nonphotosensitive trichothiodystrophy due to a splice variant in MPLKIP. BMC Medical Genetics, 2016, 17, 13.	2.1	5
100	Rare variant associations with waist-to-hip ratio in European-American and African-American women from the NHLBI-Exome Sequencing Project. European Journal of Human Genetics, 2016, 24, 1181-1187.	1.4	5
101	Panel 3: Genomics, precision medicine and targeted therapies. International Journal of Pediatric Otorhinolaryngology, 2020, 130, 109835.	0.4	5
102	Identification of Novel Candidate Genes and Variants for Hearing Loss and Temporal Bone Anomalies. Genes, 2021, 12, 566.	1.0	5
103	Delineating the genotypic and phenotypic spectrum of HECW2-related neurodevelopmental disorders. Journal of Medical Genetics, 2022, 59, 669-677.	1.5	5
104	Maternal mosaicism underlies the inheritance of a rare germline AKT3 variant which is responsible for megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome in two Roma half-siblings. Experimental and Molecular Pathology, 2020, 115, 104471.	0.9	5
105	A Monoallelic Variant in REST Is Associated with Non-Syndromic Autosomal Dominant Hearing Impairment in a South African Family. Genes, 2021, 12, 1765.	1.0	5
106	GJB2 Variants and Auditory Outcomes among Filipino Cochlear Implantees. Audiology and Neurotology Extra, 2013, 3, 1-8.	2.0	4
107	From exomes to genomes: challenges and solutions in population-based genetic association studies. European Journal of Human Genetics, 2017, 25, 395-396.	1.4	4
108	Genetic counseling in an indigenous Filipino community with a high prevalence of A2ML1-related otitis media. Journal of Community Genetics, 2019, 10, 143-151.	0.5	4

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109	ABOGenotype and Blood Type Are Associated with Otitis Media. Genetic Testing and Molecular Biomarkers, 2019, 23, 823-827.	0.3	4
110	Heterozygosity mapping for human dominant trait variants. Human Mutation, 2019, 40, 996-1004.	1.1	4
111	Multi-omic studies on missense PLG variants in families with otitis media. Scientific Reports, 2020, 10, 15035.	1.6	4
112	Wolfram-like syndrome with bicuspid aortic valve due to a homozygous missense variant in CDK13. Journal of Human Genetics, 2021, 66, 1009-1018.	1.1	4
113	The role of CDHR3 in susceptibility to otitis media. Journal of Molecular Medicine, 2021, 99, 1571-1583.	1.7	4
114	Type 1 diabetes loci display a variety of native American and African ancestries in diseased individuals from Northwest Colombia. World Journal of Diabetes, 2019, 10, 534-545.	1.3	4
115	Novel missense and 3â€²-UTR splice site variants in LHFPL5 cause autosomal recessive nonsyndromic hearing impairment. Journal of Human Genetics, 2018, 63, 1099-1107.	1.1	3
116	A quantitative trait rare variant nonparametric linkage method with application to age-at-onset of Alzheimerâ€™s disease. European Journal of Human Genetics, 2020, 28, 1734-1742.	1.4	3
117	Identification of microduplications at Xp21.2 and Xq13.1 in neurodevelopmental disorders. Molecular Genetics & Genomic Medicine, 2021, , e1703.	0.6	3
118	Phenotype Expansion for Atypical Gaucher Disease Due to Homozygous Missense PSAP Variant in a Large Consanguineous Pakistani Family. Genes, 2022, 13, 662.	1.0	3
119	Microbiota Associated With Cholesteatoma Tissue in Chronic Suppurative Otitis Media. Frontiers in Cellular and Infection Microbiology, 2022, 12, 746428.	1.8	3
120	Novel Variants in Hearing Loss Genes and Associations With Audiometric Thresholds in a Multi-ethnic Cohort of US Patients With Cochlear Implants. Otology and Neurotology, 2020, 41, 978-985.	0.7	2
121	Identification of autosomal recessive nonsyndromic hearing impairment genes through the study of consanguineous and non-consanguineous families: past, present, and future. Human Genetics, 2022, 141, 413-430.	1.8	2
122	Editorial: Otitis Media Genomics and the Middle Ear Microbiome. Frontiers in Genetics, 2021, 12, 763688.	1.1	2
123	SCN1A Variants as the Underlying Cause of Genetic Epilepsy with Febrile Seizures Plus in Two Multi-Generational Colombian Families. Genes, 2022, 13, 754.	1.0	2
124	Autosomal recessive nonsyndromic hearing impairment in two Finnish families due to the population enriched CABP2 c.637+1G>T variant. Molecular Genetics & Genomic Medicine, 2022, , e1866.	0.6	1
125	MendelProb: probability and sample size calculations for Mendelian studies of exome and whole genome sequence data. Bioinformatics, 2019, 35, 529-531.	1.8	0
126	A Start Codon Variant in NOG Underlies Symphalangism and Ossicular Chain Malformations Affecting Both the Incus and the Stapes. Case Reports in Genetics, 2019, 2019, 1-5.	0.1	0

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127	Positive Selection of a Pre-expansion CAG Repeat of the Human SCA2 Gene. PLoS Genetics, 2005, preprint, e41.	1.5	0
128	Platelet Reactivity Is Associated with VAMP8 Expression and a VAMP8 3â€²UTR Polymorphism. Blood, 2008, 112, 5366-5366.	0.6	0
129	A nonâ€coding<scp>RNASEH1</scp>gene variant associates with type 1 diabetes and interacts with<scp>HLA tagSNPs</scp>in families from Colombia. Pediatric Diabetes, 2020, 21, 1183-1192.	1.2	0
130	Exome Sequencing Identifies a Novel FBN1 Variant in a Pakistani Family with Marfan Syndrome That Includes Left Ventricle Diastolic Dysfunction. Genes, 2021, 12, 1915.	1.0	0