Regie Lyn P Santos-Cortez

List of Publications by Year
in descending order

130 papers	5,823 citations	108046 37 h-index	100535 70 g-index
134 all docs	134 docs citations	134 times ranked	13758 citing authors

#	Article	IF	Citations
1	Delineating the genotypic and phenotypic spectrum of HECW2-related neurodevelopmental disorders. Journal of Medical Genetics, 2022, 59, 669-677.	1.5	5
2	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
3	Genomic analysis of childhood hearing loss in the Yoruba population of Nigeria. European Journal of Human Genetics, 2022, 30, 42-52.	1.4	7
4	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
5	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
6	Microbiota Associated With Cholesteatoma Tissue in Chronic Suppurative Otitis Media. Frontiers in Cellular and Infection Microbiology, 2022, 12, 746428.	1.8	3
7	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
8	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
9	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
10	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
11	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
12	Identification of Novel Candidate Genes and Variants for Hearing Loss and Temporal Bone Anomalies. Genes, 2021, 12, 566.	1.0	5
13	Identification of microduplications at Xp21.2 and Xq13.1 in neurodevelopmental disorders. Molecular Genetics & Genomic Medicine, 2021, , e1703.	0.6	3
14	ADAMTS1, MPDZ, MVD, and SEZ6: candidate genes for autosomal recessive nonsyndromic hearing impairment. European Journal of Human Genetics, 2021, , .	1.4	6
15	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
16	The role of CDHR3 in susceptibility to otitis media. Journal of Molecular Medicine, 2021, 99, 1571-1583.	1.7	4
17	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	1.1	16
18	Regulatory variants in TCF7L2 are associated with thoracic aortic aneurysm. American Journal of Human Genetics, 2021, 108, 1578-1589.	2.6	17

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19	Editorial: Otitis Media Genomics and the Middle Ear Microbiome. Frontiers in Genetics, 2021, 12, 763688.	1.1	2
20	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
21	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
22	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
23	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
24	Further evidence of involvement of TMEM132E in autosomal recessive nonsyndromic hearing impairment. Journal of Human Genetics, 2020, 65, 187-192.	1.1	6
25	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
26	Panel 3: Genomics, precision medicine and targeted therapies. International Journal of Pediatric Otorhinolaryngology, 2020, 130, 109835.	0.4	5
27	Splicing Characteristics of Dystrophin Pseudoexons and Identification of a Novel Pathogenic Intronic Variant in the DMD Gene. Genes, 2020, 11, 1180.	1.0	9
28	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
29	Multi-omic studies on missense PLG variants in families with otitis media. Scientific Reports, 2020, 10, 15035.	1.6	4
30	Longâ€read wholeâ€genome sequencing for the genetic diagnosis of dystrophinopathies. Annals of Clinical and Translational Neurology, 2020, 7, 2041-2046.	1.7	22
31	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
32	Genes Implicated in Rare Congenital Inner Ear and Cochleovestibular Nerve Malformations. Ear and Hearing, 2020, 41, 983-989.	1.0	6
33	Autosomal Dominantly Inherited GREB1L Variants in Individuals with Profound Sensorineural Hearing Impairment. Genes, 2020, 11, 687.	1.0	23
34	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
35	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
36	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

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37	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
38	MYLK pathogenic variants aortic disease presentation, pregnancy risk, and characterization of pathogenic missense variants. Genetics in Medicine, 2019, 21, 144-151.	1.1	36
39	MendelProb: probability and sample size calculations for Mendelian studies of exome and whole genome sequence data. Bioinformatics, 2019, 35, 529-531.	1.8	0
40	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
41	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
42	ABOGenotype and Blood Type Are Associated with Otitis Media. Genetic Testing and Molecular Biomarkers, 2019, 23, 823-827.	0.3	4
43	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
44	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
45	Sleepwalking and Sleep Paralysis: Prevalence in Colombian Families With Genetic Generalized Epilepsy. Journal of Child Neurology, 2019, 34, 491-498.	0.7	9
46	A2ML1and otitis media: novel variants, differential expression, and relevant pathways. Human Mutation, 2019, 40, 1156-1171.	1.1	10
47	Heterozygosity mapping for human dominant trait variants. Human Mutation, 2019, 40, 996-1004.	1.1	4
48	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
49	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
50	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
51	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. Human Genetics, 2019, 138, 593-600.	1.8	16
52	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
53	Discovery of common and rare genetic risk variants for colorectal cancer. Nature Genetics, 2019, 51, 76-87.	9.4	377
54	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

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55	Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. Human Mutation, 2019, 40, 53-72.	1.1	48
56	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
57	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
58	A novel homozygous variant in BMPR1B underlies acromesomelic dysplasia Hunterâ€“Thompson type. Annals of Human Genetics, 2018, 82, 129-134.	0.3	13
59	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
60	FUT2 Variants Confer Susceptibility to Familial Otitis Media. American Journal of Human Genetics, 2018, 103, 679-690.	2.6	40
61	Confirmation of the Role ofDHX38in the Etiology of Early-Onset Retinitis Pigmentosa. , 2018, 59, 4552.		16
62	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
63	Novel candidate genes and variants underlying autosomal recessive neurodevelopmental disorders with intellectual disability. Human Genetics, 2018, 137, 735-752.	1.8	42
64	LTBP3 Pathogenic Variants Predispose Individuals to Thoracic Aortic Aneurysms and Dissections. American Journal of Human Genetics, 2018, 102, 706-712.	2.6	51
65	A variant in LMX1A causes autosomal recessive severe-to-profound hearing impairment. Human Genetics, 2018, 137, 471-478.	1.8	18
66	Novel digenic inheritance of PCDH15 and USH1G underlies profound non-syndromic hearing impairment. BMC Medical Genetics, 2018, 19, 122.	2.1	18
67	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
68	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
69	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
70	Identification of ASAH1 as a susceptibility gene for familial keloids. European Journal of Human Genetics, 2017, 25, 1155-1161.	1.4	19
71	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
72	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

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73	Fine-Mapping of Common Genetic Variants Associated with Colorectal Tumor Risk Identified Potential Functional Variants. PLoS ONE, 2016, 11, e0157521.	1.1	8
74	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
75	LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. Circulation Research, 2016, 118, 928-934.	2.0	180
76	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
77	Mutational Spectrum ofMYO15Aand the Molecular Mechanisms of DFNB3 Human Deafness. Human Mutation, 2016, 37, 991-1003.	1.1	67
78	Genetic and Environmental Determinants of Otitis Media in an Indigenous Filipino Population. Otolaryngology - Head and Neck Surgery, 2016, 155, 856-862.	1.1	19
79	Shared Genetic Risk Factors of Intracranial, Abdominal, and Thoracic Aneurysms. Journal of the American Heart Association, 2016, 5, .	1.6	45
80	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
81	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
82	Autosomal-Recessive Hearing Impairment Due to Rare Missense Variants within S1PR2. American Journal of Human Genetics, 2016, 98, 331-338.	2.6	43
83	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
84	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
85	Frequency and Complexity of De Novo Structural Mutation in Autism. American Journal of Human Genetics, 2016, 98, 667-679.	2.6	88
86	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
87	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
88	Rare Variation Facilitates Inferences of Fine-Scale Population Structure in Humans. Molecular Biology and Evolution, 2015, 32, 653-660.	3.5	38
89	Generation of sequence-based data for pedigree-segregating Mendelian or Complex traits. Bioinformatics, 2015, 31, 3706-3708.	1.8	10
90	MAT2A Mutations Predispose Individuals to Thoracic Aortic Aneurysms. American Journal of Human Genetics, 2015, 96, 170-177.	2.6	92

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91	Rare A2ML1 variants confer susceptibility to otitis media. Nature Genetics, 2015, 47, 917-920.	9.4	38
92	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
93	A homozygous missense variant in type I keratinKRT25causes autosomal recessive woolly hair. Journal of Medical Genetics, 2015, 52, 676-680.	1.5	23
94	COPA mutations impair ER-Golgi transport and cause hereditary autoimmune-mediated lung disease and arthritis. Nature Genetics, 2015, 47, 654-660.	9.4	302
95	Excess of rare, inherited truncating mutations in autism. Nature Genetics, 2015, 47, 582-588.	9.4	531
96	Genetic linkage analysis in the age of whole-genome sequencing. Nature Reviews Genetics, 2015, 16, 275-284.	7.7	225
97	Genome-wide association study of platelet aggregation in African Americans. BMC Genetics, 2015, 16, 58.	2.7	50
98	Collapsed haplotype pattern method for linkage analysis of next-generation sequence data. European Journal of Human Genetics, 2015, 23, 1739-1743.	1.4	26
99	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
100	RNF213 Rare Variants in an Ethnically Diverse Population With Moyamoya Disease. Stroke, 2014, 45, 3200-3207.	1.0	129
101	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
102	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
103	Novel somatic and germline mutations in intracranial germ cell tumours. Nature, 2014, 511, 241-245.	13.7	181
104	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
105	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
106	GJB2 Variants and Auditory Outcomes among Filipino Cochlear Implantees. Audiology and Neurotology Extra, 2013, 3, 1-8.	2.0	4
107	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
108	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

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109	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
110	Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. American Journal of Human Genetics, 2011, 88, 127-137.	2.6	108
111	DFNB89, a novel autosomal recessive nonsyndromic hearing impairment locus on chromosome 16q21-q23.2. Human Genetics, 2011, 129, 379-385.	1.8	11
112	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
113	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
114	Noncoding Mutations of HGF Are Associated with Nonsyndromic Hearing Loss, DFNB39. American Journal of Human Genetics, 2009, 85, 25-39.	2.6	119
115	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
116	Mitochondrial DNA variant interactions modify breast cancer risk. Journal of Human Genetics, 2008, 53, 924-928.	1.1	49
117	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
118	Platelet Reactivity Is Associated with VAMP8 Expression and a VAMP8 3â€²UTR Polymorphism. Blood, 2008, 112, 5366-5366.	0.6	0
119	DFNB68, a novel autosomal recessive non-syndromic hearing impairment locus at chromosomal region 19p13.2. Human Genetics, 2006, 120, 85-92.	1.8	16
120	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
121	Detection of genotyping errors and pseudo-SNPs via deviations from Hardy-Weinberg equilibrium. Genetic Epidemiology, 2005, 29, 204-214.	0.6	105
122	Novel sequence variants in theTMC1 gene in Pakistani families with autosomal recessive hearing impairment. Human Mutation, 2005, 26, 396-396.	1.1	52
123	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
124	SimPed: A Simulation Program to Generate Haplotype and Genotype Data for Pedigree Structures. Human Heredity, 2005, 60, 119-122.	0.4	64
125	Positive Selection of a Pre-expansion CAG Repeat of the Human SCA2 Gene. PLoS Genetics, 2005, preprint, e41.	1.5	0
126	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

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127	Genetic maps of microsatellite and single-nucleotide polymorphism markers: Are the distances accurate?. Genetic Epidemiology, 2003, 24, 243-252.	0.6	17
128	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
129	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
130	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