John R Shaffer

List of Publications by Year
in descending order

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Version: 2024-02-01

82 papers	3,074 citations	218677 26 h-index	206112 48 g-index
90 all docs	90 docs citations	90 times ranked	4698 citing authors

#	Article	IF	Citations
1	Heritability Analysis in Twins Indicates a Genetic Basis for Velopharyngeal Morphology. Cleft Palate-Craniofacial Journal, 2022, 59, 1340-1345.	0.9	1
2	Decreased DNA Methylation of RGMA is Associated with Intracranial Hypertension After Severe Traumatic Brain Injury: An Exploratory Epigenome-Wide Association Study. Neurocritical Care, 2022, 37, 26-37.	2.4	8
3	Genome-wide Interaction Study Implicates VGLL2 and Alcohol Exposure and PRL and Smoking in Orofacial Cleft Risk. Frontiers in Cell and Developmental Biology, 2022, 10, 621261.	3.7	3
4	Genomeâ€wide association study of multiethnic nonsyndromic orofacial cleft families identifies novel loci specific to family and phenotypic subtypes. Genetic Epidemiology, 2022, , .	1.3	4
5	Decoding the Human Face: Progress and Challenges in Understanding the Genetics of Craniofacial Morphology. Annual Review of Genomics and Human Genetics, 2022, 23, 383-412.	6.2	20
6	Racism in oral healthcare settings: Implications for dental <scp>careâ€related</scp> fear/anxiety and utilization among Black/African American women in Appalachia. Journal of Public Health Dentistry, 2022, 82, 28-35.	1.2	4
7	Parents of Children With Nonsyndromic Orofacial Clefting Show Altered Palate Shape. Cleft Palate-Craniofacial Journal, 2021, 58, 847-853.	0.9	4
8	Insights into the genetic architecture of the human face. Nature Genetics, 2021, 53, 45-53.	21.4	94
9	Genome-Wide Association Analysis of Longitudinal Bone Mineral Content Data From the Iowa Bone Development Study. Journal of Clinical Densitometry, 2021, 24, 44-54.	1.2	0
10	Impact of low-frequency coding variants on human facial shape. Scientific Reports, 2021, 11, 748.	3.3	3
11	A GWAS in Latin Americans identifies novel face shape loci, implicating VPS13B and a Denisovan introgressed region in facial variation. Science Advances, 2021, 7, .	10.3	32
12	The Intersection of the Genetic Architectures of Orofacial Clefts and Normal Facial Variation. Frontiers in Genetics, 2021, 12, 626403.	2.3	10
13	Shared heritability of human face and brain shape. Nature Genetics, 2021, 53, 830-839.	21.4	57
14	Genome-Wide Association Study of Non-syndromic Orofacial Clefts in a Multiethnic Sample of Families and Controls Identifies Novel Regions. Frontiers in Cell and Developmental Biology, 2021, 9, 621482.	3.7	16
15	The PAX1 locus at 20p11 is a potential genetic modifier for bilateral cleft lip. Human Genetics and Genomics Advances, 2021, 2, 100025.	1.7	9
16	3D facial phenotyping by biometric sibling matching used in contemporary genomic methodologies. PLoS Genetics, 2021, 17, e1009528.	3.5	13
17	Replication of GWAS significant loci in a sub-Saharan African Cohort with early childhood caries: a pilot study. BMC Oral Health, 2021, 21, 274.	2.3	3
18	Acute Brain-Derived Neurotrophic Factor DNA Methylation Trajectories in Cerebrospinal Fluid and Associations With Outcomes Following Severe Traumatic Brain Injury in Adults. Neurorehabilitation and Neural Repair, 2021, 35, 790-800.	2.9	8

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19	Genome-Wide Association Study (GWAS) of dental caries in diverse populations. BMC Oral Health, 2021, 21, 377.	2.3	16
20	The Influence of Sex and Ancestry on Three-Dimensional Palate Shape. Journal of Craniofacial Surgery, 2021, 32, 2883-2887.	0.7	1
21	Genome scans of facial features in East Africans and cross-population comparisons reveal novel associations. PLoS Genetics, 2021, 17, e1009695.	3.5	13
22	PRICKLE1 Ã— FOCAD Interaction Revealed by Genome-Wide vQTL Analysis of Human Facial Traits. Frontiers in Genetics, 2021, 12, 674642.	2.3	6
23	Oral health and related risk indicators in northâ€central Appalachia differ by rurality. Community Dentistry and Oral Epidemiology, 2021, 49, 427-436.	1.9	1
24	Characterization of cerebrospinal fluid DNA methylation age during the acute recovery period following aneurysmal subarachnoid hemorrhage. , 2021, 1, .		3
25	ANGPT1 methylation and delayed cerebral ischemia in aneurysmal subarachnoid hemorrhage patients. , 2021, 1, .		1
26	Genetic Variability and Trajectories of DNA Methylation May Support a Role for HAMP in Patient Outcomes After Aneurysmal Subarachnoid Hemorrhage. Neurocritical Care, 2020, 32, 550-563.	2.4	10
27	Methylation Data Processing Protocol and Comparison of Blood and Cerebral Spinal Fluid Following Aneurysmal Subarachnoid Hemorrhage. Frontiers in Genetics, 2020, 11, 671.	2.3	8
28	Gene-Based Association Mapping for Dental Caries in The GENEVA Consortium. Journal of Dentistry and Dental Medicine, 2020, 3, .	0.0	0
29	The impact of genetic counseling on patient engagement in a specialty cancer clinic. Journal of Genetic Counseling, 2019, 28, 974-981.	1.6	9
30	A fully adjusted twoâ€stage procedure for rankâ€normalization in genetic association studies. Genetic Epidemiology, 2019, 43, 263-275.	1.3	60
31	Genome-wide analysis of dental caries and periodontitis combining clinical and self-reported data. Nature Communications, 2019, 10, 2773.	12.8	183
32	A systematic genetic analysis and visualization of phenotypic heterogeneity among orofacial cleft GWAS signals. Genetic Epidemiology, 2019, 43, 704-716.	1.3	36
33	Predictors of dental care utilization in northâ€central Appalachia in the USA. Community Dentistry and Oral Epidemiology, 2019, 47, 283-290.	1.9	17
34	Hunting for genes that shape human faces: Initial successes and challenges for the future. Orthodontics and Craniofacial Research, 2019, 22, 207-212.	2.8	22
35	Protocols, Methods, and Tools for Genome-Wide Association Studies (GWAS) of Dental Traits. Methods in Molecular Biology, 2019, 1922, 493-509.	0.9	14
36	Association of lowâ€frequency genetic variants in regulatory regions with nonsyndromic orofacial clefts. American Journal of Medical Genetics, Part A, 2019, 179, 467-474.	1.2	18

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37	Genetic association and differential expression of PITX2 with acute appendicitis. Human Genetics, 2019, 138, 37-47.	3.8	14
38	Novel genetic loci affecting facial shape variation in humans. ELife, 2019, 8, .	6.0	58
39	Genome-wide mapping of global-to-local genetic effects on human facial shape. Nature Genetics, 2018, 50, 414-423.	21.4	205
40	Exploring the genomic basis of early childhood caries: a pilot study. International Journal of Paediatric Dentistry, 2018, 28, 217-225.	1.8	24
41	SNPs Associated With Testosterone Levels Influence Human Facial Morphology. Frontiers in Genetics, 2018, 9, 497.	2.3	23
42	Six NSCL/P Loci Show Associations With Normal-Range Craniofacial Variation. Frontiers in Genetics, 2018, 9, 502.	2.3	20
43	GWAS reveals loci associated with velopharyngeal dysfunction. Scientific Reports, 2018, 8, 8470.	3.3	8
44	Consortium-based genome-wide meta-analysis for childhood dental caries traits. Human Molecular Genetics, 2018, 27, 3113-3127.	2.9	32
45	Novel caries loci in children and adults implicated by genome-wide analysis of families. BMC Oral Health, 2018, 18, 98.	2.3	8
46	Investigating the shared genetics of non-syndromic cleft lip/palate and facial morphology. PLoS Genetics, 2018, 14, e1007501.	3.5	44
47	Mapping genetic variants for cranial vault shape in humans. PLoS ONE, 2018, 13, e0196148.	2.5	11
48	Statistics for Xâ€¢chromosome associations. Genetic Epidemiology, 2018, 42, 539-550.	1.3	16
49	Genetic variants in pachyonychia congenita-associated keratins increase susceptibility to tooth decay. PLoS Genetics, 2018, 14, e1007168.	3.5	12
50	Genome-wide meta-analyses of nonsyndromic orofacial clefts identify novel associations between FOXE1 and all orofacial clefts, and TP63 and cleft lip with or without cleft palate. Human Genetics, 2017, 136, 275-286.	3.8	139
51	Variants on chromosome 4q21 near PKD2 and SIBLINGs are associated with dental caries. Journal of Human Genetics, 2017, 62, 491-496.	2.3	11
52	Association studies of lowâ€frequency coding variants in nonsyndromic cleft lip with or without cleft palate. American Journal of Medical Genetics, Part A, 2017, 173, 1531-1538.	1.2	36
53	Multiethnic GWAS Reveals Polygenic Architecture of Earlobe Attachment. American Journal of Human Genetics, 2017, 101, 913-924.	6.2	29
54	Identification of 16q21 as a modifier of nonsyndromic orofacial cleft phenotypes. Genetic Epidemiology, 2017, 41, 887-897.	1.3	24

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55	Toward a genetic understanding of dental fear: evidence of heritability. Community Dentistry and Oral Epidemiology, 2017, 45, 66-73.	1.9	20
56	A Preliminary Genome-Wide Association Study of Pain-Related Fear: Implications for Orofacial Pain. Pain Research and Management, 2017, 2017, 1-12.	1.8	20
57	Genetic Association ofMMP10,MMP14, andMMP16with Dental Caries. International Journal of Dentistry, 2017, 2017, 1-7.	1.5	12
58	Periodontal Status and Quality of Life: Impact of Fear of Pain and Dental Fear. Pain Research and Management, 2017, 2017, 1-9.	1.8	19
59	Genome-wide association study of facial morphology reveals novel associations with FREM1 and PARK2. PLoS ONE, 2017, 12, e0176566.	2.5	68
60	Genome-Wide Association Study Reveals Multiple Loci Influencing Normal Human Facial Morphology. PLoS Genetics, 2016, 12, e1006149.	3.5	140
61	Metaâ€Analysis of Genomeâ€Wide Association Studies with Correlated Individuals: Application to the Hispanic Community Health Study/Study of Latinos (HCHS/SOL). Genetic Epidemiology, 2016, 40, 492-501.	1.3	16
62	A multi-ethnic genome-wide association study identifies novel loci for non-syndromic cleft lip with or without cleft palate on 2p24.2, 17q23 and 19q13. Human Molecular Genetics, 2016, 25, ddw104.	2.9	163
63	Depression and Rural Environment Are Associated With Poor Oral Health Among Pregnant Women in Northern Appalachia. Behavior Modification, 2016, 40, 325-340.	1.6	20
64	A Genome-wide Association Study of Nonsyndromic Cleft Palate Identifies an Etiologic Missense Variant in GRHL3. American Journal of Human Genetics, 2016, 98, 744-754.	6.2	146
65	Genome-wide association study of dental caries in the Hispanic Communities Health Study/Study of Latinos (HCHS/SOL). Human Molecular Genetics, 2016, 25, 807-816.	2.9	29
66	Genetic Diversity and Association Studies in US Hispanic/Latino Populations: Applications in the Hispanic Community Health Study/Study of Latinos. American Journal of Human Genetics, 2016, 98, 165-184.	6.2	266
67	Oral Health in a Sample of Pregnant Women from Northern Appalachia (2011â€“2015). International Journal of Dentistry, 2015, 2015, 1-12.	1.5	32
68	Caries Experience Differs between Females and Males across Age Groups in Northern Appalachia. International Journal of Dentistry, 2015, 2015, 1-8.	1.5	30
69	Genetic Susceptibility to Dental Caries Differs between the Sexes: A Family-Based Study. Caries Research, 2015, 49, 133-140.	2.0	56
70	Using genetics to test the causal relationship of total adiposity and periodontitis: Mendelian randomization analyses in the Gene-Lifestyle Interactions and Dental Endpoints (GLIDE) Consortium. International Journal of Epidemiology, 2015, 44, 638-650.	1.9	54
71	Effects of enamel matrix genes on dental caries are moderated by fluoride exposures. Human Genetics, 2015, 134, 159-167.	3.8	38
72	Genome-Wide Association Studies in Dogs and Humans Identify ADAMTS20 as a Risk Variant for Cleft Lip and Palate. PLoS Genetics, 2015, 11, e1005059.	3.5	82

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73	Genome-Wide Association Study of Periodontal Health Measured by Probing Depth in Adults Ages 18â^'49 years. G3: Genes, Genomes, Genetics, 2014, 4, 307-314.	1.8	54
74	Hair keratin mutations in tooth enamel increase dental decay risk. Journal of Clinical Investigation, 2014, 124, 5219-5224.	8.2	43
75	Genome-wide analysis of BMI in adolescents and young adults reveals additional insight into the effects of genetic loci over the life course. Human Molecular Genetics, 2013, 22, 3597-3607.	2.9	116
76	Demographic, socioeconomic, and behavioral factors affecting patterns of tooth decay in the permanent dentition: principal components and factor analyses. Community Dentistry and Oral Epidemiology, 2013, 41, 364-373.	1.9	22
77	Genome-wide association Scan of dental caries in the permanent dentition. BMC Oral Health, 2012, 12, 57.	2.3	69
78	Effects of Smoking and Genotype on the PSR Index of Periodontal Disease in Adults Aged 18â€“49. International Journal of Environmental Research and Public Health, 2012, 9, 2839-2850.	2.6	14
79	Heritable patterns of tooth decay in the permanent dentition: principal components and factor analyses. BMC Oral Health, 2012, 12, 7.	2.3	35
80	Rate of bone loss is greater in young Mexican American men than women: The San Antonio Family Osteoporosis Study. Bone, 2010, 47, 49-54.	2.9	4
81	Quantitative Trait Locus on Chromosome 1q Influences Bone Loss in Young Mexican American Adults. Calcified Tissue International, 2009, 84, 75-84.	3.1	11
82	Decreased Bone Mineral Density Is Correlated with Increased Subclinical Atherosclerosis in Older, but not Younger, Mexican American Women and Men: The San Antonio Family Osteoporosis Study. Calcified Tissue International, 2007, 81, 430-441.	3.1	64