

# 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	Genetic Diversity and Association Studies in US Hispanic/Latino Populations: Applications in the Hispanic Community Health Study/Study of Latinos. <i>American Journal of Human Genetics</i> , 2016, 98, 165-184.	6.2	266
2	Genome-wide mapping of global-to-local genetic effects on human facial shape. <i>Nature Genetics</i> , 2018, 50, 414-423.	21.4	205
3	Genome-wide analysis of dental caries and periodontitis combining clinical and self-reported data. <i>Nature Communications</i> , 2019, 10, 2773.	12.8	183
4	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. <i>Human Molecular Genetics</i> , 2016, 25, ddw104.	2.9	163
5	A Genome-wide Association Study of Nonsyndromic Cleft Palate Identifies an Etiologic Missense Variant in GRHL3. <i>American Journal of Human Genetics</i> , 2016, 98, 744-754.	6.2	146
6	Genome-Wide Association Study Reveals Multiple Loci Influencing Normal Human Facial Morphology. <i>PLoS Genetics</i> , 2016, 12, e1006149.	3.5	140
7	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. <i>Human Genetics</i> , 2017, 136, 275-286.	3.8	139
8	Genome-wide analysis of BMI in adolescents and young adults reveals additional insight into the effects of genetic loci over the life course. <i>Human Molecular Genetics</i> , 2013, 22, 3597-3607.	2.9	116
9	Insights into the genetic architecture of the human face. <i>Nature Genetics</i> , 2021, 53, 45-53.	21.4	94
10	Genome-Wide Association Studies in Dogs and Humans Identify ADAMTS20 as a Risk Variant for Cleft Lip and Palate. <i>PLoS Genetics</i> , 2015, 11, e1005059.	3.5	82
11	Genome-wide association Scan of dental caries in the permanent dentition. <i>BMC Oral Health</i> , 2012, 12, 57.	2.3	69
12	Genome-wide association study of facial morphology reveals novel associations with FREM1 and PARK2. <i>PLoS ONE</i> , 2017, 12, e0176566.	2.5	68
13	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. <i>Calcified Tissue International</i> , 2007, 81, 430-441.	3.1	64
14	A fully adjusted two-stage procedure for rank-normalization in genetic association studies. <i>Genetic Epidemiology</i> , 2019, 43, 263-275.	1.3	60
15	Novel genetic loci affecting facial shape variation in humans. <i>ELife</i> , 2019, 8, .	6.0	58
16	Shared heritability of human face and brain shape. <i>Nature Genetics</i> , 2021, 53, 830-839.	21.4	57
17	Genetic Susceptibility to Dental Caries Differs between the Sexes: A Family-Based Study. <i>Caries Research</i> , 2015, 49, 133-140.	2.0	56
18	Genome-Wide Association Study of Periodontal Health Measured by Probing Depth in Adults Ages 18~49 years. <i>G3: Genes, Genomes, Genetics</i> , 2014, 4, 307-314.	1.8	54

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19	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. <i>International Journal of Epidemiology</i> , 2015, 44, 638-650.	1.9	54
20	Investigating the shared genetics of non-syndromic cleft lip/palate and facial morphology. <i>PLoS Genetics</i> , 2018, 14, e1007501.	3.5	44
21	Hair keratin mutations in tooth enamel increase dental decay risk. <i>Journal of Clinical Investigation</i> , 2014, 124, 5219-5224.	8.2	43
22	Effects of enamel matrix genes on dental caries are moderated by fluoride exposures. <i>Human Genetics</i> , 2015, 134, 159-167.	3.8	38
23	Association studies of low-frequency coding variants in nonsyndromic cleft lip with or without cleft palate. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1531-1538.	1.2	36
24	A systematic genetic analysis and visualization of phenotypic heterogeneity among orofacial cleft GWAS signals. <i>Genetic Epidemiology</i> , 2019, 43, 704-716.	1.3	36
25	Heritable patterns of tooth decay in the permanent dentition: principal components and factor analyses. <i>BMC Oral Health</i> , 2012, 12, 7.	2.3	35
26	Oral Health in a Sample of Pregnant Women from Northern Appalachia (2011-2015). <i>International Journal of Dentistry</i> , 2015, 2015, 1-12.	1.5	32
27	Consortium-based genome-wide meta-analysis for childhood dental caries traits. <i>Human Molecular Genetics</i> , 2018, 27, 3113-3127.	2.9	32
28	A GWAS in Latin Americans identifies novel face shape loci, implicating VPS13B and a Denisovan introgressed region in facial variation. <i>Science Advances</i> , 2021, 7, .	10.3	32
29	Caries Experience Differs between Females and Males across Age Groups in Northern Appalachia. <i>International Journal of Dentistry</i> , 2015, 2015, 1-8.	1.5	30
30	Genome-wide association study of dental caries in the Hispanic Communities Health Study/Study of Latinos (HCHS/SOL). <i>Human Molecular Genetics</i> , 2016, 25, 807-816.	2.9	29
31	Multiethnic GWAS Reveals Polygenic Architecture of Earlobe Attachment. <i>American Journal of Human Genetics</i> , 2017, 101, 913-924.	6.2	29
32	Identification of 16q21 as a modifier of nonsyndromic orofacial cleft phenotypes. <i>Genetic Epidemiology</i> , 2017, 41, 887-897.	1.3	24
33	Exploring the genomic basis of early childhood caries: a pilot study. <i>International Journal of Paediatric Dentistry</i> , 2018, 28, 217-225.	1.8	24
34	SNPs Associated With Testosterone Levels Influence Human Facial Morphology. <i>Frontiers in Genetics</i> , 2018, 9, 497.	2.3	23
35	Demographic, socioeconomic, and behavioral factors affecting patterns of tooth decay in the permanent dentition: principal components and factor analyses. <i>Community Dentistry and Oral Epidemiology</i> , 2013, 41, 364-373.	1.9	22
36	Hunting for genes that shape human faces: Initial successes and challenges for the future. <i>Orthodontics and Craniofacial Research</i> , 2019, 22, 207-212.	2.8	22

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37	Depression and Rural Environment Are Associated With Poor Oral Health Among Pregnant Women in Northern Appalachia. <i>Behavior Modification</i> , 2016, 40, 325-340.	1.6	20
38	Toward a genetic understanding of dental fear: evidence of heritability. <i>Community Dentistry and Oral Epidemiology</i> , 2017, 45, 66-73.	1.9	20
39	A Preliminary Genome-Wide Association Study of Pain-Related Fear: Implications for Orofacial Pain. <i>Pain Research and Management</i> , 2017, 2017, 1-12.	1.8	20
40	Six NSCL/P Loci Show Associations With Normal-Range Craniofacial Variation. <i>Frontiers in Genetics</i> , 2018, 9, 502.	2.3	20
41	Decoding the Human Face: Progress and Challenges in Understanding the Genetics of Craniofacial Morphology. <i>Annual Review of Genomics and Human Genetics</i> , 2022, 23, 383-412.	6.2	20
42	Periodontal Status and Quality of Life: Impact of Fear of Pain and Dental Fear. <i>Pain Research and Management</i> , 2017, 2017, 1-9.	1.8	19
43	Association of low-frequency genetic variants in regulatory regions with nonsyndromic orofacial clefts. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 467-474.	1.2	18
44	Predictors of dental care utilization in north-central Appalachia in the USA. <i>Community Dentistry and Oral Epidemiology</i> , 2019, 47, 283-290.	1.9	17
45	Meta-Analysis of Genome-Wide Association Studies with Correlated Individuals: Application to the Hispanic Community Health Study/Study of Latinos (HCHS/SOL). <i>Genetic Epidemiology</i> , 2016, 40, 492-501.	1.3	16
46	Statistics for X-chromosome associations. <i>Genetic Epidemiology</i> , 2018, 42, 539-550.	1.3	16
47	Genome-Wide Association Study of Non-syndromic Orofacial Clefts in a Multiethnic Sample of Families and Controls Identifies Novel Regions. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 621482.	3.7	16
48	Genome-Wide Association Study (GWAS) of dental caries in diverse populations. <i>BMC Oral Health</i> , 2021, 21, 377.	2.3	16
49	Effects of Smoking and Genotype on the PSR Index of Periodontal Disease in Adults Aged 18-49. <i>International Journal of Environmental Research and Public Health</i> , 2012, 9, 2839-2850.	2.6	14
50	Protocols, Methods, and Tools for Genome-Wide Association Studies (GWAS) of Dental Traits. <i>Methods in Molecular Biology</i> , 2019, 1922, 493-509.	0.9	14
51	Genetic association and differential expression of PITX2 with acute appendicitis. <i>Human Genetics</i> , 2019, 138, 37-47.	3.8	14
52	3D facial phenotyping by biometric sibling matching used in contemporary genomic methodologies. <i>PLoS Genetics</i> , 2021, 17, e1009528.	3.5	13
53	Genome scans of facial features in East Africans and cross-population comparisons reveal novel associations. <i>PLoS Genetics</i> , 2021, 17, e1009695.	3.5	13
54	Genetic Association of MMP10, MMP14, and MMP16 with Dental Caries. <i>International Journal of Dentistry</i> , 2017, 2017, 1-7.	1.5	12

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55	Genetic variants in pachyonychia congenita-associated keratins increase susceptibility to tooth decay. <i>PLoS Genetics</i> , 2018, 14, e1007168.	3.5	12
56	Quantitative Trait Locus on Chromosome 1q Influences Bone Loss in Young Mexican American Adults. <i>Calcified Tissue International</i> , 2009, 84, 75-84.	3.1	11
57	Variants on chromosome 4q21 near PKD2 and SIBLINGS are associated with dental caries. <i>Journal of Human Genetics</i> , 2017, 62, 491-496.	2.3	11
58	Mapping genetic variants for cranial vault shape in humans. <i>PLoS ONE</i> , 2018, 13, e0196148.	2.5	11
59	Genetic Variability and Trajectories of DNA Methylation May Support a Role for HAMP in Patient Outcomes After Aneurysmal Subarachnoid Hemorrhage. <i>Neurocritical Care</i> , 2020, 32, 550-563.	2.4	10
60	The Intersection of the Genetic Architectures of Orofacial Clefts and Normal Facial Variation. <i>Frontiers in Genetics</i> , 2021, 12, 626403.	2.3	10
61	The impact of genetic counseling on patient engagement in a specialty cancer clinic. <i>Journal of Genetic Counseling</i> , 2019, 28, 974-981.	1.6	9
62	The PAX1 locus at 20p11 is a potential genetic modifier for bilateral cleft lip. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100025.	1.7	9
63	GWAS reveals loci associated with velopharyngeal dysfunction. <i>Scientific Reports</i> , 2018, 8, 8470.	3.3	8
64	Novel caries loci in children and adults implicated by genome-wide analysis of families. <i>BMC Oral Health</i> , 2018, 18, 98.	2.3	8
65	Methylation Data Processing Protocol and Comparison of Blood and Cerebral Spinal Fluid Following Aneurysmal Subarachnoid Hemorrhage. <i>Frontiers in Genetics</i> , 2020, 11, 671.	2.3	8
66	Acute Brain-Derived Neurotrophic Factor DNA Methylation Trajectories in Cerebrospinal Fluid and Associations With Outcomes Following Severe Traumatic Brain Injury in Adults. <i>Neurorehabilitation and Neural Repair</i> , 2021, 35, 790-800.	2.9	8
67	Decreased DNA Methylation of RGMA is Associated with Intracranial Hypertension After Severe Traumatic Brain Injury: An Exploratory Epigenome-Wide Association Study. <i>Neurocritical Care</i> , 2022, 37, 26-37.	2.4	8
68	PRICKLE1 – FOCAD Interaction Revealed by Genome-Wide vQTL Analysis of Human Facial Traits. <i>Frontiers in Genetics</i> , 2021, 12, 674642.	2.3	6
69	Rate of bone loss is greater in young Mexican American men than women: The San Antonio Family Osteoporosis Study. <i>Bone</i> , 2010, 47, 49-54.	2.9	4
70	Parents of Children With Nonsyndromic Orofacial Clefting Show Altered Palate Shape. <i>Cleft Palate-Craniofacial Journal</i> , 2021, 58, 847-853.	0.9	4
71	Genome-wide association study of multiethnic nonsyndromic orofacial cleft families identifies novel loci specific to family and phenotypic subtypes. <i>Genetic Epidemiology</i> , 2022, , .	1.3	4
72	Racism in oral healthcare settings: Implications for dental care-related fear/anxiety and utilization among Black/African American women in Appalachia. <i>Journal of Public Health Dentistry</i> , 2022, 82, 28-35.	1.2	4

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73	Impact of low-frequency coding variants on human facial shape. Scientific Reports, 2021, 11, 748.	3.3	3
74	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
75	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
76	Characterization of cerebrospinal fluid DNA methylation age during the acute recovery period following aneurysmal subarachnoid hemorrhage. , 2021, 1, .		3
77	The Influence of Sex and Ancestry on Three-Dimensional Palate Shape. Journal of Craniofacial Surgery, 2021, 32, 2883-2887.	0.7	1
78	Heritability Analysis in Twins Indicates a Genetic Basis for Velopharyngeal Morphology. Cleft Palate-Craniofacial Journal, 2022, 59, 1340-1345.	0.9	1
79	Oral health and related risk indicators in north-central Appalachia differ by rurality. Community Dentistry and Oral Epidemiology, 2021, 49, 427-436.	1.9	1
80	ANGPT1 methylation and delayed cerebral ischemia in aneurysmal subarachnoid hemorrhage patients. , 2021, 1, .		1
81	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
82	Gene-Based Association Mapping for Dental Caries in The GENEVA Consortium. Journal of Dentistry and Dental Medicine, 2020, 3, .	0.0	0