

## Curriculum Vitae

### John R. Shaffer

Department of Human Genetics  
 3120 Public Health  
 130 De Soto Street  
 Pittsburgh, PA 15261

Phone: 412-983-3816

Fax: 412-624-3020

E-mail: john.r.shaffer@pitt.edu

## Education and Training

### Undergraduate

1999-2003	Schreyer Honors College, Pennsylvania State University, State College, PA	B.S., 2003	Biology
-----------	---	------------	---------

### Graduate

2003-2008	University of Pittsburgh, Pittsburgh, PA	PhD, 2008	Human Genetics
-----------	--	-----------	----------------

### Post-Graduate

2008-2011	University of Pittsburgh, Pittsburgh, PA	Post-doctoral	Human Genetics
-----------	--	---------------	----------------

## Appointments and Positions

### Academic

2011-2013	Research Assistant Professor	Department of Human Genetics
2013-present	Assistant Professor	Graduate School of Public Health
2020-present	tenure-stream	University of Pittsburgh, Pittsburgh, PA
2015-present	Graduate Faculty	University of Pittsburgh, Pittsburgh, PA
2017-present	Assistant Professor	Department of Oral and Craniofacial Sciences School of Dental Medicine University of Pittsburgh, Pittsburgh, PA
2021-present	Vice Chair for Education	Department of Human Genetics
2021-present	Director of Graduate Studies	Graduate School of Public Health,
2021-present	Director, MS and PhD Programs	University of Pittsburgh, Pittsburgh, PA

## Membership in Professional and Scientific Societies

2004-present	American Society of Human Genetics
2005-2013	American Society of Bone and Mineral Research
2006-2007	American Public Health Association
2014-present	American Association for Dental Research

2014-present International Association for Dental Research  
 2017-present American Association for the Advancement of Science  
 2018-2019 ACPA American Cleft Palate- Craniofacial Association

## Honors

2006 Delta Omega national poster competition prize  
 American Public Health Association 134<sup>th</sup> Annual Meeting

2006 Winner in the graduate student category  
 University of Pittsburgh, Institute on Aging 1<sup>st</sup> Annual Meeting

2006 First Prize for research in the doctoral student category  
 University of Pittsburgh, Graduate School of Public Health 8<sup>th</sup> Annual Dean’s Day

2006 Herbert S. Rosenkranz Award for Public Health Research  
 University of Pittsburgh, Graduate School of Public Health 8<sup>th</sup> Annual Dean’s Day

2008 Outstanding Student Award  
 University of Pittsburgh, Graduate School of Public Health

2013 William J. Gies Award for Biological Research  
 International and American Associations for Dental Research (IADR/AADR)

2014 Excellence in Teaching Certificate  
 University of Pittsburgh, Graduate School of Public Health

2014 Innovation in Education Award  
 University of Pittsburgh, Provost’s Advisory Council on Instructional Excellence

2017 Giddon Award for Distinguished Research in the Behavioral Sciences  
 International Association for Dental Research (IADR)

2018 Nomination for the James L. Craig Excellence in Teaching Award, University of Pittsburgh, Graduate School of Public Health

2019 Nomination for the James L. Craig Excellence in Teaching Award, University of Pittsburgh, Graduate School of Public Health

2020 James L. Craig Excellence in Teaching Award, University of Pittsburgh, Graduate School of Public Health

2021 Nomination for the Chancellor’s Distinguished Teaching Award, chosen by the Graduate School of Public Health’s selection committee as the School’s nominee for this Office of the Provost Award, University of Pittsburgh

## Teaching and Mentorship

### Courses

Years	Course Number: Title Institution	Hours of Lecture, credits Average Enrollment	Role in course (% teaching effort)
2002	BIOL 110: Introductory Biology: Basic Concepts and Biodiversity The Pennsylvania State University	3 hours per week, 1 credit, 25 students	Lab instructor (100%) / teaching assistant

<b>Years</b>	<b>Course Number: Title Institution</b>	<b>Hours of Lecture, credits Average Enrollment</b>	<b>Role in course (% teaching effort)</b>
2003	BIOL 240: Introductory Biology: Function and Development of Organisms The Pennsylvania State University	3 hours per week 1 credit 25 students	Lab instructor (100%) / teaching assistant
2008	HUGEN 2022: Human Population Genetics University of Pittsburgh	2 hours per week 2 credits 30 students	Teaching assistant
2009-2012	HUGEN 2022: Human Population Genetics University of Pittsburgh	2 hours per week 2 credits 30 students	Co-instructor (30%)
2013-2015	HUGEN 2022: Human Population Genetics University of Pittsburgh	2 hours per week 2 credits 30 students	Course director / co-instructor (50%)
2016-present	HUGEN 2022: Human Population Genetics University of Pittsburgh	2 hours per week 2 credits 35-60 students	Course director / instructor (100%)
2020	HUGEN 2080: Statistical Genetics	3 hours per week 3 credits 10 students	Co-instructor (25%)

#### **Other Teaching**

<b>Year</b>	<b>Type of Teaching</b>	<b>Title</b>
2006	Guest Facilitator	Human Genetics, Department of Medicine, University of Pittsburgh "Problem-based learning: sickle cell disease"
2006	Guest Lecturer	Quantitative Genetics, Department of Human Genetics, University of Pittsburgh "Case study: linkage analysis for hip bone mineral density"
2006	Guest Lecturer	Statistical Methods & Data Mining in Microarray Analysis, Department of Biostatistics, University of Pittsburgh "Missing value imputation in microarray data"
2007	Guest Lecturer	Human Population Genetics, Department of Human Genetics, University of Pittsburgh "The hypothesis testing paradigm and the chi-squared test for Hardy-Weinberg equilibrium"
2007	Guest Lecturer	Human Population Genetics, Department of Human Genetics, University of Pittsburgh "Hardy-Weinberg equilibrium"
2008	Guest Lecturer	Human Population Genetics, Department of Human Genetics, University of Pittsburgh "Statistical distributions, p-values, and the chi-squared test for Hardy-Weinberg equilibrium"
2008	Guest Lecturer	Human Population Genetics, Department of Human Genetics, University of Pittsburgh "Hardy-Weinberg equilibrium"

Year	Type of Teaching	Title
2008	Guest Lecturer	Human Population Genetics, Department of Human Genetics, University of Pittsburgh “Genetic epidemiology: testing for genetic association”
2012	Guest Lecturer	Summer Institute for Training in Biostatistics, Department of Biostatistics, University of Pittsburgh “Oral health and disease: Epidemiology and genetics”
2012	Guest Lecturer	Summer Institute for Training in Biostatistics, Department of Biostatistics, University of Pittsburgh “GWAS of dental caries: Finding genes for tooth decay”
2015	Guest Lecturer	Introduction to Gene Mapping, Department of Human Genetics, University of Pittsburgh, “Following-up signals from genome-wide studies”
2019	Discussion Leader	Evidence-based teaching practices workshop series, Graduate School of Public Health, University of Pittsburgh; organized and co-facilitated faculty discussion for the 8-part series to discuss <i>How Learning Works: Seven Research-Based Principles for Smart Teaching</i>
2021	Guest Lecturer	PUBLTH 100 Fundamental of Public Health, University of Pittsburgh, “Introduction to Human Genetics”

#### Major Advisor for Graduate Student Essays, Theses, and Dissertations

Student	Degree, Year	Type of Document and Title • Awards	Notes • Placement
Ekaterina Orolva	MS, April 2015	MS Thesis: Putative regulatory function of GWAS-identified markers of susceptibility to acute appendicitis • Chairman’s Award for Translational Research, Dept. of Human Genetics, University of Pittsburgh	co-advisor with Michael Morowitz • doctoral program, University of Pittsburgh
Kelly Johnson	MS, April 2016	MS Thesis: Genome-wide association studies of childhood bone health • Russel Rule Rycheck Award for Public Health, Graduate School of Public Health, University of Pittsburgh • Outstanding Student Award, Graduate School of Public Health, University of Pittsburgh	• genetic counseling position
Yunqi Li	MS, April 2018	MS Thesis: “Epigenome-wide association study of recovery outcomes of traumatic brain injury patients” • Willian T. Green Award for Public Health Studies, Graduate School of Public Health, University of Pittsburgh	co-advisor with Daniel Weeks • doctoral program, University of Washington

<b>Student</b>	<b>Degree, Year</b>	<b>Type of Document and Title</b> • <b>Awards</b>	<b>Notes</b> • <b>Placement</b>
Amanda Everman	MS, April 2018	MS Thesis: "Genome-wide association study and meta-analysis of missing teeth and functional dentition" • Dean's Scholarship Award, Graduate School of Public Health, University of Pittsburgh	• Statistical Programmer/Scientific Affairs Analyst, PRA Health Sciences
Julia Verbiar	MS, April 2018	MS Thesis: "Genetics of the relationship between periodontitis and cardiovascular disease"	• genetic counseling position
Yuqiao Zhou	PhD, Dec. 2018	Dissertation: "Investigating the role of ER stress sensor IRE1 $\alpha$ in postnatal tooth development" • Dean's Scholarship Award, School of Dental Medicine • American Association for Dental Research Pittsburgh Chapter Award	• post-doctoral associate, University of Pittsburgh
Annie Infancia Arockiaraj	MS, May 2019	MS Thesis: "Epigenome-wide association studies in aneurysmal subarachnoid hemorrhage patients"	co-advisor with Daniel Weeks • doctoral program, University of Pittsburgh
Ekaterina Orlova	PhD, (in progress)	Dissertation: "Genome-wide association studies of early childhood caries"	
Dongjing Liu	PhD, Dec. 2020	Dissertation: "Integrated genome-wide analysis of normal-range facial variation" • First Place Poster Award, 2020 International Genetic Epidemiology Society • C.C. and Clara Li Award, Dept. of Human Genetics, University of Pittsburgh • Outstanding Student Award, Graduate School of Public Health, University of Pittsburgh • Robert E. Ferrell Award for Excellence, Dept. of Human Genetics, University of Pittsburgh • Frist Place Poster Award, 2021 Internation Genetic Epidemiology Society	• post-doctoral fellow, Icahn School of Medicine at Mount Sinai
Tianyu Zou	PhD (in progress)	Dissertation: (Title TBD) • C.C. and Clara Li Award, Dept. of Human Genetic, University of Pittsburgh	
Kestrel Allawos	MPH (in progress)	MPH Essay, TBD	co-advisor with Yvette Conley
Tara Harmony	MPH (in progress)	MPH Essay, TBD	co-advisor with Yvette Conley

### Service on Masters or Doctoral Committees

Year	Student	Degree	Title of Dissertation/Essay
2010-2012	Zhen Zeng	MS	Genome-wide association studies of pit and fissure surface caries and smooth surface caries in the permanent dentition (defended Oct. 1, 2012)
2012	Praewpannarai Buddadhumaruk	MS	Case-case genome-wide association study of meiosis nondisjunction in trisomy 21 (defended Nov. 30, 2012)
2012-2013	Claire Harwood	MS	Evaluation of the impact of a web-based educational tool on awareness of newborn screening (defended April 2, 2013)
2012-2013	Siddhartha Das	MS	Association of DNA repair gene variants with pancreatic cancer in patients with chronic pancreatitis (defended July 11, 2013)
2013-2014	Katherine Geroe	MS	Assessing health beliefs of women diagnosed and at-risk for Fabry Disease (defended April 11, 2014)
2013-2014	Tracy Dawson	MS	Pain and quality of life in patients with Ehlers-Danlos syndrome and mixed connective tissue disorders (defended April 11, 2014)
2013-2014	Amanda Matchette, <i>née</i> Brooker	MS	Assessing the practices of genetic counselors regarding head circumference measurements in hereditary cancer assessment (defended May 19, 2014)
2014	Katherine Rice	MS	A comprehensive examination of inflammatory bowel disease for genetic counselors (defended July 18, 2014)
2014-2015	Joshua Carpenter	MS	A connection between primary lymphedema and cystic hygromas (April 13, 2015)
2014-2015	Alicia Martinez	MS	Evaluation of genetic counseling services offered in a Huntington Disease clinic (April 1, 2015)
2014-2015	Afifa Irani	MS	Investigation of the common MELAS mutation in the Northwestern Pennsylvania Amish Community: Mutation frequency and effectiveness of an educational intervention (April 14, 2015)
2014-2015	Celeste Shelton	MS	Hereditary pancreatitis: Outcomes and Risks (April 7, 2015)
2015	Stephanie Nunziato-Virga	MS	Contribution of the candidate genes for primary lymphedema to secondary lymphedema and chronic pain in post-mastectomy patients (March 31, 2015)
2015	Eve Kalynchuk	MS	Understanding parental opinions of whole exome sequencing in the prenatal setting (April 10, 2015)
2014-2015	Anusha Vaidyanathan	MS	Treatment barriers and satisfaction of sickle-cell disease patients taking hydroxyurea (Aug. 11, 2015)
2012-2015	Deyana Lewis	PhD	Candidate Genes and Replication Studies of Dental Caries (Aug. 19, 2015)
2014-2015	Ryan Malave	MS	The association between cognitive reserve and time to conversion from normal cognition to mild cognitive impairment (Dec. 4, 2015)
2015-2016	Tricia Zion	MS	Genetic counselor's views and opinions regarding the possible implementation of prenatal exome sequencing (Mar. 29, 2016)

<b>Year</b>	<b>Student</b>	<b>Degree</b>	<b>Title of Dissertation/Essay</b>
2012-2016	Cameron Randall	PhD	Fear of Pain and Dental Care-Related Fear: Associations with the MC1R Gene (April 29, 2016)
2016-2017	Brooke Hornak	MS	Effect of standard of care changes on the no-show rate in a sickle cell clinic (April 4, 2017)
2016-2017	Emily Massiello	MS	Characterization of deletions in a cohort with cleft lip and palate (April 5, 2017)
2016-2017	Anna Zakas	MS	The impact of genetic counseling on patient engagement (April 10, 2017)
2016-2017	Samantha Wesoly	MS	From syndromes to normal variation: a candidate gene study of interorbital distances (April 14, 2017)
2017-2018	Claire Leifeste	MS	The impact of genetic counseling on patient engagement and health behavior (April 3, 2017)
2017-2018	Julia Stone	MS	Caregiver perceptions and adolescent quality of life in Duchenne Muscular Dystrophy (April 9, 2018)
2017-2018	Richard Biedrzycki	MS	Genome-wide association studies, false positives, and how we interpret them (April 5, 2018)
2017-2019	Emily Russel	PhD	Discrepant effects of CREBRF on obesity and metabolic phenotypes, and signals of selection in Samoans (June 3, 2019)
2018-2019	Samantha Afonso	MS	Universal tumor screening for Lynch Syndrome: Investigation of patient reported distress levels (April 9, 2019)
2018-2019	Rebecca Clark	MS	Barrier to uptake of genetic services in families of pediatric hypertrophic cardiomyopathy patients (April 12, 2019)
2018-2019	Charlotte Skinner	MS	Modifying the recurrence risk for nonsyndromic cleft lip with or without cleft palate based on aggregation of selected traits among unaffected family members (June 10, 2019)
2018-2019	Sindhu Gopaldaswamy	MS	Evidence of olfactory deficits in isolated clefting (March 28, 2019)
2018-2019	Rasha Alotaibi	PhD	Genetic analyses of dental anomalies and caries in multi-ethnic populations (Oct. 31, 2019)
2018-present	Jon Chernus	PhD	Genome-wide analyses of meiotic recombination phenotypes in nondisjunction of chromosome 21 and related methods (in progress)
2019	Priyanka Setty	MS	A Comparative Analysis to Predict p53 Activity Using Classification Models (July 29, 2019)
2019-2020	Caroline Bong	MS	Exploring Factors that Influence Reproductive Decision-Making in Duchenne Muscular Dystrophy Carriers (April 10, 2020)
2019-2020	Lauren Winter	MS	Pregnancy Body Mass Index and its Relationship with Maternal and Fetal Health Outcomes in Rural Appalachia (April 7, 2020)
2019-	Ahmed Elsergani	PhD	Three-dimensional Analysis of Palatal Morphology in the Unaffected Relatives of Individuals with Oro-facial Clefting (May 21, 2020)
2019-2020	Christine Drogan	MS	Alternate Genetic Services Delivery Models for Individuals with Pancreatic Cancer: An Investigation of Patient Reported Knowledge and Distress Levels (April 14, 2020)

<b>Year</b>	<b>Student</b>	<b>Degree</b>	<b>Title of Dissertation/Essay</b>
2020-2021	Revella Raker	MS	Genetic literacy and the acquisition of clinical genetics knowledge in medical students (April 9, 2021)
2020-2021	Mariele Anneling	MS	Comparison of video education and traditional genetic counseling for pancreatic cancer: Assessment of patient satisfaction and knowledge (April 5, 2021)
2021	Mark Vater	MS	Genetic Commonalities in Gynecologic Cancers Using Publicly Available Genome-Wide Association Study Summary Results: An Exploratory Meta-Analysis (August 4, 2021)
2020-	Gillian Goobie	PhD	Environmental impacts of clinical outcomes and the epigenome in patients with interstitial lung disease
2021-	Yulong Bai	PhD	TBD
2021-	Ellyn Dunbar	PhD	TBD
2021-	Haley Soller	MS	TBD

#### **Service on Comprehensive or Qualifying Examination Committees**

<b>Dates Served</b>	<b>Student</b>	<b>Type of Exam</b>
June 1, 2012	Siddhartha Das	PhD Qualifying
Aug. 22, 2012	Zhen Zeng	MS Comprehensive
Dec. 13, 2013	Cameron Randall	PhD Comprehensive
July 3, 2014	Deyana Lewis	PhD Qualifying
Jan. 22, 2015	Deyana Lewis	PhD Comprehensive
Jan. 28, 2015	Barbara Hopkins	PhD Qualifying
Oct. 13, 2015	Ryan Malave	MS Comprehensive
April 14, 2016	Ekaterina Orlova	PhD Qualifying (chair)
Feb. 16, 2017	Emily Russell	PhD Qualifying
March 29, 2017	Ekaterina Orlova	PhD Comprehensive (chair)
June 2, 2017	Kathyini Gopalakrishna	PhD Qualifying
July 10, 2017	Annie Arockiaraj	PhD Qualifying (chair)
July 26, 2017	Rasha Alotaibi	PhD Comprehensive
Sept. 12, 2017	Yuqiao Zhou	PhD Comprehensive (chair)
Sept. 29, 2017	Richard Biedrzycki	MS Comprehensive
Nov. 3, 2017	Amanda Everman	MS Comprehensive (chair)
Jan. 30, 2018	Emily Russell	PhD Comprehensive
March 13, 2018	Yunqi Li	MS Comprehensive (chair)
July, 10, 2018	Erika Dreikorn	PhD Qualifying
Dec. 7, 2018	Jon Chernus	PhD Comprehensive
Jan. 7, 2019	Dongjing Liu	PhD Qualifying (chair)
April 3, 2019	Alexandra Schmidt	PhD Qualifying
Sept. 19, 2019	Ahmed Elsergani	PhD Comprehensive
Jan. 28, 2020	Tianyu Zou	PhD Qualifying (chair)
March 12, 2020	Yulong Bai	PhD Qualifying
Jan. 12, 2021	Gillian Goobie	PhD Qualifying
Jan. 25, 2021	Ellyn Dunbar	PhD Qualifying
March 10, 2021	Yidi Qin	PhD Qualifying
May 24, 2021	Gillian Goobie	PhD Comprehensive
June 7, 2021	Ellyn Dunbar	PhD Comprehensive



<b>Dates Served</b>	<b>Student</b>	<b>Type of Exam</b>
June 9, 2021	Tianyu Zou	PhD Comprehensive

### Supervision of Post-Doctoral Students, Residents, and Fellows

<b>Year</b>	<b>Trainee</b>	<b>Position</b>
		<ul style="list-style-type: none"> <li>• Awards</li> </ul>
2013-2015	Mandy Schmella, PhD	Post-doctoral fellow (F32 training grant secondary mentor)
2015-2016	Jatinder Singh, PhD	Post-doctoral research associate (co-mentored)
2019-2021	Yuqiao Zhou, PhD, DDS	Post-doctoral research associate <ul style="list-style-type: none"> <li>• IADR Unilever Hatton Awards Competition Finalist (2020)</li> </ul>
2019-2021	Chenxing Liu, PhD	Post-doctoral research associate

### Other Teaching and Training

<b>Year</b>	<b>Teaching Activity</b>	<b>Description</b>
2010	co-mentor	rotation project student, Zhen Zeng (co-mentored with Eleanor Feingold)
2011-2014	training in genetics / supervising research	individual training grant (F31) co-mentor of Cameron Randall (PhD student, West Virginia University)
2013	mentor	mentor to Brooklyn Stanley, participating in the Training and Experimentation in Computational Biology (TECBio) program for undergraduate research experience
2013-2014	co-mentor	graduate student researcher, Jenna Carlson, <i>née</i> Colavincenzo (co-mentored with Eleanor Feingold)
2014-2016	mentor	mentor and supervisor to undergraduate computer science student Josh Rogan in developing genetics simulation software
2015	mentor	mentor to undergraduate researcher, Richard Biedrycki
2015-2016	mentor	mentor to Scott Eckert, participating in the Training and Experimentation in Computational Biology (TECBio) program for undergraduate research experience
2015-2016	mentor	mentor to Jessica Kerr, participating in the Pitt Summer EDGE program in Public and Global Health
2015-2016	co-mentor	rotation project student Annie Arockiaraj (co-mentored with Daniel Weeks)
2016-2017	mentor	mentor to Jessica LeClair, participating in the Training and Experimentation in Computational Biology (TECBio) program for undergraduate research experience
2016-2017	mentor	mentor and supervisor to undergraduate researcher, Richard Cuddy
2016	co-mentor	rotation project student Yunqi Li (co-mentored with Daniel Weeks)
2016	mentor	rotation project student Liyan Yang
2016	mentor	rotation project student Amanda Everman
2017	mentor	rotation project student Yuqiao Zhou

<b>Year</b>	<b>Teaching Activity</b>	<b>Description</b>
2017-2019	mentor	rotation project student and graduate student researcher Dongjing Liu
2018-2020	mentor	mentor to undergraduate researcher Ana Driscoll
2018-2019	mentor	rotation project student and graduate student researcher Tianyu Zou
2019	mentor	rotation project student Shenyu Sun
2019	mentor	mentor to Haley Grimason, participating in the Training and Experimentation in Computational Biology (TECBio) program for undergraduate research experience
2019-	co-mentor	research mentor to medical student Kathleen Koesarie

## Research

### Grants and Contracts

#### Principal Investigator

<b>Year</b>	<b>Grant and/or Contract Number and Title</b>	<b>Source</b>	<b>Annual Direct Costs</b>	<b>% Effort</b>
2014-2015	Online human population genetics simulator	U Pitt	\$15,000	3%
2014-2016	Genetics of bone development in children and adolescents	U Pitt / UPMC CMRF	\$25,000	-
2015-2018	R03 DE024264 Modeling childhood dental caries patterns for genomic and epigenetic analysis	NIH/NIDCR	\$150,000	30%
2017-2018	R56 DE027055 Genetic Architecture of Dental Caries of the Permanent Dentition	NIH/NIDCR	\$225,000	20%

#### Multiple Principal Investigator

<b>Year</b>	<b>Grant and/or Contract Number and Title</b>	<b>Source</b>	<b>Annual Direct Costs</b>	<b>% Effort</b>
2017-2021	X01 HG009878 Genomic Studies of Oral Health and Disease (PI: Shaffer/Feingold)	NIH / NHGRI	\$0 (\$667,040 genotyping services)	-
2017-2020	R01 DE027023 The Genetic Architecture of Human Facial Morphology (PI: Weinberg, Shaffer)	NIH/NIDCR	\$436,922	20%
2020-2025	R01 DE027023 The Genetic Architecture of Human Facial Morphology (PI: Weinberg, Shaffer, Claes, Wysoka)	NIH/NIDCR	\$575,707	10%

Year	Grant and/or Contract Number and Title	Source	Annual Direct Costs	% Effort
2020-2025	X01 HG011437 Genetics of Orofacial Clefts, Sub-types, and Subclinical Phenotypes (PI: Shaffer, Marazita)	NIH / NHGRI	0% (\$624,000 genotyping services)	-
2020-2021	R01 DE014899 Factors contributing to Oral Health Disparities in Appalachia (PIs: Marazita, McNeil, Foxman, Shaffer)	NIH / NIDCR	\$1,566,868	20%

#### Co-Investigator on Grants

Year	Grant and/or Contract Number and Title	Source	Annual Direct Costs	% Effort
2010-2012	R03 DE021425 Statistical Modeling and Genetic Epidemiology of Dental Caries in GWAS Analysis (PI: Wang/Feingold)	NIH / NIDCR	\$150,000 (\$25,000 to Shaffer)	50%
2012-2018	R01 NR013610 Epigenomics of Patient Outcomes after Aneurysmal SAH (PI: Conley)	NIH / NINR	\$454,172	30%
2014-2015	R01 DE016148 Extending the Phenotype of Nonsyndromic Orofacial Clefts Bridge Funding (PI: Marazita); <b>PI of subaward</b>	NIH / NIDCR	\$1,446,771 (\$10,000 to Shaffer)	8%
2014-2015	HHSN268201300005C OMICS in Latinos – Genetic Analysis (PI: Wier) <b>PI of subcontract</b>	NIH / NILBI	\$2,264,660 (\$25,000 to Shaffer)	20%
2014-2019	R01 DE016148 Extending the Phenotype of Nonsyndromic Orofacial Clefts (PIs: Marazita and Weinberg); <b>PI of subaward</b>	NIH / NIDCR	\$1,446,771 (\$150,000 to Shaffer)	20%
2015-2020	R01 DE014899 Factors contributing to Oral Health Disparities in Appalachia (PIs: Marazita, McNeil, Foxman); <b>PI of subaward</b>	NIH / NIDCR	\$1,566,868 (\$226,000 to Shaffer)	20%
2015-2016	R01 DE014899 Factors contributing to Oral Health Disparities in Appalachia (Supplement: Extension with Funding) (PIs: Marazita, McNeil, Foxman) <b>PI of subaward</b>	NIH / NIDCR	\$1,566,868 (\$46,000 to Shaffer)	10%
2015-2017	R21 NR015142 Epigenomics of Patient Outcomes after Traumatic Brain Injury (PI: Conley)	NIH / NINR	\$200,000	10%
2015-2016	GMKF X01 HL132363 Genomic Studies of Orofacial Cleft Birth Defects (PIs: Marazita and Feingold)	NIH common fund	\$0 (whole genome sequencing services)	-
2016-2018	R03 DE026469 Analysis of Whole Genome Sequence Data in Orofacial Clefts (PIs: Marazita and Feingold)	NIH / NIDCR	\$100,000	-

<b>Year</b>	<b>Grant and/or Contract Number and Title</b>	<b>Source</b>	<b>Annual Direct Costs</b>	<b>% Effort</b>
2018-2023	R01 CA221882 Epigenomics of Neurocognitive Function in Breast Cancer (PI: Conley)	NIH/NINR	\$290,462	10%
2021-2026	R01 NS115815 A Translational Evaluation of Sur1-Trpm4 Imaging Endophenotypes and Genetics to Direct Precision Medicine for Cerebral Edema After Traumatic Brain Injury (PI: Jha)	NIH/NNDS	\$472,560	4.5%
2022-2026	R01 DE031261 Integration of polygenic risk and facial morphometrics to decipher the genetic susceptibility of orofacial clefting (PI: Brand) <b>PI of subaward</b>	NIH/NIDCR	\$499,999	10%

#### **Invited Lectureships and Major Seminars**

<b>Date</b>	<b>Title of Presentation</b>	<b>Venue</b>
Jan. 2008	“Genetic epidemiology of bone loss in Mexican Americans”	Grand Rounds, University of Maryland School of Medicine, Baltimore, MD
Jan. 2008	“Genetic epidemiology of bone loss in Mexican Americans”	Invited Seminar, Section of Genetic Medicine, Department of Medicine, University of Chicago, Chicago, IL.
Jan. 2008	“Genetic epidemiology of bone loss in Mexican Americans”	Department of Epidemiology and Biostatistics Seminar Series, Case Western Reserve University, Cleveland, OH.
Feb. 2008	“Genetic epidemiology of bone loss in Mexican Americans”	Invited Seminar, Institute for Aging Research, Hebrew Senior Life, Harvard Medical School, Boston, MA.
Feb. 2008	“Genetic epidemiology of bone loss in Mexican Americans”	Invited Seminar, Division of Medical Genetics, Department of Medicine, University of Washington, Seattle, WA.
Feb. 2008	“Genetic epidemiology of bone loss in Mexican Americans”	Department of Human Genetics Seminar Series, University of Pittsburgh, Pittsburgh, PA
Sept. 2011	“GWAS of dental caries: Finding genes for tooth decay”	Department of Human Genetics Seminar Series, University of Pittsburgh, Pittsburgh, PA
Dec. 2011	“Genome-wide association study of dental caries: Finding genes for cavities”	Friends of the National Institute for Dental and Craniofacial Research Annual Conference, Washington, DC
July 2013	“Genome-wide association study of dental caries: Finding genes for cavities”	Training and Experimentation in Computational Biology (TECBio) Research and Career Seminar Series, University of Pittsburgh, Pittsburgh, PA

Date	Title of Presentation	Venue
Aug. 2014	“Keratins discovered in tooth cause dental defects and tooth decay”	Department of Human Genetics Seminar Series, University of Pittsburgh, Pittsburgh, PA
Nov. 2014	“Genetic epidemiology of dental caries”	Department of Oral Biology Research Seminar Series, University of Pittsburgh, Pittsburgh, PA
May 2016	“Genetic epidemiology of dental caries”	Development Skin Biology Section Seminar, NIAMS, NIH, Bethesda, MD
Aug. 2016	“Genetic epidemiology of dental caries”	Department of Cariology, Restorative Sciences and Endodontics, Special Seminar, University of Michigan, Ann Arbor, MI
April. 2017	“Oral-systemic health: the genomic connection”	Oral-Systemic Health Connection. 2 <sup>nd</sup> International Quintessence Symposium on Oral Health, Charlotte, NC
Jan. 2019	“The genetic architecture of human facial morphology”	Department of Human Genetics Seminar Series, University of Pittsburgh, Pittsburgh, PA

#### Other Research and Training Activities

Date	Position	Description of Activity
2014	consultant to Dr. Mei R. Fu, New York University, “Lymphedema risk reduction for Chinese breast cancer survivors”	Provided expertise on study design and oversight of analysis of genetic data
2014-2015	consultant to Dr. Lina Moreno-Urbe, University of Iowa, on genomic studies of malocclusion	Provided expertise on study design, training of research personnel, and oversight of analysis of genetic data
2015-2017	consultant to Dr. Nianjun Liu, University of Alabama at Birmingham 2015-2016, University of Indiana 2016-2017, on “Secondary analysis and risk prediction of dental caries and oral clefts”	Provided expertise on dental and orofacial phenotypes and study populations

## Publications

#### Peer-reviewed Articles

1. Lamb NE, Yu K, **Shaffer J**, Feingold E, Sherman SL. An association between maternal age and meiotic recombination for trisomy 21. *American Journal of Human Genetics*. 2005. 76:91-99.
2. **Shaffer JR (corresponding author)**, Kammerer CM, Reich D, McDonald G, Patterson N, Goodpaster B, Bauer DC, Li J, Newman AB, Cauley JA, Harris TB, Tyllavsky F, Ferrell RE, Zmuda JM for the Health ABC study. Genetic markers for ancestry are correlated with musculoskeletal traits in older African Americans. *Osteoporosis International*. 2007. 18:733-741.

3. **Shaffer JR (corresponding author)**, Kammerer CM, Rainwater DL, O'Leary DH, Bruder JM, Bauer RL, Mitchell BD. 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 Tissues International*. 2007. 81:430-441.
4. Miller-Butterworth CM, Kaplan JR, **Shaffer JR**, Devlin B, Manuck SB, Ferrell RE. Sequence variation in the primate dopamine transporter gene and its relationship to social dominance. *Molecular Biology and Evolution*. 2008. 25:18-28.
5. Brock GN, **Shaffer JR**, Blakesley RE, Lotz MJ, Tseng GC. Schemes for method selection in missing value imputation for expression profiles. *BMC Bioinformatics*. 2008. 9:1-12.
6. **Shaffer JR (corresponding author)**, Kammerer CM, Bruder JM, Cole SA, Dyer TD, Almasy L, MacCluer JW, Blangero J, Bauer RL, Mitchell BD. Genetic influences on bone loss in The San Antonio Family Osteoporosis Study. *Osteoporosis International*. 2008. 19:1759-1767.
7. **Shaffer JR (corresponding author)**, Kammerer CM, Bruder JM, Cole SA, Dyer TD, Almasy L, MacCluer JW, Blangero J, Bauer RL, Mitchell BD. Quantitative trait locus on chromosome 1q influences bone loss in young Mexican American adults. *Calcified Tissue International*. 2009. 84:75-84.
8. Ding H, Wu B, Lu Z, Yan J, Hou L, Hui R, Wang X, **Shaffer JR**, Wang DW. A novel loss-of-function *DDAH1* promoter polymorphism is associated with increased susceptibility to stroke and CHD. *Cardiovascular Research*. 2010. 106:1019-1021.
9. Wang X\*, **Shaffer JR\***, \*joint first authors, Weyant RJ, Cuenco KT, Crout R, McNeil DW, Marazita ML. Genes and their effects on dental caries may differ between primary and permanent dentitions. *Caries Research*. 2010. 44:277-84.
10. **Shaffer JR (corresponding author)**, Kammerer CM, Dressen AS, Bruder JM, Bauer RL, Mitchell BD. Rate of bone loss is greater in young Mexican American men than women: The San Antonio Family Osteoporosis Study. *Bone*. 2010. 47:49-54.
11. Hoppman N, McLenithan JC, McBride DJ, Shen H, Bruder J, Bauer R, **Shaffer JR**, Liu J, Streeten E, Shuldiner A, Kammerer C, Mitchell BD. A common variant in fibroblast growth factor binding protein 1 (*FGFBP1*) is associated with bone mineral density and influences gene expression in vitro. *Bone*. 2010. 47:272-80.
12. **Shaffer JR (corresponding author)**, Kammerer CM, Dorn J, Ferrell RE, Iacoviello L, Trevisan M, Donahue RP. Polymorphisms in the platelet-specific collagen receptor *GP6* are associated with risk of nonfatal myocardial infarction in Caucasians. *Nutrition, Metabolism and Cardiovascular Diseases*. 2011. 21:546-52.
13. Geller F, Feenstra B, Zhang H, **Shaffer JR**, Hansen T, Boyd HA, Nohr EA; Timpson NJ, Evans DM, Weyant RJ, Levy SM, Lathrop M, Smith GD, Murray JC, Werge T, Marazita ML, Sorensen TIA, Melbye M. Genome-Wide Association Study Identifies Four Loci Associated with Eruption of Permanent Teeth. *PLoS Genet*. 2011. 7(9): e1002275.
14. **Shaffer JR**, Wang X, Feingold E, Lee M, Begum F, Weeks DE, T. Cuenco K, Barmada MM, Wendell SK, Crosslin DR, Laurie CC, Doheny KF, Pugh EW, Zhang Q, Feenstra B, Geller F, Boyd HA, Zhang H, Melbye M, Murray JC, Weyant RJ, Crout R, McNeil DW, Levy SM, Slayton RL, Willing MC, Broffitt B, Marazita ML. Genome-wide Association Scan for Childhood Caries Implicates Novel Genes. *J Dent Res*. 2011. 90(12):1457-62.
15. Ryckman KK, Feenstra B, **Shaffer JR**, Bream ENA, Geller F, Feingold E, Weeks DE, Gadow E, Cosentino V, Saleme C, Simhan HN, Merrill D, Fong CT, Busch T, Berends SK, Comas B, Camelo JL, Boyd H, Laurie C, Crosslin D, Zhang Q, Doheny KF, Pugh E, Melbye M, Marazita M, Dagle JM, Murray JC. Replication of a Genome-Wide Association Study of Birth Weight in Preterm Neonates. *Journal of Pediatrics*. 2012. 160(1):19-24.

16. Hartz SM, Short S, Saccone NL, Culverhouse R, Chen LS, Schwantes-An T, Coon H, Han Y, Stephens SH, Sun J, Chen X, Ducci F, Dueker N, Franceschini N, Frank J, Geller F, Guðbjartsson D, Hansel NH, Jiang C, Keskitalo-Vuokko K, Liu J, Lyytikäinen L, Michel M, Rawal R, Rosenberger A, Scheet P, **Shaffer JR**, Teumer A, Thompson JR, Vink JM, Vogelzangs N, Wenzlaff A, Wheeler W, Xiangjun X, Yang B, Aggen SH, Balmforth AJ, Baumeister SE, Beaty T, Bennett S, Bergen A, Boyd H, Broms U, Campbell H, Chatterjee N, Chen J, Cheng Y, Cichon S, Couper D, Cucca F, Dick D, Foroud T, Furberg H, Giegling I, Gu F, Hall AS, Hällfors J, Han S, Hartmann AM, Hayward C, Heikkilä K, Hottenga JJ, Jousilahti P, Kaakinen M, Kittner SJ, Konte B, Korhonen T, Landi M, Laatikainen T, Leppert M, Levy SM, Mathias T, McNeil DW, Medland SE, Montgomery G, Muley T, Murray T, Nauck M, North K, Paré PD, Peltonen L, Pergadia M, Polasek O, Ramos E, Ripatti S, Risch A, Ruczinski I, Rudan I, Salomaa V, Schlessinger D, Stykárdsdóttir U, Terracciano A, Uda M, Willemsen G, Abecasis G, Barnes K, Bickeböller H, Boerwinkle E, Boomsma D, Caporaso N, Duan J, Edenberg HJ, Francks C, Gejman PV, Gelernter J, Grabe HJ, Hops H, Jarvelin M, Jorma V, Kähönen M, Kendler KS, Lehtimäki T, Levinson DF, Marazita ML, Marchini J, Melbye M, Mitchell BD, Murray JC, Nöthen M, Penninx BW, Raitakari O, Rietschel M, Rujescu D, Samani NJ, Sanders AR, Schwartz A, Shete S, Shi J, Spitz M, Stefansson D, Swan G, Thorgeirsson T, Völzke H, Wei Q, Wichmann H, Amos C, Breslau N, Cannon D, Ehringer M, Gruzca R, Hatsukami D, Heath A, Johnson EO, Kaprio J, Madden P, Martin NG, Stevens VL, Stitzel JA, Weiss RB, Kraft P, Bierut LJ. Increased genetic vulnerability to smoking at *CHRNA5* in early-onset smokers. *Archives of General Psychiatry*. 2012. 69(8):854-60.
17. Zheng X, **Shaffer JR**, McHugh CP, Laurie C, Melbye M, Murray JC, Marazita ML, Feingold E. Using Family Data as a Verification Standard to Evaluate Copy Number Variation Calling Strategies for Genetic Association Studies. *Genetic Epidemiology*. 2012. 36(3):253-62.
18. **Shaffer JR\***, Wang X\*, \*joint first authors, DeSensi RS, Wendell S, Weyant RJ, T.Cuenco K, Crout R, McNeil DW, Marazita ML. Genetic susceptibility to dental caries on pit and fissure and smooth surfaces. *Caries Research*. 2012. 46(1):38-46
19. Ding H, Chen C, **Shaffer JR**, Liu L, Xu Y, Wang X, Hui T, Wang DW. Telomere length and risk of stroke in Chinese. *Stroke*. 2012 43(3):658-63.
20. Agrawal A, Cheng Y\*, Lin P\*, **Shaffer JR\***, Sun Q\*, Taylor K\*, Yaspan B\*, \*joint second authors, Cole J, Cornelis M, DeSensi RS, Fitzpatrick A, Heiss G, Kang JH, O'Connell J, Bennett S, Bookman E, Bucholz K, Caporaso N, Crout R, Dick D, Edenberg HJ, Freedman N, Goate A, Hesselbrock V, Kittner S, Kramer J, Nurnberger JI, Qi L, Rice J, Schuckit M, van Dam RM, Boerwinkle E, Hu F, Levy S, Marazita M, Mitchell B, Pasquale L, Bierut L, for the GENEVA Consortium. Measuring Alcohol Consumption for Genomic Meta-Analyses: Opportunities and Challenges. *American Journal of Clinical Nutrition*. 2012. 95(3):539-47.
21. **Shaffer JR (corresponding author)**, Feingold E, Wang X, T.Cuenco K, Weeks DE, DeSensi RS, Polk DE, Wendell S, Weyant RJ, Crout R, McNeil DW, Marazita ML. Heritable patterns of tooth decay in the permanent dentition: Principal components and factor analyses. *BMC Oral Health*. 2012. 12(1):7.
22. **Shaffer JR (corresponding author)**, Feingold E, Marazita ML. Genome-wide association studies: prospects and challenges for oral health. *Journal of Dental Research*. 2012. 91(7):637-41.
23. Polk DE, Wang X, Feingold E, **Shaffer JR**, Weeks DE, Weyant RJ, McNeil DW, Marazita ML. Effects of Smoking and Genotype on the PSR Index of Periodontal Disease in Adults Aged 18 – 49. *International Journal of Environmental Research and Public Health*. 2012. 9(8):2839-50.
24. Alleman BW, Myking S, Ryckman KK, Myhre R, Feingold E, Feenstra B, Geller F, Boyd HA, **Shaffer JR**, Zhang Q, Begum F, Crosslin D, Doheny K, Pugh E, Pay AS, Ostensen IH, Morken NH, Magnus P, Marazita ML, Jacobsson B, Melbye M, Murray JC. No observed association for mitochondrial SNPs with preterm delivery and related outcomes. *Pediatric Research*. 2012. 72(5):539-44.

25. Wang X, **Shaffer JR**, Zeng Z, Begum F, Vieira AR, T.Cuenco K, Lee M, Beck J, Boerwinkle E, Cornelis M, Hu F, Crosslin DR, Laurie CC, Doheny KF, Pugh EW, Polk DE, Weyant RJ, Crout R, McNeil DW, Weeks DE, Feingold E, Marazita ML. Genome-wide Association Scan of Dental Caries in the Permanent Dentition. *BMC Oral Health*. 2012. 12(1):57.
26. **Shaffer JR (corresponding author)**, Feingold EF, Wang X, Weeks DE, Weyant RJ, Crout R, McNeil DW, Marazita ML. Clustering tooth surfaces into biologically-informative caries outcomes. *Journal of Dental Research*. 2013. 92(1):32-7.
27. **Shaffer JR (corresponding author)**, Feingold EF, Wang X, Lee M, T.Cuenco K, Weeks DE, Weyant RJ, Crout R, McNeil DW, Marazita ML. GWAS of dental caries patterns in the permanent dentition. *Journal of Dental Research*. 2013. 92(1):38-44.
28. Schreiber KL, Martel MO, Shnol H, **Shaffer JR**, Greco C, Taylor LC, Viray N, McLaughlin M, Brufsky A, Ahrendt G, Bovbjerg D, Edwards RR, Belfer I. Persistent Pain In Postmastectomy Patients: Comparison of Psychophysical, Medical, Surgical and Psychosocial Characteristics between Patients with and without Pain. *Pain*. 2013. 154(5):660-8.
29. Zeng Z\* (mentee), **Shaffer JR\*** (corresponding author), \* joint first authors, Wang X, Feingold E, Weeks DE, Lee M, T.Cuenco K, Wendell SK, Weyant RJ, Crout R, McNeil DW, Marazita ML. Genome-wide Association Studies of Pit-and-Fissure- and Smooth-surface Caries in Permanent Dentition. *Journal of Dental Research*. 2013. 92(5):43207.
30. **Shaffer JR (corresponding author)**, Polk DE, Feingold E, Wang X, T.Cuenco K, Weeks DE, DeSensi RS, Weyant RJ, Crout R, McNeil DW, Marazita ML. Demographic, socioeconomic, and behavioral factors affecting patterns of tooth decay in the permanent dentition: Principal components and factor analysis. *Community Dentistry and Oral Epidemiology* 2013 41(4)364-73.
31. Graff M, Ngwa JS, Workalemahu T, Homuth G, Schipf S, Teumer A, Volzke H, Wallaschofski H, Abecasis GR, Edward L, Francesco C, Sanna S, Scheet P, Schlessinger D, Sidore C, Xiao X, Wang Z, Chanock SJ, Jacobs KB, Hayes RB, Hu F, Van Dam RM, The GIANT Consortium, Crout RJ, Marazita ML, **Shaffer JR**, Atwood LD, Fox CS, Heard-Costa NL, White C, Choh AC, Czerwinski SA, Demerath ED, Dyer TD, Towne B, Amin N, Oostra BA, vanDuijn CM, Zillikens MC, Esko T, Nelis M, Nikopensius T, Metspalu A, Strachan DP, Monda K, Qi L, North KE, Cupples LA, Gordon-Larsen P, Berndt SI. 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(17):3597-607.
32. Belfer I, Schreiber KL, **Shaffer JR**, Shnol H, Blaney K, Morando A, Englert D, Greco C, Brufsky A, Ahrendt G, Kehlet H, Edwards RR, Bovbjerg DH. Persistent Post-Mastectomy Pain in Breast Cancer Survivors: Analysis of Clinical, Demographic and Psychosocial Factors. *The Journal of Pain* 2013 14(10):1185-1195.
33. S.H. Stephens, S.M. Hartz, N.R. Hoft, N.L. Saccone, R.C. Corley, J.K. Hewitt, C.J. Hopfer, N. Breslau, H. Coon, X. Chen, F. Ducci, N. Dueker, N. Franceschini, J. Frank, Y. Han, N.N. Hansel, C. Jiang, T. Korhonen, P.A. Lind, J. Liu, M. Michel, L-P. Lyytikäinen, **J.R. Shaffer**, S.E. Short, J. Sun, A. Teumer, J.R. Thompson, N. Vogelzangs, J.M. Vink, A. Wenzlaff, W. Wheeler, B-Z. Yang, S.H. Aggen, A.J. Balmforth, S. Baumesiter, T.H. Beaty, D.J. Benjamin, A.W. Bergen, U. Broms, D. Cesarini, N. Chatterjee, J. Chen, Y-C. Cheng, S. Cichon, D. Couper, F. Cucca, D. Dick, T. Foround, H. Furberg, I. Giegling, N.A. Gillespie, F. Gu, A.S. Hall, J. Hällfors, S. Han, A.M. Hartmann, K. Heikkilä, I.B. Hickie, J.J. Hottenga, P. Jousilahti, M. Kaakinen, M. Kähönen, P.D. Koellinger, S. Kittner, B. Konte, M-T. Landi, T. Laatikainen, M. Leppert, S.M. Levy, R.A. Mathias, D.W. McNeil, S.E. Medlund, G.W. Montgomery, T. Murray, M. Nauck, K.E. North, P.D. Paré, M. Pergadia, I. Ruczinski, V. Salomaa, J. Viikari, G. Willemsen, K.C. Barnes, E. Boerwinkle, D.I. Boomsma, N. Caporaso, H.J. Edenberg, C. Francks, J. Gelernter, H.J. Grabe, H. Hops, M-R. Jarvelin, M. Johannesson, K.S. Kendler, T. Lehtimäki, P.K.E. Magnusson, M.L. Marazita, J. Marchini, B.D. Mitchell, M.M. Nöthen, B.W.



- Penninx, O, Raitakari, M, Rietschel, D, Rujescu, N.J, Samani, A.G, Schwartz, S, Shete, M, Spitz, G.E, Swan, H, Völzke, J, Veijola, Q, Wei, C, Amos, D.S, Canon, R, Grucza, D, Hatsukami, A, Heath, E.O, Johnson, J, Kaprio, P, Madden, N.G, Martin, V.L, Stevens, R.B, Weiss, P, Kraft, L.J, Bierut, M.A, Ehringer. Distinct Loci in the *CHRNA5/CHRNA3/CHRN4* Gene Cluster are Associated with Onset of Regular Smoking. *Genetic Epidemiology*. 2013 37(8):846-859.
34. Zheng X, Feingold E, Ryckman KK, **Shaffer JR**, Boyd HA, Feenstra B, Melbye M, Marazita ML, Murray JC, T.Cuenco K. Association of maternal CNVs in *GSTT1/GSTT2* with smoking, preterm delivery and low birth weight. *Frontiers in Genetics: Statistical Genetics and Methodology* 2013 4:196.
  35. **Shaffer JR (corresponding author)**, Polk DE, Wang X, Feingold E, Weeks DE, Lee M, T.Cuenco K, Weyant RJ, Crout RJ, McNeil DW, Marazita ML. GWAS of Periodontal Health Measured by Probing Depth in Adults Aged 18-49 years. *Genes | Genomes | Genetics* 2014 4:307-314.
  36. Zeng Z (mentee), Feingold E, Wang X, Weeks DE, Lee M, T.Cuenco K, Broffitt B, Weyant RJ, Crout RJ, McNeil DW, Levy SM, Marazita ML, **Shaffer JR (corresponding author)**. Genome-wide association study of primary dentition pit-and-fissure and smooth surface caries. *Caries Research* 2014 48:330-338.
  37. Stanley BOC (mentee), Feingold E, Cooper M, Vayukov MM, Maher BS, Slayton RL, Willing MC, Rein SE, McNeil DW, Crout RJ, Weyant RJ, Levy SM, Vieira AR, Marazita ML, **Shaffer JR (corresponding author)**. Genetic association of *MPPED2* and *ACTN2* with dental caries. *Journal of Dental Research* 2014 93(7):626-632.
  38. Duverger O, Ohara T, **Shaffer JR**, Donahue D, Dullnig A, Crecelius C, Beniash E, Marazita ML, Morasso MI. Epithelial hair keratins in enamel: novel susceptibility loci for human tooth decay. *Journal of Clinical Investigation* 2014 124(12):5219-24.
  39. **Shaffer JR (corresponding author)**, Carlson J, Stanley BOC, Feingold E, Cooper M, Vanyukov MM, Maher BS, Slayton RL, Willing MC, Rein SE, McNeil DW, Crout RJ, Weyant RJ, Levy SM, Vieira AR, Marazita ML. Effects of enamel matrix genes on dental caries are moderated by fluoride exposures. *Human Genetics* 2015 134(2):159-167.
  40. **Shaffer JR (corresponding author)**, Wang X, McNeil DW, Weyant RJ, Crout RJ, Marazita ML. Genetic susceptibility to dental caries differs between the sexes: A family-based study. *Caries Research* 2015 49(2):133-140.
  41. Wolf ZT\*, Brand HA\*, **Shaffer JR\***, \*joint first authors, Leslie EJ, Willet C, McHenry T, Ariz B, Narayan N, Feingold E, Wang X, Sliskovic S, Karmi N, Safra N, Sanchez C, Deleyiannis FWB, Murray JC, Wade CM, Marazita ML, Bannasch DL. Genetic studies in human and canines implicate *ADAMTS20* as a novel risk gene for nonsyndromic cleft lip and palate. *PLoS Genetics* 2015 11(3):e1005059.
  42. Neiswanger K, McNeil DW, Foxman B, Govil M, Cooper ME, Weyant RJ, **Shaffer JR**, Crout RJ, Simhan HN, Beach SR, Chapman S, Zovko JG, Brown LJ, Strotmeyer SJ, Maurer JL, Marazita ML. Oral Health in a Sample of Pregnant Women from Northern Appalachia (2011-2015). *Int J Dent* 2015 469376:1-12.
  43. **Shaffer JR (corresponding author)**, Leslie EJ, Feingold EF, Govil M, McNeil DW, Crout RJ, Weyant RJ, Marazita ML. Caries experience differs between females and males across age groups in Northern Appalachia. *Int J Dent* 2015 938213:1-8.
  44. Shungin D, Cornelis M, Divaris K, Holtfreter B, **Shaffer JR**, Yu Y, Barros SP, Beck JD, Biffar R, Boerwinkle EA, Crout RJ, Ganna A, Hallmans G, Hindy G, Hu FB, McNeil DW, Melander S, Moss KL, North KE, Orho-Melander M, Pedersen NJ, Ridker PM, Rose LM, Rukh G, Teumer A, Weyant RJ, Chasman DI, Joshipura K, Kocher T, Magnusson PKE, Marazita ML, Nilsson P, Offenbacher S, Smith GD, Lundberg P, Palmer TM, Timpson NJ, Johansson I, Franks PW. Is Body Mass Index

- Causally Related with Periodontitis? Results from the Gene-Lifestyle Interactions and Dental Endpoints (GLIDE) Consortium. *Int J Epidemiol* 2015 44(2):638-50.
45. McNeil DW, Hayes S, Randall CL, Polk DE, Neiswanger K, **Shaffer JR**, Weyant RJ, Foxman B, Crout RJ, Chapman S, Brown LJ, Maurer JL, Marazita ML. Depression and a rural environment are associated with poor oral health among pregnant women in Northern Appalachia. *Behav Modif* 2016 40(1-2):325-40.
  46. Conomos MP, Laurie CA, Stilp AM, Gogarten SM, McHugh CP, Nelson SC, Sofer T, Fernández-Rhodes L, Justice AE, Graff M, Young KL, Seyerle AA, Avery CL, Taylor KD, Rotter JI, Talavera GA, Daviglus ML, Wassertheil-Smoller S, Schneiderman N, Heiss G, Kaplan RC, Franceschini N, Reiner AP, **Shaffer JR**, Barr RG, Kerr KF, Browning SR, Browning BL, Weir BS, Avilés-Santa ML, Papanicolaou GJ, Lumley T, Szpiro AA, North KE, Rice K, Thornton TA, Laurie CC. Genetic Diversity and Association Studies in U.S. Hispanic/Latino Populations: Applications in the Hispanic Community Health Study/Study of Latinos. *Am J Hum Genet* 2016 98(1):165-84.
  47. Morrison J, Laurie CC, Marazita ML, Sander AE, Offenbacher S, Salazar CR, Conomos M, Thornton T, Dain D, Laurie CA, Kerr K, Papanicolaou G, Taylor K, Kaste L, Finlayson TL, Beck J, **Shaffer JR (corresponding author)**. Genome-wide association study of dental caries in the Hispanic Communities Health Study / Study of Latinos. *Human Molecular Genetics* 2016 Feb 15;25(4):807-16.
  48. Leslie EJ, Liu H, Carlson JC, **Shaffer JR**, Feingold E, Wehby G, Laurie CA, Jain D, Laurie CC, Doheny K, McHenry T, Resick J, Sanchez C, Jacobs J, Emanuele B, Neiswanger K, Czeizel AE, Deleyiannis F, Christensen K, Munger R, Lie RT, Willcox A, Romitti P, Field LL, Padilla C, Lidral AC, Valencia-Ramirez LC, Lopez-Palacio AM, Palacio DR, Arcos-Burgos M, Castilla EE, Poletta FA, Orioli IM, Carvalho FM, Hecht JT, Blanton SH, Buxó CJ, Butali A, Mossey PA, Adeyemo WL, James O, Braimah RO, Aregbesola BS, Eshete MA, Deribew M, Vieira AR, Koruyucu M, Seymen F, Ma L, de Salamanca JE, Moreno L, Weinberg S, Cornell RA, Murray JC, Marazita ML. A genome-wide association study of nonsyndromic cleft palate identifies an etiologic missense variant in *GRHL3*. *Am J Hum Genet* 2016 Apr 7;98(4):744-54.
  49. Leslie EJ\*, Carlson JC\*, **Shaffer JR\***, **\*joint first authors**, Feingold E, Wehby G, Laurie CA, Jain D, Laurie CC, Doheny K, McHenry T, Resick J, Sanchez C, Jacobs J, Emanuele B, Neiswanger K, Lidral AC, Valencia-Ramirez LC, Lopez-Palacio AM, Palacio DR, Arcos-Burgos M, Czeizel AE, Field LL, Padilla C, Deleyiannis F, Christensen K, Munger R, Lie RT, Willcox A, Romitti P, Castilla EE, Poletta FA, Orioli IM, Carvalho FM, Hecht JT, Blanton SH, Buxó CJ, Butali A, Mossey PA, Adeyemo WL, James O, Braimah RO, Aregbesola BS, Eshete MA, Deribew M, Vieira AR, Koruyucu M, Seymen F, Ma L, de Salamanca JE, Moreno L, Weinberg S, Murray JC, Marazita ML. A multi-ethnic genome-wide association study identifies novel loci for nonsyndromic cleft lip with or without cleft palate on 2p24.2, 17q23 and 19q31. *Human Molecular Genetics* 2016 Jul 1;25(13):2862-2872.
  50. Sofer T, **Shaffer JR**, Graff M, Qi Q, Stilp AM, Gogarten SM, North KE, Isasi CR, Weir BS, Laurie CC, Szpiro AA. Meta-analysis of genome-wide association studies with correlated individuals: application to the Hispanic Community Health Study / Study of Latinos (HCHS/SOL). *Genetic Epidemiology* 2016 Sep;40(6):492-501..
  51. Randall CL, McNeil DW, **Shaffer JR**, Crout RJ, Weyant RJ, Marazita ML. Fear of pain mediates the association between MC1R genotype and dental fear. *J Dent Res* 2016 Sep;95(10):1132-7.
  52. **Shaffer JR**, Orlova E (mentee), Lee MK, Leslie EJ, Raffensperger ZD, Heike CL, Cunningham ML, Hecht JT, Kau CH, Nidey NL, Moreno LM, Wehby GL, Murray JC, Laurie CA, Laurie CC, Cole J, Ferrara T, Santorico S, Klein O, Mio W, Feingold E, Hallgrimsson B, Spritz RA, Marazita ML, Weinberg SE. Genome-wide association study reveals multiple loci influencing normal human facial morphology. *PLoS Genetics* 2016 Aug 25;12(8):e1006149.

53. Lewis DD (mentee), **Shaffer JR (corresponding author)**, Feingold E, Cooper M, Vanyukov MM, Maher BS, Slayton RL, Willing MC, Reis SE, McNeil DW, Crout RJ, Weyant RJ, Levy SM, Vieira AR, Marazita ML. Genetic association of MMP10, MMP14 and MMP16 with dental caries. *International Journal of Dentistry* 2017 2017:8465125:1-7.
54. Sanders AE, Wong Q, Sofer T, Agler C, **Shaffer JR**, Beck JD, Offenbacher S, Atkinson J, Salazar CR, North KE, Marazita ML, Laurie CC, Divaris K. Chronic periodontitis genome-wide association study among Hispanics/Latinos. *Journal of Dental Research* 2017 Jan;96(1):64-72..
55. Randall CL, **Shaffer JR (corresponding author)**, McNeil DW, Crout RJ, Weyant RJ, Marazita ML. Toward a Genetic Understanding of Dental Fear: Evidence of Heritability. *Community Dentistry and Oral Epidemiology* 2017 Feb;45(1):66-73.
56. Sanders AE, Jain D, Sofer T, Kerr KF, Laurie CL, **Shaffer JR**, Marazita ML, Kaste LM, Slade GD, Fillingim RB, Ohrbach R, Maixner W, Kocher T, Bernhardt O, Teumer A, Schwahn C, Sipilä K, Lähdesmäki R Mannikko M, Pesonen P, Järvelin MR, Rizzatti-Barbosa CM, Meloto CB, Serrano P, Ribeiro-Dasilva M, Diatchenko L, Smith SB. GWAS Identifies New Loci for Painful Temporomandibular Disorder. *Journal of Dental Research* 2017 Mar;96(3):277-284.
57. Eckert S (mentee), Feingold E, Cooper M, Vanyukov MM, Maher BS, Slayton RL, Willing MC, Reis SE, McNeil DW, Crout RJ, Weyant RJ, Levy SM, Vieira AR, Marazita ML, **Shaffer JR (corresponding author)**. Variants on chromosome 4q21 near PKD2 and SIBLINGs are associated with dental caries. *Journal of Human Genetics* 2017 Apr;62(4):491-496.
58. Wright CD, Edwards CB, McNeil DW, Crout RJ, Neiswanger K, Weyant RJ, Polk DE, **Shaffer JR**, Foxman B, Marazita ML. Periodontal Disease and Quality of Life: Impact of Fear of Pain and Dental Fear. *Pain Research and Management* 2017 2017:5491923.
59. Leslie EJ, Carlson JC, **Shaffer JR**, Butali A, Buxo CJ, Castilla EE, Christensen K, Keleyiannis FWB, Field LL, Hecht JT, Moreno L, Orioli IM, Padilla C, Vieira AR, Wehby GL, Feingold E, Weinberg SM, Murray JC, Beaty TH, Marazita ML. 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 Mar;136(3):275-286.
60. Leslie EJ, Carlson JC, **Shaffer JR**, Buxo CJ, Castilla EE, Christensen K, Deleyiannis FWB, Field LL, Hecht JT, Moreno L, Orioli IM, Padilla C, Vieira AR, Wehby GL, Feingold E, Weinberg SM, Murray JC, Marazita ML. Association studies of low-frequency coding variants in nonsyndromic cleft lip with or without cleft palate. *American Journal of Medical Genetics Part A* 173(6):1531-1538.
61. Lee MK\*, **Shaffer JR\* (corresponding author), joint first authors**, Leslie EJ, Orlova E, Carlson JC, Feingold E, Marazita ML, , Weinberg SM. Genome-wide association study of facial morphology reveals novel associations with *PARK2* and *FREM1*. *PLoS ONE* 2017 Apr 25;12(4):e0176566.
62. Randall CL, Wright CD, Chernus J, McNeil DW, Feingold E, Crout RJ, Neiswanger K, Weyant RJ, **Shaffer JR (corresponding author)**, Marazita ML. A preliminary genome-wide association study of fear of pain: Implications for orofacial pain. *Pain Research and Management*, 2017, 2017:7375468:1-12.
63. Ballantine J, Carlson JC, Zandona A, Agler CS, Zeldin LP, Rozier G, Roberts MW, Basta PV, Luo J, Antonio-Obese ME, McNeil D, Weyant R, Crout RJ, Slayton R, Levy S, **Shaffer JR**, Marazita ML, North KE, Divaris K. Exploring the genomic basis of early childhood caries: a pilot study. *Int J Paed Dentistry*. 2018 Mar;28(2):217-225.
64. Jenna C. Carlson, Jennifer Standley, Aline Petrin, **John R. Shaffer**, Azeez Butali, Carmen J. Buxo, Kaare Christensen, Frederic W-D Deleyiannis, Jacqueline T. Hecht, L. Leigh Field, Lina M. Moreno, Ieda M. Orioli, Carmencita Padilla, Satoshi Suzuki, Alexandre R. Vieira, Seth M. Weinberg, George L. Wehby, Eleanor Feingold, Jeffrey C. Murray, Mary L. Marazita, Elizabeth J. Leslie. Identification of 16q21 as a modifier of nonsyndromic orofacial cleft phenotypes. *Genetic Epidemiology* 2017 Dec;41(8):887-897.

65. **John R. Shaffer**, Jinxi Li, Myoung Keun Lee, Jasmien Roosenboom, Ekaterina Orlova (mentee), Kaustabh Adhikari, 23andMe Research Team, Carla Gallo, Giovanni Poletti, Lavinia Schuler-Faccini, Maria-Cátira Bortolini, Samuel Canizales-Quinteros, Francisco Rothhammer, Gabriel Bedoya, Rolando González-José, Paige E. Pfeffer, Christopher A. Wollenschlaeger, Jacqueline T. Hecht, George L. Wehby, Lina M. Moreno, Anan Ding, Li Jin, Yajun Yang, Jenna C. Carlson, Elizabeth J. Leslie, Eleanor Feingold, Mary L. Marazita, David Hinds, Timothy C. Cox, Sijia Wang, Andrés Ruiz-Linares, Seth M. Weinberg. Multiethnic GWAS reveals polygenic architecture of earlobe attachment. *Am J Hum Genet* 2017 Dec 7;101(6):913-924.
66. Peter Claes, Jasmien Roosenboom, Julie White, Tomek Swigut, Dzemila Sero, Jiarui Li, Myoung Keun Lee, Arslan Zaidi, Brooke Mattern, Corey Liebowitz, Tomás González, Elizabeth J. Leslie, Jenna C. Carlson, Ekaterina Orlova (mentee), Paul Suetens, Dirk Vandermeulen, Eleanor Feingold, Mary L. Marazita, **John R. Shaffer**, Joanna Wysocka, Mark Shriver, Seth M. Weinberg. Genome-wide mapping of global-to-local genetic effects on human facial shape. *Nat Genet* 2018 50(3):414-423.
67. Olivier Duverger, Jenna C. Carlson, CM Karacz, M Schwartz, Mary L. Marazita, **John R. Shaffer**, and Maria Morasso. Genetic variants in pachyonychia congenita-associated keratins increase susceptibility to tooth decay. *PLOS Genetics* 2018 Jan 22;14(1):e1007168.
68. Jonathan Chernus, Jasmien Roosenboom, Matthew Ford, Myoung Keun Lee, Beth Emanuele, Joel Anderton, Jacqueline T. Hecht, Carmencita Padilla, Frederic W.B. Deleyiannis, Carmen J. Buxo, Eleanor Feingold, Elizabeth J. Leslie, **John R Shaffer**, Seth M. Weinberg, Mary L. Marazita. GWAS reveals loci associated with velopharyngeal dysfunction. *Scientific Reports*. 2018 May 31;8(1):8470.
69. Govil M, Mukhopadhyay N, Weeks DE, Feingold E, **Shaffer JR**, Levy SM, Vieira AR, Slayton RL, McNeil DW, Weyant RJ, Crout RJ, Marazita ML. Novel caries loci in children and adults implicated by genome-wide linkage. *BMC Oral Health* 2018 Jun 1;18(1):98.
70. Simon Haworth, Dmitry Shungin, Justin T van der Tas, Strahinja Vucic, Carolina Medina-Gomez, Victor Yakimov, Bjarke Feenstra, **John R Shaffer**, Myoung Keun Lee, Marie Standl, Elisabeth Thiering, Carol Wang, Klaus Bønnelykke, Johannes Waage, Leon Eyrych Jessen, Pia Elisabeth Nørrisgaard, Raimo Joro, Ilkka Seppälä, Olli Raitakari, Tom Dudding, Olja Grgic, Edwin Ongkosuwito, Anu Vierola, Aino-Maija Eloranta, Nicola X West, Steven J Thomas, Daniel W McNeil, Steven M Levy, Rebecca Slayton, Ellen A Nohr, Terho Lehtimäki, Timo Lakka, Hans Bisgaard, Craig Pennell, Jan Kühnisch, Mary L Marazita, Mads Melbye, Frank Geller, Fernando Rivadeneira, Eppo B Wolvius, Paul W Franks, Ingegerd Johansson, Nicholas J Timpson. Consortium genome-wide meta-analysis for childhood dental caries traits. *Human Molecular Genetics*. 2018 Sep 1;27(17):3113-3127.
71. Laurence J. Howe, Myoung Keun Lee, Gemma C. Sharp, George Davey Smith, Beate St Pourcain, **John R. Shaffer**, Kerstin U. Ludwig, Elisabeth Mangold, Mary L. Marazita, Eleanor Feingold, Alexei Zhurov, Evie Stergiakouli, Jonathan Sandy, Stephen Richmond, Seth M Weinberg, Gibran Hemani, Sarah J. Lewis. Investigating the shared genetics of non-syndromic cleft lip/palate and facial morphology. *PLOS Genetics* 2018 Aug 1;14(8):e1007501.
72. Jasmien Roosenboom, Myoung Keun Lee, Jacqueline T. Hecht, Carrie L. Heike, George L. Wehby, Kaare Christensen, Eleanor Feingold, Mary L. Marazita, A. Murat Maga, **John R. Shaffer**, Seth M. Weinberg. Mapping genetic variants for cranial vault shape in humans. *PLOS One*. 13(4): e0196148.
73. Jasmien Roosenboom, Karlijne Indencleef, Myoung Keun Lee, Hanne Hoskens, Julie D. White, Dongjing Liu, Jacqueline T. Hecht, George L. Wehby, Lina M. Moreno, Carolyn Hodges-Simeon, Eleanor Feingold, Mary L. Marazita, Stephen Richmond, Mark D. Shriver, Peter Claes, **John R.**

- Shaffer**, Seth M. Weinberg. SNPs associated with testosterone levels influence human facial morphology. *Frontiers in Genetics* 2018 Oct 23;9:497.
74. Karlijne Indencleef, Jasmien Roosenboom, Hanne Hoskens, Julie D. White, Mark D. Shriver, Stephen Richmond, Hilde Peeters, Eleanor Feingold, Mary L. Marazita, **John R. Shaffer**, Seth M. Weinberg, Greet Hens, Peter Claes. Six NSCL/P loci show associations with normal-range craniofacial variation. *Frontiers in Genetics* 2018 Oct 25;9:502.
  75. Umut Özbek, Hui-Min Lin, Yan Lin, Daniel E. Weeks, Wei Chen, **John R. Shaffer**, Shaun M. Purcell, Eleanor Feingold. Statistics for X-chromosome Associations. *Genetic Epidemiology* 2018 Sep;42(6):539-550.
  76. Ekaterina Orlova (mentee), Andrew Yeh, Min Shi, Brian Firek, Sarangarajan Ranganathan, 23andMe Research Team, David C. Whitcomb, David N. Finegold, Robert E. Ferrell, M. Michael Barmada, Mary L. Marazita, David A. Hinds, **John R. Shaffer (corresponding author)**, Michael J. Morowitz. Genetic association and differential expression of PITX2 with acute appendicitis. *Human Genetics* 2019 138(1), 37-47
  77. **John R. Shaffer (corresponding author)**, Jessica LeClair (mentee), Jenna C. Carlson, Eleanor Feingold, Carmen J. Buxó, Kaare Christensen, Frederic W.B. Deleyiannis, L. Leigh Field, Jacqueline T. Hecht, Lina Moreno, Ieda M. Orioli, Carmencita Padilla, Alexandre R. Vieira, George L. Wehby, Jeffrey C. Murray, Seth M. Weinberg, Mary L. Marazita, Elizabeth J. Leslie. Association of Low-Frequency Genetic Variants in Regulatory Regions with Non-Syndromic Orofacial Clefts. *American Journal of Medical Genetics Part A* 2019 Mar;179(3):467-474.
  78. Tamar Sofer, Xiuwen Zheng, Stephanie M. Gogarten, Cecelia A. Laurie, Kelsey Grinde, **John R. Shaffer**, Dmitry Shungin, Jeffrey R. O'Connell, Ramon A. Durazo-Arviso, Laura Raffield, Leslie Lange, Solomon Musani, Ramachandran S. Vasan, L. Adrienne Cupples, Alexander P. Reiner, Cathy C. Laurie, Kenneth M. Rice. A Fully-Adjusted Two-Stage Procedure for Rank Normalization in Genetic Association Studies. *Genetic Epidemiology* 2019 Apr;43(3):263-275.
  79. Seth Weinberg, Jasmien Roosenboom, **John R. Shaffer**, Mark D. Shriver, Joanna Wysocka, Peter Claes. Hunting for genes that shape human faces: initial successes and challenges for the future. *Orthodontics & Craniofacial Research* 2019; 22(Suppl. 1):207–212.
  80. Mengxia Chen, Casey D. Wright, Oluwabunmi Tokede, Alfa Yansane, Alexander Montasem, E. Kalenderian, Terri Beaty, Eleanor Feingold, Richard J. Crout, Katherine Neiswanger, **John R. Shaffer**, Robert J. Weyant, Mary L. Marazita, Daniel W. McNeil. Predictors of Dental Care Utilization in North Central Appalachia in the USA. *Community Dent Oral Epidemiol* 2019 Aug;47(4):283-290.
  81. Agler CS, Shungin D, Ferreira Zandoná AG, Basta PV, Luo J, Cantrell J, Pahel TD, Meyer B, **Shaffer JR**, Schäefer AS, North KE, Divaris K. Protocols, methods and tools for genome-wide association studies (GWAS) of dental traits. *Methods in Molecular Biology* 2019; 1922:493-509.
  82. Jenna C. Carlson, Deepti Anand, Carmen J. Buxo, Kaare Christensen, Fred Deleyiannis, Jacqueline T. Hecht, Lina M. Moreno, Ieda M. Orioli, Carmencita Padilla, **John R. Shaffer**, Alexandre R. Vieira, George L. Wehby, Seth M. Weinberg, Jeffrey C. Murray, Terri H. Beaty, Irfan Saadi, Salil A. Lachke, Mary L. Marazita, Eleanor Feingold, Elizabeth J. Leslie. A systematic genetic analysis and visualization of phenotypic heterogeneity among orofacial cleft GWAS signals. *Genetic Epidemiology* 2019 Sep;43(6):704-716.
  83. Dmitry Shungin, Simon Haworth, Kimon Divaris, Yoichiro Kamatani, Myoung Keun Lee, Kelsey Grinde, George Hindy, Viivi Alaraudanjoki, Paula Pesonen, Alexander Teumer, Birte Holtfreter, Saori Sakaue, Jun Hirata, Franco Giulianini, Daniel I Chasman, Paul M Ridker, Yau-Hua Yu, Patrik Magnusson, Takeaki Sudo, Yukinori Okada, Thomas Kocher, Vuokko Anttonen, Marja-Liisa Laitala, Marju Orho-Melander, **John R. Shaffer**, Mary Marazita, Michiaki Kubo, Yasushi Furuichi, Steve Offenbacher, Cary Agler, Paul W. Franks, Nicholas Timpson, Ingegerd Johansson.

- Understanding dental diseases through combined genome-wide analyses of clinical and self-reported data. *Nature Communications* 2019 Jun 24;10(1):2773.
84. Anna L. Zakas, Claire Leifeste, Beth Dudley, Eve Karloski, Samantha Afonso, Robin E. Grubs, **John R. Shaffer**, Andrea L. Durst, Michael D. Parkinson, Randall Brand. The Impact of Genetic Counseling on Patient Engagement in a Specialty Cancer Clinic. *Journal of Genetic Counseling*. 2019 Oct;28(5):9740981.
  85. Ekaterina Orlova (mentee), Jenna C. Carlson, Myoung Keun Lee, Eleanor Feingold, Daniel W. McNeil, Richard J. Crout, Robert J. Weyant, Mary L. Marazita, **John R. Shaffer (corresponding author)**. Pilot GWAS of Dental Caries in African-Americans shows Genetic Heterogeneity. *BMC Oral Health* 2019 Sep 18;19(1):215.
  86. Yueyao Wang, Dipankar Bandyopadhyay, **John R. Shaffer**, Xiaowei Wu. Gene-based association mapping for dental caries in the GENEVA program. *Journal of Dentistry and Dental Medicine* 2020; 3(4):156.
  87. Xiong Z, Dankova G, Howe LJ, Lee MK, Hysi PG, de Jong MA, Zhu G, Adhikari K, Li D, Li Y, Pan B, Feingold E, Marazita ML, **Shaffer JR**, McAloney K, Xu SH, Jin L, Wang S, de Vrij FM, Lendemeijer B, Richmond S, Zhurov A, Lewis S, Sharp GC, Paternoster L, Thompson H, Gonzalez-Jose R, Bortolini MC, Canizales-Quinteros S, Gallo C, Poletti G, Bedoya G, Rothhammer F, Uitterlinden AG, Ikram MA, Wolvius E, Kushner SA, Nijsten TE, Palstra RT, Boehringer S, Medland SE, Tang K, Ruiz-Linares A, Martin NG, Spector TD, Stergiakouli E, Weinberg SM, Liu F, Kayser M; International Visible Trait Genetics (VisiGen) Consortium. Novel genetic loci affecting facial shape variation in humans. *Elife* 2019 Nov 26;8. pii: e49898.
  88. Annie I. Arockiaraj (mentee), Dongjing Liu (mentee), **John R Shaffer**, Theresa A. Koleck, Elizabeth A. Crago, Daniel E. Weeks, Yvette P. Conley. Methylation Data Processing Protocol and Comparison of Blood and Cerebral Spinal Fluid Following Aneurysmal Subarachnoid Hemorrhage. *Frontiers in Genetics*. 2020 Jun 26;11:671. doi: 10.3389/fgene.2020.00671. eCollection 2020.
  89. Lacey W. Heinsberg, Annie I. Arockiaraj, Elizabeth A. Crago, Samuel M. Poloyac, Dianxu Ren, **John R. Shaffer**, Paula R. Sherwood, Susan M. Sereika, Daniel E. Weeks, Yvette P. Conley. Genetic variability and trajectories of DNA methylation support a role for HAMP in patient outcomes after aneurysmal subarachnoid hemorrhage. *Neurocritical Care* 2020 Apr;32(2):550-563.
  90. Camden P. Bay, Steven M. Levy, Kethleen F. Janz, Brian J. Smith, **John R. Shaffer**, Mary L. Marazita, Trudy L. Burns. Genome-wide Association Analysis of Longitudinal Bone Mineral Content Data from the Iowa Bone Development Study. *Journal of Clinical Densitometry*. 2021 Jan-Mar;24(1):44-54.
  91. Julie D. White, Karlijne Indencleef, Sahin Naqvi, Ryan J. Eller, Jasmien Roosenboom, Myoung Keun Lee, Jiarui Li, Jaaved Mohammed, Stephen Richmond, Ellen E. Quillen, Heather L. Norton, Eleanor Feingold, Tomek Swigut, Mary L. Marazita, Hilde Peeters, Greet Hens, **John R. Shaffer**, Joanna Wysocka, Susan Walsh, Seth M. Weinberg, Mark D. Shriver, Peter Claes. Insights into the genetic architecture of the human face. *Nature Genetics*. 2021 Jan;53(1):45-53.
  92. Dongjing Liu (mentee), Nora Alhazmi, Harold Matthews, Myoung Keun Lee, Jiarui Li, Jacqueline T. Hecht, George L. Wehby, Lina M. Moreno, Carrie L. Heike, Jasmien Roosenboom, Eleanor Feingold, Mary L. Marazita, Peter Claes, Eric C. Liao, Seth M. Weinberg, **John R. Shaffer (corresponding author)**. Impact of low-frequency Coding Variants on Human Facial Shape. *Scientific Reports*. 2021 Jan 12;11(1):748.
  93. Betty Bonfante, Pierre Faux, Nicolas Navarro, Javier Mendoza-Revilla, Morgane Dubied, Charlotte Montillot, Emma Wentworth, Lauriane Poloni, Ceferino Varón-González, Phil Jones, Ziyi Xiong, Macarena Fuentes-Guajardo, Juan Camilo Chacón-Duque, Malena Hurtado, Valeria Villegas, Vanessa Granja, Victor Acuña-Alonzo, Claudia Jaramillo, William Arias, Rodrigo Barquera Lozano,

- Paola Everardo, Mirsha Sánchez-Quinto, Jorge Gómez-Valdés, Hugo Villamil-Ramírez, Caio C. Silva de Cerqueira, Tábita Hunemeier, Virginia Ramallo, Fan Liu, Seth Weinberg, **John R. Shaffer**, Evie Stergiakouli, Laurence J. Howe, Pirro G. Hysi, Timothy D. Spector, Lavinia Schuler-Faccini, Rolando Gonzalez-José, Maria-Cátira Bortolini, Samuel Canizales-Quinteros, Carla Gallo, Giovanni Poletti, Gabriel Bedoya, Francisco Rothhammer, Christel Thauvin, Laurence Faivre, Caroline Costedoat, David Balding, Timothy Cox, Manfred Kayser, Laurence Duplomb, Binnaz Yalcin, Justin Cotney, Kaustubh Adhikari and Andrés Ruiz- Linares. Archaic Introgression impacts on the human face and VPS13B influences natural facial variation in human and mice. *Science Advances*. 2021 Feb 5;7(6):eabc6160.
94. Ahmed M. El Sergani, Stephanie Brandebura, Carmencita Padilla, Azeez Butali, Wasiu L. Adeyemo, Consuelo Valencia-Ramírez, Claudia P. Restrepo Muñeton, Lina M. Moreno, Carmen J. Buxó, Katherine Neiswanger, **John R. Shaffer**, Mary L. Marazita, Seth M. Weinberg. The Influence of Sex and Ancestry on Three-dimensional Palate Shape. *Cleft Palate Craniofacial Journal*. 2021 (in press)
  95. Yuqiao Zhou (mentee), Richard Cuddy (mentee), Daniel W. McNeil, Casey Wright, Richard J. Crout, Eleanor Feingold, Katherine Neiswanger, Mary L. Marazita, **John R. Shaffer (corresponding author)**. Oral health and related risk factors in Appalachia differ by rurality. *Community Dentistry and Oral Epidemiology* (in press)
  96. Sahin Naqvi, Yoeri Sleyp, Hanne Hoskens, Karlijne Indencleef, Jeffrey P. Spence, Rose Bruffaerts, Ahmed Radwan, Ryan J. Eller, Stephen Richmond, Mark D. Shriver, **John R. Shaffer**, Seth M. Weinberg, Susan Walsh, James Thomson, Jonathan K. Pritchard, Stefan Sunaert, Hilde Peeters, Joanna Wysocka, Peter Claes. Shared heritability of face and brain shape distinct from cognitive traits. *Nature Genetics* 2021 Jun;53(6):830-839.
  97. Karlijne Indencleef, Hanne Hoskens, Myoung Keun Lee, Julie D White, Chenxing Liu (mentee), Ryan J Eller, Sahin Naqvi, George L Wehby, Lina M Moreno Uribe, Jacqueline T. Hecht, Ross E Long, Kaare Christensen, Frederic W Deleyiannis, Susan Walsh, Mark Shriver, Stephen Richmond, Joanna Wysocka, Hilde Peeters, **John R Shaffer**, Mary L Marazita, Greet Hens, Seth M Weinberg, Peter Claes. The intersection of the genetic architectures of orofacial clefts and normal facial variation. *Frontiers in Genetics* 2021 Feb 22;12:626403.
  98. Sarah W. Curtis, Daniel Chang, Myoung Keun Lee, **John R. Shaffer**, Karlijne Indencleef, Michael P. Epstein, David J. Cutler, Jeffrey C. Murray, Eleanor Feingold, Terri H. Beaty, Peter Claes, Seth M. Weinberg, Mary L. Marazita, Jenna C. Carlson, Elizabeth J. Leslie. The PAX1 locus at 20p11 is a potential genetic modifier for bilateral cleft lip. *Human Genetics and Genomics Advances* 2021 Apr 8;2(2):100025.
  99. Olubukola O. Olatosi, Azeez A . Alade, Afolabi Oyapero, Tamara Busch, Mary Li, John Pape, Joy Olotu, Waheed Awotoye, Mohaned Hassan, Chinyere Adeleke, Wasiu L. Adeyemo, **John R. Shaffer**, Mary Mazarita, Azeez Butali. Dental caries severity and nutritional status of Nigerian pre-school children. *JDR Clinical and Translational Research*. 2021 May 20;21(1):274.
  100. Nandita Mukhopadhyay, Eleanor Feingold, Lina Moreno-Urbe, George Wehby, Luz Consuelo Valencia-Ramirez, Claudia Restrepo Muneton, Carmencita Padilla, Frederic Deleyiannis, Kaare Christensen, Fernando Poletta, Ieda Orioli, Jacqueline T. Hecht, Carmen Buxo, Azeez Butali, Wasiu Lanre Adeyemo, Alexandre Vieira, **John R Shaffer**, Jeff Murray, Seth M Weinberg, Elizabeth Leslie, Mary L Marazita. 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 Apr 9;9:621482.
  101. Hanne Hoskens, Dongjing Liu (mentee), Sahin Naqvi, Myoung Keun Lee, Ryan J Eller, Karlijne Indencleef, Julie D. White, Jiarui Li, Greet Hens, Joanna Wysocka, Susan Walsh, Stephen Richmond, Mark D. Shriver, **John R. Shaffer**, Hilde Peeters, Seth M. Weinberg, Peter Claes. 3D

- facial phenotyping by biometric sibling matching used in contemporary genomic methodologies. *PLOS Genetics* 2021 May 13;17(5):e1009528.
102. [Dongjing Liu \(mentee\)](#), [Annie I. Arockiaraj \(mentee\)](#), **John R. Shaffer**, Elizabeth A. Crago, Daniel E. Weeks, Yvette P. Conley. ANGPT1 methylation is associated with delayed cerebral ischemia after acute subarachnoid hemorrhage – An epigenome-wide association study. *Epigenetics Communications*. (in press)
  103. Olubukola O. Olatosi, Mary Li, Azeez A. Alade, Afolabi Oyapero, Tamara Busch, John Pape, Joy Olotu, Waheed Awotoye, Mohaned Hassan, Chinyere Adeleke, Wasiu L. Adeyemo, Elizabeth O. Sote, **John R. Shaffer**, Mary Marazita, Azeez Butali. Replication of GWAS significant loci in a Sub-Saharan African Cohort with Early Childhood Caries: a pilot study. *BMC Oral Health*. (in press)
  104. Amery Treble-Barna, Lacey W. Heinsberg, Ava M. Puccio, **John R. Shaffer**, David O. Okonkwo, Sue R. Beers, Daniel E. Weeks, Yvette P. Conley. Acute Brain-Derived Neurotrophic Factor DNA Methylation Trajectories in CSF and Associations with Outcomes following Severe Traumatic Brain Injury in Adults. *Neurorehabilitation and Neural Repair* (in press).
  105. Lacey W. Heinsberg, [Dongjing Liu \(mentee\)](#), **John R. Shaffer**, Daniel E. Weeks, Yvette P. Conley. Characterization of Cerebrospinal Fluid DNA Methylation Age During the Acute Recovery Period Following Aneurysmal Subarachnoid Hemorrhage. *Epigenetics Communications*. (2021) 1:2.
  106. [Dongjing Liu \(mentee\)](#), Hyo-Jeong Ban, Ahmed El Sergani, Myoung Keun Lee, Jacqueline T. Hecht, George L. Wehby, Lina M. Moreno, Carrie L. Heike, Eleanor Feingold, Mary L. Marazita, Seongwon Cha, Heather Szabo-Rogers, Seth M. Weinberg, **John R. Shaffer (corresponding author)**. PRICKLE1 x FOCAD interaction revealed by genome-wide vQTL analysis of human facial traits. *Frontiers in Genetics* *Front Genet*. 2021 Aug 9;12:674642.
  107. [Chenxing Liu \(mentee\)](#), Myoung Keun Lee, Sahin Naqvi, Hanne Hoskens, [Dongjing Liu \(mentee\)](#), Julie D. White, Karlijne Indencleef, Harold Matthews, Ryan J. Eller, Jiarui Li, Jaaved Mohammed, Tomek Swigut, Stephen Richmond, Mange Manyama, Benedikt Hallgrímsson, Richard A. Spritz, Eleanor Feingold, Mary L. Marazita, Joanna Wysocka, Susan Walsh, Mark D. Shriver, Peter Claes, Seth M. Weinberg, **John R. Shaffer (corresponding author)**. Genome Scans of Facial Features in East Africans and Cross-Population Comparisons Reveal Novel Associations. *PLoS Genet*. 2021 Aug 19;17(8):e1009695.
  108. Myoung Keun Lee, [Chenxing Liu \(mentee\)](#), Elizabeth J. Leslie, **John R. Shaffer**, Jamie L. Perry, Seth M. Weinberg. Heritability Analysis in Twins Indicates a Genetic Basis for Velopharyngeal Morphology. *Cleft Palate-Craniofacial Journal* (in press)
  109. Rasha N. Alotaibi, Brian J. Howe, Lina Moreno Uribe, Consuelo Valencia, Claudia Restrepo, Frederic W. B. Deleyiannis, Carmencita Padilla, Ieda M. Orioli, Carmen J Buxó, Jacqueline T. Hecht, George L. Wehby, Katherine Neiswanger, **John R. Shaffer**, Seth M. Weinberg, Mary L. Marazita. Multivariate GWAS of Structural Dental Anomalies and Dental Caries in a Multi-Ethnic Cohort. *Frontiers in Dental Medicine*. (in press).
  110. [Dongjing Liu \(mentee\)](#), Benjamin E. Zusman, **John R. Shaffer**, [Yunqi Li \(mentee\)](#), [Annie I. Arockiaraj \(mentee\)](#), Shuwei Liu, Daniel E. Weeks, Shashvat M. Desai, Patrick M. Kochanek, Ava M. Puccio, David O. Okonkwo, Yvette P. Conley, Ruchira M. Jha. Decreased DNA methylation of RGMA is associated with intracranial hypertension after severe traumatic brain injury: an exploratory epigenome-wide association study. *Neurocritical Care*. (in press)
  111. Jenna Carlson, **John R. Shaffer**, Fred Deleyiannis, Jacqueline T. Hecht, George L. Wehby, Kaare Christensen, Eleanor Feingold, Seth M. Weinberg, Mary L. Marazita, Elizabeth Leslie. Genome-wide interaction study implicates VGLL2 and alcohol exposure and PRL and smoking in orofacial cleft risk. *Frontiers in Cell and Developmental Biology*. (in press)
  112. Nandita Mukhopadhyay, Eleanor Feingold, Lina Moreno-Urbe, George Wehby, Luz Consuelo Valencia-Ramirez, Claudia P. Restrepo Muñeton, Carmencita Padilla, Frederic Deleyiannis, Kaare



Christensen, Fernando A. Poletta, Ieda M Orioli, Jacqueline T. Hecht, Carmen J. Buxó, Azeez Butali, Wasiu L. Adeyemo, Alexandre R. Vieira, **John R. Shaffer**, Jeffrey C. Murray, Seth M. Weinberg, Elizabeth J. Leslie, Mary L. Marazita. Genome-wide association study of multiethnic non-syndromic orofacial cleft families identifies novel loci specific to family and phenotypic subtypes. *Genetic Epidemiology*. (in press)

#### submitted

113. Lewis DD (mentee), **Shaffer JR (corresponding author)**, Feingold E, Cooper M, Vanyukov MM, Maher BS, Slayton RL, Willing MC, Reis SE, McNeil DW, Crout RJ, Weyant RJ, Levy SM, Vieira AR, Marazita, ML. Ion channel genes CACNA2D1, KCNH1, and KCNK5 are associated with dental caries (submitted).
114. Emery LS, Orlova E (mentee), Sofer T, Laurie CC, Marazita ML, Sander AE, Offenbacher S, Salazar CR, Laurie CA, Kerr KF, Beck JD, **Shaffer JR (corresponding author)**. Genome-wide association study of pit-and-fissure and smooth tooth surfaces caries in the Hispanic Communities Health Study/Study of Latinos (HCHS/SOL). (submitted).
115. Zion TC, **Shaffer JR**, Weltmer EC, Grubs RE. Genetic Counselors' Views and Perceptions Regarding the Implementation of Prenatal Exome Sequencing. (submitted).
116. Salazar CR, Emery LS, Sofer T, Laurie CL, Kerr K, Louie T, Morse DE, Badner V, Sanders AE, **Shaffer JR**, Qi Q, Kaste L, Singer RH, Kaplan RC, Lamster IB. No association of IL-1 $\beta$  polymorphisms with chronic periodontitis in the Hispanic Community Health Study/ Study of Latinos (HCHS/SOL). (submitted)
117. Leslie S. Emery, Ekaterina Orlova (mentee), Nandita Mukhopadhyay, Christian R. Salazar, Tamar Sofer, Marston Youngblood, Cathy C. Laurie, Alexandre R. Vieira, Mary L. Marazita, Ieda M. Orioli, Eduardo E. Castilla, Fred Deleyiannis, Anne E. Sanders, Steven Offenbacher, Cecelia A. Laurie, Kathleen F. Kerr, Shirley M. Beaver, James D. Beck, **John R. Shaffer (corresponding author)**. Genome-wide association study of tooth loss in the Hispanic Community Health Study/Study of Latinos (HCHS/SOL). (submitted)
118. Yuqiao Zhou (mentee), Daniel W. McNeil, Simon Haworth, Tom Dudding, Jonathan M. Chernus, Chenxing Liu (mentee), Dongjing Liu (mentee), Casey D. Wright, Jamey Brumbaugh, Cameron L. Randall, Robert J. Weyant, Richard J. Crout, Betsy Foxman, Steven Reis, Nicolas J. Timpson, Mary L. Marazita, **John R. Shaffer (corresponding author)**. Genome-wide scan of dental fear and anxiety nominates novel genes. (submitted)
119. Tiwari T, Randall CL, Rai N, Carey C, **Shaffer JR**, Finlayson T, Kaste LM, Daviglius ML, Laniado N, Singer R. Acculturation and Dental Care-Related Anxiety: An Assessment from the Hispanic Community Health Study / Study of Latinos. (submitted)
120. Kalo Sokoto, Lisa Platt, Linda Alexander, Betsy Foxman, **John R. Shaffer**, Mary L. Marazita, Daniel W. McNeil. Racism, Dental Care-Related Fear/Anxiety, and Oral Healthcare Utilization among Black/African American Women in Appalachia. (submitted)
121. Sahin Naqvi, Hanne Hoskens, Franziska Wilke, Seth M. Weinberg, **John R. Shaffer**, Susan Walsh, Mark D. Shriver, Joanna Wysocka, Peter Claes. Decoding the human face: challenges and progress in understanding the genetics of craniofacial morphology. (submitted)

#### Book Chapters

1. **Shaffer JR**, Marazita ML. Genetic and genomic studies of dental caries: insights into the promise of personalized medicine. 2015. (Sonis ST, ed.) *Genomics, personalized medicine and oral diseases*. Springer International Publishing. 117-144. doi 10.1007/978-3-319-17942-1.

2. **Shaffer JR**, Weyant RJ. Oral-systemic health: the genomic connection 2019. (Glick M, ed.) *The Oral-Systemic Health Connection, 2nd Edition*. Quintessence.

#### Published Abstracts

1. **John R. Shaffer**, Candace M. Kammerer, David L. Rainwater, David H. O'Leary, Richard L. Bauer, Braxton D. Mitchell. (2004) Relationship between bone mineral density and cardiovascular disease differs by sex. *American Society of Human Genetics, 54th Annual Meeting. Abstracts*: 511.
2. **John R. Shaffer**, Candace M. Kammerer, Jan M. Bruder, Richard L. Bauer, Braxton D. Mitchell. (2005) Five-year change in bone mineral density is heritable in Mexican Americans: The San Antonio Family Osteoporosis Study. American Society of Bone and Mineral Research, *27th Annual Meeting. Journal of Bone and Mineral Research* **20 Suppl 1**:S67.
3. Candace M. Kammerer, **John R. Shaffer**, David Reich, Douglas C. Bauer, Jing Li, Anne B. Newman, Jane A. Cauley, Tamara B. Harris, Francis Tylavsky, Joseph M. Zmuda. (2005) Genetic markers for ancestry are correlated with bone density and size in African Americans. *American Society of Bone and Mineral Research, 27th Annual Meeting. Journal of Bone and Mineral Research* **20 Suppl 1**:S338.
4. **John R. Shaffer**, Candace M. Kammerer, Jan M. Bruder, Richard L. Bauer, Braxton D. Mitchell. (2006) Sex-specific heritability of five-year change in bone mineral density in Mexican Americans: The San Antonio Family Osteoporosis Study. *American Society of Bone and Mineral Research, 28th Annual Meeting. Journal of Bone and Mineral Research* **21 Suppl 1**:S241.
5. **John R. Shaffer**, Candace M. Kammerer, Jan M. Bruder, Richard L. Bauer, Braxton D. Mitchell (2006) Assessment of genetic factors affecting bone loss: The San Antonio Family Osteoporosis Study. *University of Pittsburgh Institute on Aging, 1st Annual Meeting Abstracts*: 41.
6. **John R. Shaffer**, Candace M. Kammerer, Jan M. Bruder, Richard L. Bauer, Braxton D. Mitchell (2006) Assessment of genetic factors affecting bone loss: The San Antonio Family Osteoporosis Study. *American Public Health Association, 134th Annual Meeting Final Program*: 53.
7. Guy Brock, **John Shaffer**, Richard Blakesley, Meredith Lotz, George C. Tseng (2007) Which Missing Value Imputation Method To Use in Expression Profiles: A Comparative Study and Two Selection Schemes. *International Biometric Society Eastern North American Region 2007 Spring Meeting Program and Abstracts*: 374.
8. Guy Brock, **John Shaffer**, Richard Blakesley, Meredith Lotz, George C. Tseng (2007) Which Missing Value Imputation Method To Use in Expression Profiles: A Comparative Study and Two Selection Schemes. *American Statistical Association, Institute of Mathematical Statistics, International Biometric Society, Statistical Society of Canada 2007 Joint Statistical Meetings Abstract Book*: 362.
9. **John R. Shaffer**, Candace M. Kammerer, Jan M. Bruder, Shelley Cole, Thomas D. Dyer, Laura Almasy, Jean W. MacCluer, John Blangero, Richard L. Bauer, Braxton D. Mitchell (2007) Suggestive Evidence for Linkage of 5-Year Change in Bone Mineral Density to Chromosome 6q: The San Antonio Family Osteoporosis Study. *American Society of Human Genetics, 57th Annual Meeting. Abstracts*: 244.
10. **John R. Shaffer**, Candace M. Kammerer, Jan M. Bruder, Shelley Cole, Thomas D. Dyer, Laura Almasy, Jean W. MacCluer, John Blangero, Richard L. Bauer, Braxton D. Mitchell (2008) Genetic Locus on Chromosome 1q Influences Bone Loss in Young Mexican American Adults. *American Society of Bone and Mineral Research, 30th Annual Meeting. Journal of Bone and Mineral Research* **23 Suppl 1**:S435.

11. **John R. Shaffer**, Candace M. Kammerer, Jan M. Bruder, Richard L. Bauer, Braxton D. Mitchell (2008) Rate of Bone Loss is Greater in Young Adult Men than Women. *American Society of Bone and Mineral Research, 30th Annual Meeting. Journal of Bone and Mineral Research* **23 Suppl 1**:S316.
12. Candace Kammerer, **John Shaffer**, Jan Bruder, Shelley Cole, Thomas Dyer, Laura Almasy, Jean MacCluer, John Blangero, Richard Bauer, Braxton Mitchell (2008) Evidence that Different Genes Influence Bone Metabolism at Different Ages. *American Society of Human Genetics, 58<sup>th</sup> Annual Meeting. Abstracts*: 440.
13. **John Shaffer**, Candace Kammerer, Licia Iacoviello, Maurizio Trevisan, Robert Ferrell, Richard Donahue (2008) Association between GP6 Polymorphisms and Coronary Heart Disease in a Community-based Population: The Western New York Acute Myocardial Infarction Study. *American Society of Human Genetics, 58<sup>th</sup> Annual Meeting. Abstracts*: 386.
14. **John R. Shaffer**, Candace M. Kammerer, Jan M. Bruder, Richard L. Bauer, Braxton D. Mitchell. (2009) Different genes contribute to variation in peak bone density and bone loss. *American Society for Bone and Mineral Research 31<sup>st</sup> Annual Meeting. Journal of Bone and Mineral Research* **24 Suppl 1**:S55.
15. Laura Yerges-Armstrong, **John Shaffer**, K Fyan, J Liu, Candace Kammerer, Braxton Mitchell. (2009) Joint genetic influences on height and bone mineral density. *American Society of Human Genetics 59<sup>th</sup> Annual Meeting Abstracts*: 922W.
16. **John R. Shaffer**, Xiaojing Wang, Robert J. Weyant , Karen T. Cuenco, Richard Crout, Daniel W. McNeil, Mary L. Marazita. (2009) Evidence of age-dependent genetic effects on tooth decay: heritability estimates suggest genes influencing dental caries differ between primary and permanent dentitions. *American Society of Human Genetics 59<sup>th</sup> Annual Meeting Abstracts*: 1800T.
17. Mary L. Marazita, **John R. Shaffer**, Xiaojing Wang, Myoungkeun Lee, Ferdouse Begum, Eleanor Feingold, Daniel E. Weeks, Michael Barmada, Karen T. Cuenco, S. Wendell, D. Crosslin, C. Laurie, B. Weir, K. Doheny, E. Pugh, Robert J. Weyant, R. Crout, D. McNeil, S.M. Levy, R. Slayton, M. Willing, B. Broffitt, A. Vieira. (2010) Genome-wide association study of caries in the primary dentition. *American Association for Dental Research Annual Meeting*: 1514.
18. Heather Boyd, Bjarke Feenstra, Frank Geller, Eleanor Feingold, Kelli Ryckman, **John Shaffer**, Cathy Laurie, Qi Zhang, Kim Doheny, Elizabeth Pugh, Teri Manolio, Mary Marazita, Mads Melbye, Jeff Murray, GENEVA Consortium (2010) A genome-wide association study of spontaneous preterm delivery. *Pediatric Academic Societies' Annual Meeting*.
19. Brandon Alleman, Kelli Ryckman, Eleanor Feingold, Bjarke Feenstra, Frank Geller, Heather Boyd, **John Shaffer**, Cathy Laurie, Qi Zhang, Kim Doheny, Elizabeth Pugh, Teri Manolio, Mary Marazita, Mads Melbye, Jeff Murray, GENEVA Consortium (2010) Lack of Mitochondrial Association with Preterm Birth in Mothers and Neonates. *Pediatric Academic Societies' Annual Meeting*.
20. Kelli Ryckman, Ferdouse Begum, Bjarke Feenstra, Eleanor Feingold, Frank Geller, Heather Boyd, **John Shaffer**, Cathy Laurie, Qi Zhang, Kim Doheny, Elizabeth Pugh, Teri Manolio, Mary Marazita, Jeffery C Murray, Mads Melbye, GENEVA Consortium (2010) A Genome-Wide Association Study of Birthweight in Preterm and Term Neonates. *Pediatric Academic Societies' Annual Meeting*.
21. **John R. Shaffer**, Eleanor Feingold, Kelli Ryckman, Ferdouse Begum, Bjarke Feenstra, Frank Geller, Heather A. Boyd, Daniel E. Weeks, M. Michael Barmada, Cathy Laurie, Qi Zhang, Kim Doheny, Elizabeth Pugh, Mary L. Marazita, Mads Melbye, Jeffery C. Murray, GENEVA Consortium (2010) Intergenerational allelic interaction: Excess maternal-fetal allele sharing in the HLA genomic region is associated with preterm birth. *American Society of Human Genetics 60<sup>th</sup> Annual Meeting Abstracts*: 984.

22. Eleanor Feingold, **John R. Shaffer**, Xiaojing Wang, Myoungkeun Lee, Ferdouse Begum, Daniel E. Weeks, M. Michael Barmada, Karen T. Cuenco, S. Wendell, David Crosslin, Cathy Laurie, Kim Doheny, Elizabeth Pugh, Frank Geller, Bjarke Feenstra, Qi Zhang, Heather Boyd, Mads Melbye, Robert J. Weyant, Richard Crout, D. McNeil, S. M. Levy, R. L. Slayton, M. Willing, B. Broffitt, Mary L. Marazita (2010) Whole genome-scan of genetic determinants for dental caries in the primary dentition. *American Society of Human Genetics 60<sup>th</sup> Annual Meeting Abstracts*: 1043.
23. Myoungkeun Lee, Karen T. Cuenco, Xiaojing Wang, **John R. Shaffer**, Ferdouse Begum, Eleanor Feingold, Daniel E. Weeks, M. Michael Barmada, S. Wendell, David Crosslin, Cathy Laurie, Bruce Weir, Kim F. Doheny, Elizabeth Pugh, R. J. Weyant, R. J. Crout, D. W. McNeil, Mary L. Marazita (2010) Genome-Wide Association Study of Saliva Flow Rates from the COHRA-GENEVA Study. *American Society of Human Genetics 60<sup>th</sup> Annual Meeting Abstracts*: 1075.
24. Xiaojing Wang, **John R. Shaffer**, Eleanor Feingold, Myoungkeun Lee, Karen T. Cuenco, Ferdouse Begum, Daniel E. Weeks, M. Michael Barmada, S. Wendell, David Crosslin, Cathy Laurie, Kim Doheny, Elizabeth Pugh, R. J. Weyant, A. R. Vieira, R. J. Crout, D. W. McNeil, Mary L. Marazita (2010) Whole genome-scan of genetic determinants for dental caries in the permanent dentition. *American Society of Human Genetics 60<sup>th</sup> Annual Meeting Abstracts*: 1093.
25. Laura M. Yerges-Armstrong, **John R. Shaffer**, Jan M. Bruder, Thomas D. Dyer, M. A. Carless, J. E. Curran, HHH. Göring, John Blangero, Candace M. Kammerer, Braxton D. Mitchell (2010) The value of mapping genome-wide associations across other ethnic groups: An analysis of BMD in Mexican Americans. *American Society of Human Genetics 60<sup>th</sup> Annual Meeting Abstracts*: 1098.
26. Xiaojing Zheng, **John R. Shaffer**, C. McHugh, Cathy Laurie, Mads Melbye, Jeffery C. Murray, Mary L. Marazita, Eleanor Feingold (2010) Using family data as a verification standard to evaluate CNV calling strategies. *American Society of Human Genetics 60<sup>th</sup> Annual Meeting Abstracts*: 1842.
27. Kelli K. Ryckman, E. N. A. Bream, **John R. Shaffer**, Eleanor Feingold, S. K. Berends, E. Gadow, C. Saleme, H. N. Simhan, D. Merrill, C. T. Fong, T. Busch, Bjarke Feenstra, Frank Geller, Mads Melbye, Mary L. Marazita, J. M. Dagle, Jeffery C. Murray (2010) Replication of a Genome-Wide Association Study of Birth Weight in Preterm Neonates. *American Society of Human Genetics 60<sup>th</sup> Annual Meeting Abstracts*: 2462.
28. Bjarke Feenstra, Frank Geller, Qi Zhang, Heather A. Boyd, Kelli K. Ryckman, **John R. Shaffer**, J. M. Dagle, Daniel E. Weeks, Mary L. Marazita, Eleanor Feingold, Jeffery C. Murray, Mads Melbye (2010) Assessing the Combined Significance of SNPs in Candidate Genes for Preterm Birth. *American Society of Human Genetics 60<sup>th</sup> Annual Meeting Abstracts*: 2479.
29. H. Ling, **John R. Shaffer**, Terri H. Beaty, Mary L. Marazita, I. Ruczinski, Eleanor Feingold, Cathy C. Laurie, David Crosslin, AF. Scott, Kim F. Doheny, Elizabeth W. Pugh (2010) Description of transmission distortion in two family-based GWAS. *American Society of Human Genetics 60<sup>th</sup> Annual Meeting Abstracts*: 2965.
30. M. Graff, P. Gordon-Larsen, C.C. White, J. Ngwa, T. Workalemahu, T. Esko, N. Amin, P. Scheet, C. Schurmann, A. Teumer, C.S. Fox, L. Qi., R.M. van Dam, D. Strachan, A. Metspalu, C.M. van Duijn, D. Schlessinger, H. Völzke, **J.R. Shaffer**, M.L. Marazita, R.J. Crout, K.E. North, S. Berndt, L.A. Cupples (2011) The association between 32 common genetic BMI loci and longitudinal BMI across the transition from adolescence to young adulthood: a meta-analysis. *Obesity Society 29<sup>th</sup> Annual Scientific Meeting*.
31. M. Graff, P. Gordon-Larsen, C.C. White, J. Ngwa, T. Workalemahu, T. Esko, N. Amin, P. Scheet, C. Schurmann, A. Teumer, C.S. Fox, L. Qi., R.M. van Dam, D. Strachan, A. Metspalu, C.M. van Duijn, D. Schlessinger, H. Völzke, **J.R. Shaffer**, M.L. Marazita, R.J. Crout, K.E. North, S. Berndt, L.A. Cupples (2011) A meta-analysis of genetic loci for BMI in the lifecycle period between adolescence and young adulthood. *Obesity Society 29<sup>th</sup> Annual Scientific Meeting*.

32. MyoungKeun Lee, Karen T. Cuenco, Xiaojing Wang, **John R. Shaffer**, Ferdouse Begum, Eleanor Feingold, Daniel E. Weeks, Michael M. Barmada, Steven Wendell, David Crosslin, Cathy Laurie, Kimberly F. Doheny, Elizabeth Pugh, Robert J. Weyant, Richard J. Crout, Daniel W. McNeil, Mary L. Marazita. Genome-wide association study of saliva flow rate and integration of protein-gene webs. (2011) *European Human Genetics Conference 2011*.
33. Xiaojing Wang, Alex R. Vierira, **John R. Shaffer**, Ferdouse Begum, Myoungkeun Lee, Karen T. Cuenco, M. Michael Barmada, J Noel, I Anjomshoaa, Daniel E. Weeks, Eleanor Feingold, Mary L. Marazita. (2011) Follow-up GWAS analysis and in silico candidate gene study for dental caries in permanent teeth. *American Society of Human Genetics 61st Annual Meeting Abstracts*.
34. Zhen Zeng, **John R. Shaffer**, Xiaojing Wang, Myoungkeun Lee, Karen T. Cuenco, M. Michael Barmada, Robert J. Weyant, Richard Crout, Daniel W. McNeil, Steven Levy, Daniel E. Weeks, Eleanor Feingold, Mary L. Marazita. GWAS for childhood tooth decay implicated novel genes for pit and fissure and smooth surfaces. (2011) Genome-wide association study of saliva flow rate and integration of protein-gene webs. *American Society of Human Genetics 61st Annual Meeting Abstracts*.
35. Myoungkeun Lee, Karen T. Cuenco, Zhen Zeng, **John R. Shaffer**, Xiaojing Wang, M. Michael Barmada, Deborah E. Polk, Robert J. Weyant, Richard Crout, Daniel W. McNeil, Daniel E. Weeks, Eleanor Feingold, Mary L. Marazita. (2011) Identification of gene networks for caries of pit and fissures vs. smooth tooth surfaces through GWAS and protein network integration. *American Society of Human Genetics 61st Annual Meeting Abstracts*.
36. Karen T. Cuenco, Myoungkeun Lee, **John R. Shaffer**, Xiaojing Wang, M. Michael Barmada, Deborah E. Polk, Robert J. Weyant, Richard Crout, Daniel W. McNeil, Steven Levy, Daniel E. Weeks, Eleanor Feingold, Mary L. Marazita. (2011) Genome-wide association study of primary caries and integration of protein networks. *American Society of Human Genetics 61st Annual Meeting Abstracts*.
37. **John R. Shaffer**, Zeng Zhen, Xiaojing Wang, Karen T. Cuenco, M. Michael Barmada, Deborah E. Polk, Robert J. Weyant, Richard Crout, Daniel W. McNeil, Daniel E. Weeks, Eleanor Feingold, Mary L. Marazita. (2011) In silico candidate gene study for childhood tooth decay. *American Society of Human Genetics 61st Annual Meeting Abstracts*.
38. Frank Geller; Bjarke Feenstra; Hao Zhang; **John R Shaffer**; Thomas Hansen; Heather A Boyd; Ellen A Nohr; Nicholas J Timpson; David M Evans; Robert J Weyant; Steven M Levy; Mark Lathrop; George Davey Smith; Jeffrey C Murray; Thomas Werge; Mary L Marazita; Thorkild I A Sorensen; Mads Melbye. (2011) Genome-Wide Association Study Identifies Four Loci Associated with Eruption of Permanent Teeth. *American Society of Human Genetics 61st Annual Meeting Abstracts*.
39. Mary L. Marazita, John R. Shaffer, Eleanor Feingold, Xiaojing Wang, Steven Levy. (2012) Caries in the Primary and Permanent Dentitions. *81<sup>st</sup> Annual Meeting of the American Association of Physical Anthropologists*.
40. Cameron L. Randall, **John R. Shaffer**, Xiaojing Wang, Daniel W. McNeil, Richard J. Crout, Robert J. Weyant, Mary L. Marazita (2012) Relation between dental fear and fear of pain: How heritable? *International Association for Dental Research*.
41. Cameron L. Randall, Daniel W. McNeil, **John R. Shaffer**, Xiaojing Wang, Richard J Crout, Robert J. Weyant, Mary Marazita. (2012) Heritability of phobia: contributions of fears about pain. *Association for Behavioral and Cognitive Therapies*.
42. U. Ozbek, D. E. Weeks, W. Chen, **J. Shaffer**, S. M. Purcell, E. Feingold. (2012) Statistics for X-chromosome association. *American Society of Human Genetics 62<sup>nd</sup> Annual Meeting Abstracts*.
43. K. Cuenco, M. K. Lee, Z. Zeng, **J. R. Shaffer**, X. Wang, E. Feingold, D. E. Weeks, S. M. Levy, R. J. Weyant, R. J. Crout, D. W. McNeil, M. L. Marazita. (2012) Overlapping G x G interactions

- associated with caries in permanent and primary dentition. *American Society of Human Genetics 62<sup>nd</sup> Annual Meeting Abstracts*.
44. D. Lewis, E. Feingold, Z. Zeng, **J. Shaffer**, X. Wang, R. Weyant, D. McNeil, M. Marazita. (2012) The effect of race, sex, age, socioeconomic, behavioral factors, and genetics on dental caries in different tooth surfaces. *American Society of Human Genetics 62<sup>nd</sup> Annual Meeting Abstracts*.
  45. **J. R. Shaffer**, E. Feingold, X. Wang, M. Lee, K. T. Cuenco, D. E. Weeks, R. J. Weyant, R. Crout, D. W. McNeil, M. L. Marazita. (2012) GWAS needs a good phenotype: clustering tooth surfaces into biologically informative dental caries outcomes. *American Society of Human Genetics 62<sup>nd</sup> Annual Meeting Abstracts*.
  46. N. Mukhopadhyay, M. Govil, Z. Zheng, E. Feingold, D. E. Weeks, **J. R. Shaffer**, X. Wang, R. J. Weyant, R. Crout, D. W. McNeill, M. L. Marazita. (2012) Whole Genome Linkage Analysis to Identify Genes for Childhood Dental Caries. *American Society of Human Genetics 62<sup>nd</sup> Annual Meeting Abstracts*.
  47. M. Lee, K. T. Cuenco, Z. Zeng, X. Wang, **J. R. Shaffer**, E. Feingold, D. E. Weeks, R. J. Weyant, R. J. Crout, D. W. McNeil, M. L. Marazita. (2012) Identification of host gene-gene interaction in saliva flow and *Streptococcus mutans* using MDR. *American Society of Human Genetics 62<sup>nd</sup> Annual Meeting Abstracts*.
  48. X. Wang, Z. Zeng, **J. R. Shaffer**, E. Feingold, D. E. Weeks, M. Lee, K. T. Cuenco, R. J. Weyant, R. Crout, D. W. McNeil, M. L. Marazita. (2012) OFCD syndrome gene BCOR is associated with dental caries. *American Society of Human Genetics 62<sup>nd</sup> Annual Meeting Abstracts*.
  49. O. Harari, A. Goate, GENEVA Alcohol Consumption Working Group. (2012) Pathway analysis of the GENEVA Alcohol Consumption GWAS Meta-Analysis. *American Society of Human Genetics 62<sup>nd</sup> Annual Meeting Abstracts*.
  50. **J. R. Shaffer**, X. Wang, R. J. Weyant, R. Crout. D. W. McNeil, M. L. Marazita. (2012) Genetic Susceptibility to Dental Caries Differs between the Sexes. *5<sup>th</sup> American Association for Dental Research Fall Focused Symposium: Translational Genetics -- Advancing Front for Craniofacial Health*.
  51. Nandita Mukhopadhyay, Manika Govil, Zhen Zeng, Eleanor Feingold, Daniel E. Weeks, **John. R. Shaffer**, Xiaojing Wang, Robert J. Weyant, Richard Crout, Daniel W. McNeil, Mary L. Marazita. (2013) Genome-Wide Linkage Analysis to Identify Risk Genes for Dental Caries. *International Association for Dental Research/American Association for Dental Research/Canadian Association for Dental Research General Session & Exhibition*.
  52. Brooklyn Stanley, John R. Shaffer. Replication of Genetic Associations with Dental Caries (2013) *Summer Undergraduate Research Symposium. Bayer Learning Center & Mellon Hall of Science, Duquesne University*.
  53. **John R. Shaffer**, Robert J. Weyant, Richard Crout, Daniel W. McNeil, Mary L. Marazita. (2013) Unexpected pleiotropy: Do asthma and dental caries share a genetic basis? *American Society of Human Genetics 63<sup>rd</sup> Annual Meeting Abstracts*.
  54. D. Lewis, **J. Shaffer**, E. Feingold, M. Cooper, R. Weyant, D. McNeil, R. Crout, S. Reis, A. Vieira, M. Vanyukov, M. Marazita (2013) Replication of GWAS results for dental caries in the permanent dentition. *American Society of Human Genetics 63<sup>rd</sup> Annual Meeting Abstracts*.
  55. Amy Lowery, Dana H. Bovbjerg, Ellen Ingalls Beckjord, Adam Brufsky, Carol Marie Greco, Carissa A. Low, Jodi Martin, Donna M. Posluszny, John E. Schmidt, **John R. Shaffer**, Inna Belfer. (2014) Persistent pain following breast cancer surgery: Why does it hurt more for some survivors than others? *American Society of Clinical Oncology 50<sup>th</sup> Annual Meeting*.
  56. Zena T. Wolf, Boaz Arzi, Elizabeth J. Leslie, Harrison A. Brand, **John R. Shaffer**, Cali Willet, Nili Karmi1, Toby McHenry, Eleanor Feingold, Xioajing Wang, Jeffrey C. Murray, Mary L. Marazita,

- Claire M. Wade, Danika L. Bannasch. (2014) Advances in cleft lip and palate research utilizing less conventional model organisms. *American Society of Human Genetics 64<sup>th</sup> Annual Meeting*.
57. Deyana Lewis, **John R. Shaffer**, Eleanor Feingold, Margaret. Cooper, Michael M. Vanyukov, Brion S. Maher, Steven E. Reis, Daniel W. McNeil, Richard J. Crout, Robert J. Weyant, Steven M. Levy, Alexandre R.Vieira, Mary L. Marazita. (2014) The role of selected ion channel genes in dental caries. *American Society of Human Genetics 64<sup>th</sup> Annual Meeting*.
58. Jenna Colavincenzo, **John R. Shaffer**, Eleanor Feingold, Carla Sanchez, Toby McHenry, Frederic W. B. Deleyiannis, Daniel W. McNeil, Richard Crout, Robert J. Weyant, Mary L. Marazita. Genome-wide association studies for dental caries in African American and Latino populations: Novel genes and replication. *American Society of Human Genetics 64<sup>th</sup> Annual Meeting*.
59. Dmitry Shungin, Marilyn C. Cornelis, Kimon Divaris, Birte Holtfreter, **John R. Shaffer**, Yau-Hua Yu, Silvana P. Barros, James D. Beck, Reiner Biffar, Eric A. Boerwinkle, Richard J. Crout, Andrea Ganna, Goran Hallmans, George Hindy, Frank B. Hu, Peter Kraft, Daniel W. McNeil, Olle Melander, Kevin L. Moss, Kari E. North, Marju Orho-Melander, Nancy L. Pedersen, Paul M. Ridker, Eric B. Rimm, Lynda M. Rose, Gull Rukh, Alexander Teumer, Robert J. Weyant, Daniel I. Chasman, Kaumudi Joshipura, Thomas Kocher, Patrik K.E. Magnusson, Mary L. Marazita, Peter Nilsson, Steve Offenbacher, George Davey Smith, Pernilla Lundberg, Tom M. Palmer, Nicholas J. Timpson, Ingegerd Johansson, Paul W. Franks. (2014) Is Body Mass Index Causally Related with Periodontitis? Results from Mendelian Randomization study in the Gene-Lifestyle Interactions and Dental Endpoints (GLIDE) Consortium. *The Obesity Society and American Society for Metabolic and Bariatric Surgery 2014 Annual Meetings*.
60. Dmitry Shungin, Marilyn C. Cornelis, Kimon Divaris, Birte Holtfreter, **John R. Shaffer**, Yau-Hua Yu, Silvana P. Barros, James D. Beck, Reiner Biffar, Eric A. Boerwinkle, Richard J. Crout, Andrea Ganna, Goran Hallmans, George Hindy, Frank B. Hu, Peter Kraft, Daniel W. McNeil, Olle Melander, Kevin L. Moss, Kari E. North, Marju Orho-Melander, Nancy L. Pedersen, Paul M. Ridker, Eric B. Rimm, Lynda M. Rose, Gull Rukh, Alexander Teumer, Robert J. Weyant, Daniel I. Chasman, Kaumudi Joshipura, Thomas Kocher, Patrik K.E. Magnusson, Mary L. Marazita, Peter Nilsson, Steve Offenbacher, George Davey Smith, Pernilla Lundberg, Tom M. Palmer, Nicholas J. Timpson, Ingegerd Johansson, Paul W. Franks. (2014) Is Body Mass Index Causally Related with Periodontitis? Results from Mendelian Randomization study in the Gene-Lifestyle Interactions and Dental Endpoints (GLIDE) Consortium. *The International Genetics Epidemiology Society 2014 Annual Meeting*.
61. **John R. Shaffer**, Jenna C. Carlson, Daniel McNeil, Robert J. Weyant, Richard J. Crout, Eleanor Feingold, Mary L. Marazita. (2015) Dioxin receptor gene AHR is associated with early childhood caries. *International Association for Dental Research/American Association for Dental Research/Canadian Association for Dental Research General Session & Exhibition*.
62. Nandita Mukhopadhyay, Manika Govil, Eleanor Feingold, Daniel E. Weeks, **John R. Shaffer**, Robert J. Weyant, Richard Crout, Daniel W. McNeil, Mary L. Marazita. (2015) Multi-population Study of Risk Genes for Age-adjusted Dental Caries. *International Association for Dental Research/American Association for Dental Research/Canadian Association for Dental Research General Session & Exhibition*.
63. Mary L. Marazita, **John R. Shaffer**, Eleanor Feingold, Brooklyn Stanley, Jenna C. Carlson, Deyana Lewis, Xiaojing Wang, Daniel McNeil, Robert J. Weyant, Katherine Neiswanger, Betsy Foxman, Richard W. Crout. (2015) Genetic Risk Factors for Dental Caries in Children and Adults. *International Association for Dental Research/American Association for Dental Research/Canadian Association for Dental Research General Session & Exhibition*.
64. Steven F. Miller, **John R. Shaffer**, Mary L. Marazita, Steven M. Levy, John J. Warren, James K. Hartsfield, Lina M. Moreno. (2015) Exploratory Genome-wide Association Study of Dental Arch

- Shape. *International Association for Dental Research/American Association for Dental Research/Canadian Association for Dental Research General Session & Exhibition.*
65. Olivier Duverger, Takahiro Ohara, **John R. Shaffer**, Danielle Donahue, Patricia Zervas, Andrew Dullnig, Christopher Crecelius, Elia Beniash, Mary Marazita and Maria I. Morasso. (2015) Hair keratins as structural organic components of mature enamel: the link between hair disorders and susceptibility to dental caries. *International Association for Dental Research/American Association for Dental Research/Canadian Association for Dental Research General Session & Exhibition.*
  66. Deyana Lewis, Margaret Cooper, **John R. Shaffer**, Eleanor Feingold, Robert J. Weyant, Daniel McNeil, Richard W. Crout, Steven E. Reis, Steven M. Levy, Alexandre R. Vieira, Michael M. Vanyukov, Mary L. Marazita. (2015) GWAS Follow-Up Study of Caries in the Permanent Dentition. *International Association for Dental Research/American Association for Dental Research/Canadian Association for Dental Research General Session & Exhibition.*
  67. Cameron L. Randall, **John R. Shaffer**, Daniel W. McNeil, Richard J. Crout, Robert J. Weyant, and Mary L. Marazita. (2015) Dental Fear and MC1R Genotype: Fear of Pain as Mediator. *International Association for Dental Research/American Association for Dental Research/Canadian Association for Dental Research General Session & Exhibition.*
  68. **John R. Shaffer**, Joshua Rogan. (2015) Online human population genetics simulator: a tool for genetics/genomics education and research. *American Society of Human Genetics 65<sup>th</sup> Annual Meeting.*
  69. Leslie EJ, Carlson JC, **Shaffer JR**, Feingold E, Laurie C, Doheny K, Moreno L, Czeizel AE, Field LL, Deleyiannis F, Wehby G, Castilla EE, Vieira AR, Butali A, Christensen K, Ma L, Neiswanger K, Lidral A, Beaty T, Murray JC, Weinberg SW, Marazita ML. (2015) Uncovering novel genes contributing to phenotypic heterogeneity in a common birth defect. *American Society of Human Genetics 65<sup>th</sup> Annual Meeting.*
  70. Jenna C. Carlson, Elizabeth J. Leslie, Eleanor Feingold, **John R. Shaffer**, Holger Schwender, Margaret A. Taub, Ingo Ruczinski, Jeffrey C. Murray, Terri H. Beaty, Mary L. Marazita. (2015) Evaluating methods for identifying sources of phenotypic heterogeneity in family-based samples. *American Society of Human Genetics 65<sup>th</sup> Annual Meeting.*
  71. Tamar Sofer, **John R. Shaffer**, Misa Graff, Qibin Qi, Adrienne M. Stilp, Stephanie M. Gogarten, Kari E. North, Carmen Isasi, Bruce Weir, Cathy Laurie and Adam A. Szpiro. (2015) Meta-analysis of genome-wide association studies with correlated individuals: application to the Hispanic Community Health Study / Study of Latinos (HCHS/SOL). *American Society of Human Genetics 65<sup>th</sup> Annual Meeting.*
  72. Scott Eckert, **John R. Shaffer**. Genetic association of PKD2. (2015) *Summer Undergraduate Research Symposium. Bayer Learning Center & Mellon Hall of Science, Duquesne University.*
  73. Elizabeth J. Leslie, **John R. Shaffer**, Jenna C. Carlson, Eleanor Feingold, Cecelia Laurie, Kim Doheny, Katherine Neiswanger, Lina Moreno, Andrew Lidral, Andrew Czeizel, Carmencita Padilla, Frederic Deleyannis, L. Leigh Field, George Wehby, Alexandre Vieira, Eduardo Castilla, Azeez Butali, Jacqueline Hecht, Carmen Buxo, Kaare Christensen, Lian Ma, Jeffrey C. Murray, Seth M. Weinberg, Mary L. Marazita. (2015) Multi-ethnic genome-wide association studies of CL/P identify novel risk genes and evidence for genetic modifiers. *38<sup>th</sup> Annual Meeting of the Society of Craniofacial Genetics and Developmental Biology.*
  74. Seth M. Weinberg, Myoung Keun Lee, Elizabeth J. Leslie, Jacqueline T. Hecht, Carrie L. Heike, Michael L. Cunningham, George L. Wehby, Lina M. Moreno, Nichole L. Nidey, Eleanor Feingold, Mary L. Marazita, **John R. Shaffer** (2016) Genome-wide association study reveals novel loci influencing normal human facial morphology. *American Cleft Palate and Craniofacial Association 73<sup>rd</sup> Annual Meeting.*



75. Elizabeth J. Leslie, **John R. Shaffer**, Jenna C. Carlson, Huan Liu, Lina Moreno, George Wehby, Robert A. Cornell, Jeffrey C. Murray, Seth M. Weinberg, Mary L. Marazita (2016) Association of missense variant in GRHL3 in a genome-wide association study of isolated cleft palate. *American Cleft Palate and Craniofacial Association 73<sup>rd</sup> Annual Meeting*.
76. Rasha N Alotaibi, Myoungkeun Lee, Jenna Carlson, Katherine Neiswanger, **John Shaffer**, Margaret Cooper, Daniel McNeil, Robert Weyant, Betsy Foxman, Richard J Crout, Mary L. Marazita (2016) Role of Environmental and Behavioral Factors on Dental Caries Patterns. *American Association for Dental Research/Canadian Association for Dental Research Annual Meeting & Exhibition*.
77. Ballantine JL, Carlson JC, Zandona AF, Agler C, Zeldin LP, Rozier RG, Roberts MW, Slayton RL, Levy SM, **Shaffer JR**, Marazita ML, Divaris K (2016) Exploring the Genomic Basis of Early Childhood Caries. *American Academy of Pediatric Dentistry Annual Meeting*.
78. Lee MK, Leslie EJ, Carlson JC, Orlova E, Feingold E, Marazita ML, **Shaffer JR**, Weinberg SM. (2016) Genome-wide association study of multivariate measure of normal human facial morphology. *American Society of Human Genetics 66<sup>th</sup> Annual Meeting*.
79. Orlova E, Lee MK, Adkhar K, Li J, Callo C, Poletti G, Schuler-Faccini L, Bortolini MC, Canizales-Quinteros S, Rothhammer F, Bedoya G, Gonzalez-Jose R, Balding D, Leslie EJ, Feingold E, Heike CL, Cunningham ML, Hecht JT, Wehby GL, Moreno LM, Ding A, Tan J, Yang Y, Jin L, Marazita ML, Cox T, Wang S, Ruiz-Linares A, **Shaffer JR**, Weinberg SM. (2016) Trans-ethnic genome-wide meta-analysis of over 9000 individuals reveals multiple associations with earlobe attachment. *American Society of Human Genetics 66<sup>th</sup> Annual Meeting*.
80. Carlson JC, Leslie EJ, **Shaffer JR**, Christensen K, Deleyiannis F, Hecht JT, Vieira AR, Wehby GL, Feingold E, Weinberg SM, Marazita ML. (2016) Genome-wide interaction study implicates *VGLL2* and alcohol exposure in orofacial clefting. *American Society of Human Genetics 66<sup>th</sup> Annual Meeting*.
81. Leslie EJ, Carlson JC, **Shaffer JR**, Butali A, Buxo CJ, Christensen K, Deleyiannis F, Hecht JT, Moreno L, Orioli LM, Vieira AR, Wehby GL, Feingold E, Weinberg SM, Murray JC, Beaty TH, Marazita ML. (2016) Genome-wide analyses reveal subtype-specific genetic effects in orofacial cleft birth defects. *American Society of Human Genetics 66<sup>th</sup> Annual Meeting*.
82. Randall CL, McNeil DW, **Shaffer JR**, Weyant RJ, Crout RJ, Marazita ML. (2016) MC1R gene variation and fear related to dental care: Evidence of fear of pain as mediator. *American Society of Human Genetics 66<sup>th</sup> Annual Meeting*.
83. Arockiaraj A, Koleck TA, Shaffer JR, Sherwood PR, Crago EA, Poloyac SM, Weeks DE, Conley YP. (2016) Epigenome-wide association study of delayed cerebral ischemia in stroke patients. *American Society of Human Genetics 66<sup>th</sup> Annual Meeting*.
84. Duverger O, **Shaffer JR**, Carlson JC, Marazita ML, Morasso MI. (2016) Mutations in keratins involved in pachyonychia congenita are associated with dental enamel defects. *Enamel Symposium 2016 – Enamel9*.
85. Alotaibi RN, Howe BJ, Chernus JM, Mukhopadhyay N, Sanchez CA, Cooper ME, Deleyiannis FW, Neiswanger K, Resick J, Vieira AR, **Shaffer JR**, Moreno-Urbe M, Marazita ML. (2017) Genome-Wide Association Study of Dental Caries in Orofacial Cleft Families. *International Association for Dental Research/American Association for Dental Research/Canadian Association for Dental Research General Session & Exhibition*.
86. Mukhopadhyay N, Howe BJ, Chernus JM, Alotaibi R, Sanchez CA, Cooper ME, Emanuele BJ, Deleyiannis FW, Neiswanger K, Resick J, Vieira AR, Govil M, **Shaffer, JR**, Moreno-Urbe LM, Marazita ML. (2017) Multi-ethnic genome-wide association of caries combining adults and children. *International Association for Dental Research/American Association for Dental Research/Canadian Association for Dental Research General Session & Exhibition*.

87. LeClair J, Leslie EJ, Carlson JC, Feingold E, Buxo CJ, Castilla EE, Christensen K, Deleyiannia F, Field LL, Hecht JT, Moreno L, Orioli LM, Padilla C, Vieira AR, Wehby GL, Weinberg SM, Murray JC, Marazita ML, **Shaffer JR**. (2017) Association of Low-Frequency Variants in Regulatory Regions with Non-Syndromic Orofacial Clefts. *Joint Mathematics Meetings: 123<sup>rd</sup> Annual Meeting of the American Mathematical Society and 100<sup>th</sup> Annual winter meeting of the Mathematical Association of America*.
88. Wright CD, Randall CL, Chernus J, McNeil DW, Feingold E, Crout RJ, Neiswanger K, Weyant RJ, **Shaffer JR**, & Marazita ML (2017) Fear of Minor Pain is Associated with Specific Genetic Loci: Relations to Oral Health. *Association for Psychological Science 29<sup>th</sup> Annual Convention*.
89. Seth M. Weinberg, Myoung Keun Lee, Elizabeth J. Leslie, Ekaterina Orlova, Jenna C. Carlson, Brooke C. Mattern, Corey R. Liebowitz, Julie D. White, Arslan Zaidi, Diego Hernandez, Tomas Gonzalez, Laurel N. Pearson, Dzemila Sero, Jiarui Li, Jasmien Roosenboom, Eleanor Feingold, Mary L. Marazita, **John R. Shaffer**, Joanna Wysocka, Mark D. Shriver, Peter Claes. (2017) Modular 3D dense surface analysis and GWAS reveal localized genetic effects on human facial morphology involving multiple novel loci. *American Association of Anatomists Annual Meeting at Experimental Biology*.
90. Anna Zakas, R. Beth Dudley, Robin E. Grubs, **John R. Shaffer**, Michael Parkinson, Eve Karloski, Randall Brand. (2017) The Impact of Genetic Counseling on Patient Engagement. *National Society of Genetic Counselors 36<sup>th</sup> Annual Conference*.
91. Jenna C. Carlson, Nichole L. Nidey, **John R. Shaffer**, Azeez Butali, Carmen J. Buxo, Kaare Christensen, Frederic W-D Deleyiannis, Jacqueline T. Hecht, L. Leigh Field, Lina M. Moreno, Ieda M. Orioli, Carmencita Padilla, Alexandre R. Vieira, Seth M. Weinberg, George L. Wehby, Eleanor Feingold, Jeffrey C. Murray, Mary L. Marazita, Elizabeth J. Leslie. (2017) Exploring the impact of sex-specific genetic effects on orofacial clefting. *American Society of Human Genetics 67<sup>th</sup> Annual Meeting*.
92. Elizabeth J. Leslie, Jenna C. Carlson, Jennifer Standley, Aline Petrin, **John R. Shaffer**, Azeez Butali, Carmen J. Buxo, Kaare Christensen, Frederic W-D Deleyiannis, Jacqueline T. Hecht, L. Leigh Field, Lina M. Moreno, Ieda M. Orioli, Carmencita Padilla, Satoshi Suzuki, Natsume Nagato, Ariuntuul Garidkhuu, Alexandre R. Vieira, Seth M. Weinberg, George L. Wehby, Eleanor Feingold, Jeffrey C. Murray, Mary L. Marazita. (2017) Identification of 16q21 as a modifier locus for nonsyndromic orofacial cleft subphenotypes. *American Society of Human Genetics 67<sup>th</sup> Annual Meeting*.
93. Myoung Keun Lee, Jasmien Roosenboom, Jenna C. Carlson, Elizabeth J. Leslie, Eleanor Feingold, Mary L. Marazita, **John R. Shaffer**, Seth M. Weinberg. (2017) Genome-wide association study of cranial vault shape reveals novel loci at 15q26.3 and 17q11.2. *American Society of Human Genetics 67<sup>th</sup> Annual Meeting*.
94. Simon Haworth, Dmitry Shungin, Justin van der Tas, Strahinja Vucic, Caroline Medina Gomez, Victor Yakimov, **John Shaffer**, Myoung Keun Lee, Marie Standl, Carol Wang, Klaus Bønnelykke, Johannes Waage, Leon Eyrich Jesson, Pia Nørrisgaard, Raimo Joro, Ilkka Seppälä, Terho Lehtimäki, Timo Lakka, Hans Bisgaard, Craig Pennell, Jan Kühnisch, Mary Marazita, Frank Geller, Fernando Rivadeneira, Eppo Wolvius, Nicola West, Steve Thomas, Paul Franks, Nicholas Timpson, Ingegerd Johansson. (2017) Genome wide meta-analysis for dental caries in childhood. *American Society of Human Genetics 67<sup>th</sup> Annual Meeting*.
95. Ekaterina Orlova, Myoung Keun Lee, Eleanor Feingold, Daniel W. McNeil, Richard Crout, Robert J. Weyant, Mary L. Marazita, John R. Shaffer. (2017) GWAS of Early Childhood Caries in an Appalachian Population. *American Society of Human Genetics 67<sup>th</sup> Annual Meeting*.
96. A. Arockiaraj, **J.R. Shaffer**, P.R. Sherwood, E.A. Crago, S.M. Poloyac, D.E. Weeks, Y.P. Conley. (2017) Correlation of methylomic profiles between blood and cerebral spinal fluid in aneurysmal subarachnoid hemorrhage patients. *American Society of Human Genetics 67<sup>th</sup> Annual Meeting*.

97. **John R. Shaffer**, Jonathan M. Chernus, Cameron L. Randall, Casey Wright, Daniel W. McNeil, Richard J. Crout, Robert J. Weyant, Mary L. Marazita. (2017) Genome-wide association study of dental treatment-related fear nominates novel genes. *American Society of Human Genetics 67<sup>th</sup> Annual Meeting*.
98. Jessica LeClair, Elizabeth J. Leslie, Jenna C. Carlson, Eleanor Feingold, Carmen J. Buxo, Eduardo E. Castilla, Kaare Christensen, Frederic Deleyiannis, L. Leigh Field, Jacqueline T. Hecht, Lina M. Moreno, Leda M. Orioli, Carmencita Padilla, Alexandre R. Vieira, George L. Wehby, Seth M. Weinberg, Jeffrey C. Murray, Seth M. Weinberg, Mary L. Marazita, **John R. Shaffer**. (2018) Association of Low-Frequency Variants in Regulatory Regions with Non-Syndromic Orofacial Clefts. *American Cleft Palate-Craniofacial Association 75<sup>th</sup> Annual Meeting*.
99. Roosenboom J, Lee MK, Feingold E, Marazita ML, Maga M, **Shaffer JR**, Weinberg SM. (2018) Mapping Genetic Variants for Cranial Vault Shape in Humans. *American Cleft Palate-Craniofacial Association 75<sup>th</sup> Annual Meeting*.
100. Matthew Ford, Myoung Keun Lee, Jasmien Roosenboom, Jon Chernus, **John R. Shaffer**, Eleanor Feingold, Mary L. Marazita, Seth M. Weinberg. (2018) Velopharyngeal incompetence in a multiethnic cohort of families with a history of nonsyndromic orofacial clefting and population-based controls. *American Cleft Palate-Craniofacial Association 75<sup>th</sup> Annual Meeting*.
101. Jenna C. Carlson, Deepti Anand, **John R. Shaffer**, Carmen J. Buxo, Kaare Christensen, Fred Deleyiannis, Jacqueline T. Hecht, Lina Moreno, Ieda M. Orioli, Alexandre R. Vieira, George L. Wehby, Seth M. Weinberg, Jeffrey C. Murray, Terri H. Beaty, Eleanor Feingold, Mary L. Marazita, Elizabeth J. Leslie. (2018) A systematic analysis of orofacial clefts reveals subtype-specific genetic effects. *American Cleft Palate-Craniofacial Association 75<sup>th</sup> Annual Meeting*.
102. Jonathan M. Chernus, Jasmien Roosenboom, Matthew Ford, Lina M. Moreno Uribe, Luz Consuelo Valencia-Ramirez, Carmen J. Buxó, Jacqueline T. Hecht, Frederick W. B. Deleyiannis, Carmencita Padilla, Eleanor Feingold, **John R. Shaffer**, Elizabeth J. Leslie, Seth M. Weinberg, Mary L. Marazita. (2018) A genome-wide association study of velopharyngeal insufficiency. *American Cleft Palate-Craniofacial Association 75<sup>th</sup> Annual Meeting*.
103. Mary Marazita, Myoung Keun Lee, Jasmien Roosenboom, Elizabeth Leslie, Carla Sanchez, **John Shaffer**, Eleanor Feingold, George Wehby, Lina Moreno, Luz Valencia-Ramirez, Frederic Deleyiannis, Alexandre Vieira, Azeez Butali, Carmencita Padilla, Ieda Orioli, Carmen Buxo, Kaare Christensen, Jacqueline Hecht, Jeffrey Murray, Seth Weinberg. (2018) Subepithelial *orbicularis oris* muscle discontinuities in a multiethnic sample of individuals with nonsyndromic isolated cleft palate versus controls. *American Cleft Palate-Craniofacial Association 75<sup>th</sup> Annual Meeting*.
104. Laurence J Howe, Myoung Keun Lee, Gemma C Sharp, George Davey Smith, Beate St Pourcain, **John R Shaffer**, Mary L Marazita, Eleanor Feingold, Alexei Zhurov, Evie Stergiakouli, Jonathan Sandy, Stephen Richmond, Seth M Weinberg, Gibran Hemani, Sarah J Lewis (2018) Genetic variants for non-syndromic cleft influence philtrum width in the general population. *Craniofacial Society of Great Britain and Ireland Annual Meeting*.
105. Yuqiao Zhou, Richard Cuddy, Daniel McNeil, Casey Wright, Deborah Polk, Richard Crout, Robert Weyant, Eleanor Feingold, Katherine Neiswanger, Mary L. Marazita, **John R. Shaffer**. (2018) Oral Health Status and Dental Utilization Differs by Rurality in Appalachia. *American Association for Dental Research/Canadian Association for Dental Research joint General Session and Exhibition*
106. Daniel W. McNeil, **John R. Shaffer**, Jonathan M. Chernus, Cameron L. Randall, Casey D. Wright, Dongjing Liu, Richard J. Crout, Robert J. Weyant, Katherine Neiswanger, Mary L. Marazita (2018) Biological Influences on Dental Fear and Anxiety: Genome-Wide Association Study. *International Association for Dental Research 96<sup>th</sup> General Session and Exhibition*.
107. Rasha N. Alotaibi, Jonathan M. Chernus, Katherine Neiswanger, Nandita Mukhopadhyay, Brian Howe, Lina Moreno Uribe, Carla Sanchez, Alexandre R. Vieira, Daniel McNeil, **John R. Shaffer**,

- Mary L. Marazita (2018) Genome-Wide Association Study of Dental Caries in Diverse Caucasian Populations. *International Association for Dental Research 96<sup>th</sup> General Session and Exhibition*.
108. Jasmien Roosenboom, Myoung Keun Lee, Karlijne Indencleef, Eleanor Feingold, Mary L. Marazita, Mark D. Shriver, Peter Claes, **John R. Shaffer**, Seth M. Weinberg. (2018) Testosterone-related genetic variants are associated with human facial morphology. *International Association for Dental Research 96<sup>th</sup> General Session and Exhibition*.
  109. Ekaterina Orlova, Jenna C Carlson, Myoung Keun Lee, Eleanor Feingold, Daniel W McNeil, Richard J Crout, Robert J Weyant, Mary L Marazita, **John R Shaffer**. (2018) Genome Scans of Caries in African-Americans Show Genetic Heterogeneity. *International Association for Dental Research 96<sup>th</sup> General Session and Exhibition*.
  110. Ekaterina Orlova, Jon Chernus, Jenna C Carlson, Myoung Keun Lee, Eleanor Feingold, Daniel W McNeil, Richard J Crout, Robert J Weyant, Mary L Marazita, **John R Shaffer**. (2018) Genome Scans of Early Childhood Caries Phenotypes Identify Novel Associations. *International Association for Dental Research 96<sup>th</sup> General Session and Exhibition*.
  111. Lee MK, Roosenboom J, Feingold E, Marazita ML, **Shaffer JR**, Weinberg SM (2018) Genome-wide association studies of human facial width-to-height ratios. *American Society of Human Genetics 68<sup>th</sup> Annual Meeting*.
  112. Yuqiao Zhou, Jasmien Roosenboom, Myoung Keun Lee, Jacqueline T Hecht, Carrie L Heike, George L Wehby, Kaare Christensen, Lina M. Moreno, Eleanor Feingold, Mary L. Marazita, Peter Claes, Seth M. Weinberg, **John R. Shaffer** (2018) Genetic association of ER stress sensors IRE1 and XBP1 with cranial vault shape and nasal shape in humans. *American Society of Human Genetics 68<sup>th</sup> Annual Meeting*.
  113. Yuqiao Zhou, Hongjiao Ouyang, Daniel W. McNeil, Robert J. Weyant, Richard J. Crout, Rebecca Slayton, Trudy Burns, Steven M. Levy, Mary L. Marazita, **John R. Shaffer**. (2018) Genetic variants in ER stress sensors signaling molecules IRE1 and XBP1 are associated with human bone and tooth phenotypes. *American Society of Human Genetics 68<sup>th</sup> Annual Meeting*.
  114. Dongjing Liu, Myoung Keun Lee, Jasmien Roosenboom, Jacqueline T. Hecht, George L. Wehby, Lina M. Moreno, Carrie L. Heike, Mary L. Marazita, Eleanor Feingold, Seth M. Weinberg, **John R. Shaffer**. (2018) Low-frequency genetic variants contribute to human craniofacial morphology. *American Society of Human Genetics 68<sup>th</sup> Annual Meeting*.
  115. **John R. Shaffer**, Jessica LeClair, Jenna C. Carlson, Eleanor Feingold, Carmen J. Buxó, Kaare Christensen, Frederic W.B. Deleyiannis, L. Leigh Field, Jacqueline T. Hecht, Lina Moreno, Ieda M. Orioli, Carmencita Padilla, Alexandre R. Vieira, George L. Wehby, Jeffrey C. Murray, Seth M. Weinberg, Mary L. Marazita, Elizabeth J. Leslie. (2018) Association of Low-Frequency Variants in Regulatory Regions with Non-Syndromic Orofacial Clefts. *American Society of Human Genetics 68<sup>th</sup> Annual Meeting*.
  116. Roosenboom J, Lee MK, Feingold E, Marazita ML, Shriver MD, Wysocka J, Claes P, **Shaffer JR**, Weinberg SM. (2018) Detailed annotation of genetic signals associated with human nose morphology. *American Society of Human Genetics 68<sup>th</sup> Annual Meeting*.
  117. Sareena Ali, Daniel E. Ehrlich, Lina M. Moreno Uribe, Brad A. Amendt, Myoung Keun Lee, **John R. Shaffer**, Mary L. Marazita, Seth M. Weinberg, Steven F. Miller. (2019) Linking the Expression of Facial Shape and BMI via the Hippo Signaling Pathway. *Experimental Biology 2019 Annual Meeting*.
  118. Dongjing Liu, Myoung Keun Lee, Jiarui Li, Jacqueline T. Hecht, George L. Wehby, Lina M. Moreno, Carrie L. Heike, Jasmien Roosenboom, Eleanor Feingold, Mary L. Marazita, Peter Claes, Seth M. Weinberg, **John R. Shaffer**. (2019) Low-frequency coding variants contribute to normal human facial variation. *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition*.

119. Karlijne Indencleef, Jasmien Roosenboom, Hanne Hoskens, Julie D. White, Mark D. Shriver, Stephen Richmond, Hilde Peeters, Eleanor Feingold, Mary L. Marazita, **John R. Shaffer**, Seth M. Weinberg, Greet Hens, Peter Claes. (2019) Six NSCL/P loci show associations with normal-range craniofacial variation. *European Cleft Palate Craniofacial Association 2019 Congress*.
120. Hanne Hoskens, Karlijne Indencleef, Jiarui Li, Julie D. White, Alejandra Ortega-Castrillon, Joanna Wysocka, Susan Walsh, Stephen Richmond, **John R. Shaffer**, Seth M. Weinberg, Mark D. Shriver, Hilde Peeters, Peter Claes. A GWAS on data-driven 3D facial phenotypes selected by matching siblings reveals 310 genetic loci. *European Human Genetics Conference 2019*.
121. Ryan Eller, Julie D. White, Karlijne Indencleef, Sahin Naqvi, Jasmien Roosenboom, Myoung Keun Lee, Jaaved Mohammed, Stephen Richmond, Mary Marazita, Eleanor Feingold, Hilde Peeters, Greet Hens, Joanna Wysocka, **John R. Shaffer**, Seth M. Weinberg, Mark D. Shriver, Peter Claes, Susan Walsh. (2019) Approaches to help explain and potentially predict the complex architecture of the human face. *International Society for Forensic Genetics 28<sup>th</sup> International Congress*.
122. Dongjing Liu, Nora Alhazmi, Myoung Keun Lee, Jiarui Li, Jacqueline T. Hecht, George L. Wehby, Lina M. Moreno, Carrie L. Heike, Jasmien Roosenboom, Eleanor Feingold, Mary L. Marazita, Peter Claes, Chienwei E. Liao, Seth M. Weinberg, **John R. Shaffer**. (2019) Exome-wide low-frequency genetic variants contribute to human craniofacial morphology. *International Genetic Epidemiology Society 2019 Annual Conference*.
123. Elizabeth J. Leslie, Daniel Chang, Nadia Harerimana, Miranda R. Sun, Carmen Buxo, Kaare Christensen, Frederic Deleyiannis, Jacqueline T. Hecht, L. Leigh Field, Lina M. Moreno, Ieda M. Orioli, Carmencita Padilla, **John R. Shaffer**, Alexandre R. Vieira, Seth M. Weinberg, George L. Wehby, Eleanor Feingold, Jeffrey C. Murray, Mary L. Marazita, Robert J. Lipinski, Jenna C. Carlson. (2019) A locus on 4q28 is a laterality modifier with opposite effects for left- and right-sided cleft lip. *American Society of Human Genetics 69<sup>th</sup> Annual Meeting*.
124. Julie D. White, Karlijne Indencleef, Ryan J. Eller, Sahin Naqvi, Jasmien Roosenboom, Myoung Keun Lee, Jiarui Li, Jaaved Mohammed, Stephen Richmond, Eleanor Feingold, Mary L. Marazita, Hilde Peeters, Greet Hens, **John R. Shaffer**, Joanna Wysocka, Susan Walsh, Seth M. Weinberg, Mark D. Shriver, Peter Claes. (2019) 203 SNPs with combinatorial effects contribute to the genetic architecture of facial morphology. *American Society of Human Genetics 69<sup>th</sup> Annual Meeting*.
125. Yuqiao Zhou, Daniel W. McNeil, George L. Wehby, Frederic W. Deleyiannis, Jacqueline T. Hecht, Robert J. Weyant, Richard J. Crout, Eleanor Feingold, Mary L. Marazita, **John R. Shaffer**. (2019) Common and low-frequency coding variants at the ERN1 and XBP1 loci are associated with dental caries in the primary dentition. *American Society of Human Genetics 69<sup>th</sup> Annual Meeting*.
126. Lee MK, Roosenboom J, Indencleef K, White JD, Carlson JC, Feingold E, Beaty T, Leslie EJ, Marazita ML, Walsh S, Shriver MD, Claes P, **Shaffer JR**, Weinberg SM. (2019) Investigating genetic links between normal-range facial variation and orofacial clefting. *American Society of Human Genetics 69<sup>th</sup> Annual Meeting*.
127. Chenxing Liu, Myoung Keun Lee, Mary L. Marazita, Seth M. Weinberg, **John R. Shaffer**. (2019) Comparing archaic to modern human-derived sites reveals the evolution of human facial morphology. *American Society of Human Genetics 69<sup>th</sup> Annual Meeting*.
128. Tianyu Zou, Myoung Keun Lee, Luiza A.F. De Souza, Charlotte E. Skinner, Alyssa M. Azevedo, Mary L. Marazita, Seth M. Weinberg, **John R. Shaffer**. (2019) Genome-wide mapping for palpebral fissure inclination. *American Society of Human Genetics 69<sup>th</sup> Annual Meeting*.
129. Dongjing Liu, Myoung Keun Lee, Jiarui Li, Jacqueline T. Hecht, George L. Wehby, Lina M. Moreno, Carrie L. Heike, Jasmien Roosenboom, Eleanor Feingold, Mary L. Marazita, Peter Claes, Seth M. Weinberg, **John R. Shaffer**. (2019) Exome-wide low-frequency genetic variants contribute to human craniofacial morphology. *American Society of Human Genetics 69<sup>th</sup> Annual Meeting*.

130. Ana C. Driscoll, Myoung Keun Lee, Mary L. Marazita, Peter Claes, Seth M. Weinberg, **John R. Shaffer**. (2019) Association of human accelerated regions with craniofacial and subcortical morphology. *American Society of Human Genetics 69<sup>th</sup> Annual Meeting*.
131. Julie D. White, Karlijne Indencleef, Sahin Naqvi, Ryan J. Eller, Stephen Richmond, Mary L. Marazita, **John R. Shaffer**, Joanna Wysocka, Susan Walsh, Seth M. Weinberg, Mark D. Shriver, Peter Claes. (2020) Mapping genes that drive variation in human facial shape. *2020 Gordon Research Conference on Craniofacial Morphogenesis & Tissue Regeneration*.
132. Rasha N. Alotaibi, Brian J. Howe, Lina Moreno Uribe, Consuelo Valencia Ramirez, Claudia Restrepo, Frederic W.B. Deleyiannis, Carmencita Padilla, Ieda M. Orioli, Carmen J. Buxó, Jacqueline T. Hecht, George L. Wehby, Katherine Neiswanger, **John R. Shaffer**, Seth M. Weinberg, Mary L. Marazita. (2020) Multivariate GWAS of Correlated Dental Traits in a Multiethnic Cohort. *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition*.
133. Chenxing Liu, Karlijne Indencleef, Jiarui Li, Myoung Keun Lee, Dongjing Liu, Harry Matthews, Julie D. White, Sahin Naqvi, Ryan J. Eller, Jaaved Mohammed, Tomek Swigit, Stephen Richmond, Benedikt Hallgrímsson, Richard A. Spritz, Eleanor Feingold, Mary L. Marazita, Joanna Wysocka, Susan Walsh, Mark D. Shriver, Peter Claes, **John R. Shaffer**, Seth M. Weinberg. (2020) Genome Scan of Facial Features in Africans Reveals Novel Associations. *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition*.
134. Yuqiao Zhou, Daniel W. McNeil, Richard J. Crout, Robert J. Weyant, Steve Reis, Mary L. Marazita, **John R. Shaffer**. Exome-wide low-frequency genetic variants contribute to chronic periodontitis-related phenotype. (2020) *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition*.
135. Tianyu Zou, Mary L. Marazita, John J. Warren, Steven M. Levy, **John R. Shaffer** Genome-wide Association Study of Longitudinal Dental Caries Trajectory Clusters. (2020) *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition*.
136. Jonathan M. Chernus, Jasmien Roosenboom, Myoung Keun Lee, Azeez Butali, Carmen J. Buxó, Kaare Christensen, Frederic W. B. Deleyiannis, Jacqueline T. Hecht, Lina M. Moreno Uribe, Iêda M. Orioli, Carmencita D. Padilla, Fernando A. Poletta, Luz Consuelo Valencia-Ramirez, George L. Wehby, Katherine Neiswanger, Eleanor Feingold, **John R. Shaffer**, Elizabeth J. Leslie, Jeffrey C. Murray, Seth M. Weinberg, Mary L. Marazita. Subclinical Lip Defects and Clefts Associated with Different Genetics Factors (2020) *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition*.
137. **John R. Shaffer**, Daniel McNeil, Yuqiao Zhou, Jamey Brumbaugh, Robert J. Weyant, Richard J. Crout, Steven Reis, Mary L. Marazita. Genome-wide scan of dental fear and anxiety nominates novel genes. (2020) *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition*.
138. Olubukola O. Olatosi, **John R. Shaffer**, Afolabi Oyapero, Tamara Busch, Mary Li, John Pape, Mohamed Hassan, Joy Olotu, Waheed Awotoye, Azeez Alade, Chinyere Adeleke, Wasiu L. Adeyemo, Mary L. Marazita, Azeez Butali. Early childhood caries in Nigerian children. (2020) *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition*.
139. Nandita Mukhopadhyay, **John Shaffer**, Katherine Neiswanger, Robert J. Weyant, Betsy Foxman, Richard J. Crout, Dan McNeil, Mary L. Marazita. Identifying Genes For Lifetime Caries Risk In An

- Appalachian Sample. (2020) *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition*.
140. E Orlova, N Mukhopadhyay, M Lee, DW McNeil, RJ Crout, RJ Weyant, E Feingold, **JR Shaffer**, ML Marazita. Exome-wide rare variant analysis of early childhood caries. (2020) *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition*.
  141. Yuqiao Zhou, Daniel W. McNeil, George L. Wehby, Frederic W. Deleyiannis, Jacqueline T. Hecht, Robert J. Weyant, Richard J. Crout, Eleanor Feingold, Mary L. Marazita, **John R. Shaffer**. (2019) Common and low-frequency coding variants at the ERN1 and XBP1 loci are associated with dental caries in the primary dentition. *University of Pittsburgh 2<sup>nd</sup> Annual Interdepartmental Genetics Retreat*.
  142. Lee MK, Roosenboom J, Indencleef K, White JD, Carlson JC, Feingold E, Beaty T, Leslie EJ, Marazita ML, Walsh S, Shriver MD, Claes P, **Shaffer JR**, Weinberg SM. (2019) Investigating genetic links between normal-range facial variation and orofacial clefting. *University of Pittsburgh 2<sup>nd</sup> Annual Interdepartmental Genetics Retreat*.
  143. Chenxing Liu, Myoung Keun Lee, Mary L. Marazita, Seth M. Weinberg, **John R. Shaffer**. (2019) Comparing archaic to modern human-derived sites reveals the evolution of human facial morphology. *University of Pittsburgh 2<sup>nd</sup> Annual Interdepartmental Genetics Retreat*.
  144. Tianyu Zou, Myoung Keun Lee, Luiza A.F. De Souza, Charlotte E. Skinner, Alyssa M. Azevedo, Mary L. Marazita, Seth M. Weinberg, **John R. Shaffer**. (2019) Genome-wide mapping for palpebral fissure inclination. *University of Pittsburgh 2<sup>nd</sup> Annual Interdepartmental Genetics Retreat*.
  145. Dongjing Liu, Myoung Keun Lee, Jiarui Li, Jacqueline T. Hecht, George L. Wehby, Lina M. Moreno, Carrie L. Heike, Jasmien Roosenboom, Eleanor Feingold, Mary L. Marazita, Peter Claes, Seth M. Weinberg, **John R. Shaffer**. (2019) Exome-wide low-frequency genetic variants contribute to human craniofacial morphology. *University of Pittsburgh 2<sup>nd</sup> Annual Interdepartmental Genetics Retreat*.
  146. Rasha N. Alotaibi, Brian Howe, Lina Moreno Uribe, Katherine Neiswanger, **John R. Shaffer**, Seth M. Weinberg, Mary L. Marazita. (2019) GWAS of Enamel Hypoplasia in a Multiethnic Cohort. *10th International Symposium of Dental Enamel*.
  147. Dongjing Liu, Myoung Keun Lee, Jacqueline T. Hecht, George L. Wehby, Lina M. Moreno, Carrie L. Heike, Mary L. Marazita, Peter Claes, Seth M. Weinberg, **John R. Shaffer**. (2020) Transcriptome-wide association study of human facial shape identifies potential mediating genes. *International Genetic Epidemiology Society 2020 Annual Conference*. **first-place poster award**
  148. Lacey W. Heinsberg, Daniel E. Weeks, **John R. Shaffer**, Yvette P. Conley. (2020) Characterization of Cerebrospinal Fluid DNA Methylation Age Following Subarachnoid Hemorrhage. *International Society of Nurses in Genetics 2020 World Congress*.
  149. Chenxing Liu, Myoung Keun Lee, Dongjing Liu, Julie D. White, Karlijne Indencleef, Sahin Naqvi, Ryan J. Eller, Jiarui Li, Stephen Richmond, Benedikt Hallgrímsson, Richard A. Spritz, Eleanor Feingold, Mary L. Marazita, Joanna Wysocka, Susan Walsh, Mark D. Shriver, Peter Claes, Seth M. Weinberg, **John R. Shaffer**. (2020) Genome Scan of Facial Features in Africans Reveals Novel Associations. *American Society of Human Genetics 70<sup>th</sup> Annual Meeting*.
  150. Dongjing Liu, Myoung Keun Lee, Jacqueline T. Hecht, George L. Wehby, Lina M. Moreno, Carrie L. Heike, Mary L. Marazita, Peter Claes, Seth M. Weinberg, **John R. Shaffer**. (2020) Transcriptome-wide association study of human facial morphology identified putative mediating genes. *American Society of Human Genetics 70<sup>th</sup> Annual Meeting*.
  151. Yuqiao Zhou, Daniel W. McNeil, Simon Haworth, Jonathan M. Chernus, Chenxing Liu, Dongjing Liu, Casey D. Wright, Jamey Brumbaugh, Cameron L. Randall,

- Robert J. Weyant, Richard J. Crout, Steven Reis, Nicolas J. Timpson, Mary L. Marazita, **John R. Shaffer**. (2020) Genome-wide scan of dental fear and anxiety nominates novel genes. *American Society of Human Genetics 70<sup>th</sup> Annual Meeting*.
152. Sarah W. Curtis, Daniel Chang, Myoung Keun Lee, **John R. Shaffer**, Karlijne Indecleef, Peter Claes, Jeff Murray, Eleanor Feingold, Terri H. Beaty, Seth M. Weinberg, Mary L. Marazita, Jenna C. Carlson, Elizabeth J. Leslie. (2020) 20p11 as a laterality modifier of orofacial clefts and the distinct genetic etiology of nonsyndromic bilateral cleft lip. *American Society of Human Genetics 70<sup>th</sup> Annual Meeting*.
153. Tianyu Zou, Mary L. Marazita, John J. Warren, Steven M. Levy, **John R. Shaffer**. (2020) Genome-wide association study of longitudinal dental caries trajectory clusters. *American Society of Human Genetics 70<sup>th</sup> Annual Meeting*.
154. Ahmed El Sergani, Stephanie Brandebura, Carmencita Padilla, Azeez Butali, Wasiu Adeyemo, Luz Valencia-Ramirez, Claudia Restrepo, Lina Moreno, Carmen Buxo, Ross Long, **John R. Shaffer**, Katherine Neiswanger, Mary Marazita, Seth Weinberg. (2020) Altered Palate Shape as a Subclinical Phenotypic Risk Factor for Orofacial Clefting. *American Cleft Palate-Craniofacial Association 78th Annual Meeting*.
155. Rasha N. Alotaibia, Brian J. Howe, Jonathon M. Chernus, Nandita Mukhopadhyay, Carla Sanchez, Frederic W.B. Deleyiannis, Katherine Neiswanger, Carmencita Padilla, Fernando A. Poletta, Ieda M. Orioli, Carmen J. Buxó, Jacqueline T. Hecht, George L. Wehby, Ross E. Long, Alexandre R. Vieira, Jeffrey C. Murray, Seth M. Weinberg, **John R. Shaffer**, Lina Moreno Uribe, Mary L. Marazita. (2021) Genome-Wide Association Study of Dental Caries in Families with Non-syndromic Orofacial Clefts. *American Cleft Palate-Craniofacial Association 78th Annual Meeting*.
156. Nandita Mukhopadhyay, Eleanor Feingold, Lina Moreno-Urbe, George Wehby, Luz Consuelo Valencia-Ramirez, Claudia P. Restrepo Muñeton, Carmencita Padilla, Frederic Deleyiannis, Kaare Christensen, Fernando A. Poletta, Ieda M Orioli, Jacqueline T. Hecht, Carmen J. Buxó, Azeez Butali, Wasiu L. Adeyemo, Alexandre R. Vieira, **John R. Shaffer**, Jeffrey C. Murray, Seth Weinberg, Elizabeth J. Leslie, Mary L. Marazita. (2020) Genome-wide association study of orofacial clefts in a multiethnic sample of families to identify etiological differences between types of clefts. *Society for Craniofacial Genetics and Developmental Biology 43<sup>rd</sup> Annual Meeting*.
157. Caroline Bong, Lauren Morgenroth, Robin E. Grubs, Roxanna M. Bendixen, **John Shaffer**. (2020) Exploring Factors that Influence Reproductive Decision-Making in Duchenne Muscular Dystrophy Carriers. *39<sup>th</sup> National Society of Genetic Counselors Annual Conference*.
158. T. Tiwari, C. L. Randall, N. Rai, **J. R. Shaffer**, T. L. Finlayson, L. M. Kaste, N. Laniado, R. H. Singer. Association Between Acculturation and Dental care-related Anxiety in Hispanic/Latino Population. (2021) *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition*.
159. J. An, K. C. Sokoto, E. C. Kao, K. Neiswanger, B. Foxman, **J. R. Shaffer**, M. L. Marazita, D. W. McNeil. Effects of Depression and Stress on Maternal Oral Self-Care. (2021) *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition*.
160. Y. Zhou, D. W. McNeil, S. Haworth, T. Dudding, J. M. Chernus, C. Liu, D. Liu, C. D. Wright, J. Brumbaugh, C. L. Randall, R. Weyant, R. J. Crout, B. Foxman, S. Reis, N. Timpson, M. L. Marazita, **J. R. Shaffer**. Exploring the genetic basis of dental fear and anxiety. (2021) *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition*.
161. T. Zou, K. Neiswanger, E. Feingold, B. Foxman, D. W. McNeil, M. L. Marazita, **J. Shaffer**. Genome-wide survival analysis of dental caries incidence. (2021) *International Association for Dental*



- Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition.*
162. K. C. Sokoto, J. Brumbaugh, J. An, D. E. Polk, L. Platt, E. C. Kao, K. Neiswanger, B. Foxman, **J. R. Shaffer**, M. L. Marazita, L. A. Alexander, D. W. McNeil. Associations between Racial Discrimination and Anxiety/Fear in Dental Settings. (2021) *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition.*
  163. R. N. Alotaibi, B. J. Howe, L. M. Moreno Uribe, C. Valencia Ramirez, C. Restrepo Muñeton, F. Deleyiannis, C. D. Padilla, I. Orioli, C. J. Buxó-Martínez, J. T. Hecht, G. L. Wehby, K. Neiswanger, **J. R. Shaffer**, S. M. Weinberg, M. L. Marazita. Multivariate GWAS of Correlated Dental Traits in a Multiethnic Cohort. (2021) *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition.*
  164. E. Orlova, R. Alotaibi, J. M. Chernus, M. Lee, N. Mukhopadhyay, E. Feingold, D. W. McNeil, R. J. Crout, B. Foxman, M. L. Marazita, **J. R. Shaffer**. Genome Scan of Early Childhood Caries Again Implicates Beta-Defensins. (2021) *International Association for Dental Research/American Association for Dental Research/ Canadian Association for Dental Research General Session & Exhibition.*
  165. J.T. Brumbaugh, M.C. Arias, C.D. Wright, K. Neiswanger, **J.R. Shaffer**, B. Foxman, M.L. Marazita, D.W. McNeil. Infant Nighttime Sleep Trajectories in Northern Appalachia. (2021) *Association for Behavioral and Cognitive Therapies 55th Annual Convention.*
  166. B. E. Zusman, D. Liu, **J. R. Shaffer**, P. M. Kochanek, D. E. Weeks, Y. P. Conley, A. M. Puccio, D. O. Okonkwo, A. Rani, S. Raikwar, R. M. Jha. Epigenetic regulation of ABCC8 and TRPM4 is associated with intracranial hypertension and outcome after severe TBI. (2021) *American Neurological Association 146<sup>th</sup> Annual Meeting.*
  167. Treble-Barna A, Heinsberg LW, Puccio AM, **Shaffer JR**, Okonkwo DO, Beers SR, Weeks DE, Conley YP. Acute Brain-Derived Neurotrophic Factor DNA Methylation Trajectories in CSF and Associations with Outcomes following Severe Traumatic Brain Injury in Adults. (2021) *Annual Meeting of the National Neurotrauma Society.*
  168. Dongjing Liu, Hyo-Jeong Ban, Ahmed M. El Sergani, Myoung Keun Lee, Jacqueline T. Hecht, George L. Wehby, Lina M. Moreno, Eleanor Feingold, Mary L. Marazita, Seongwon Cha, Heather Szabo-Rogers, Seth M. Weinberg, **John R. Shaffer**. PRICKLE1 x FOCAD interaction revealed by genome-wide vQTL analysis of human facial traits. *2021 International Genetic Epidemiology Society Annual Meeting. first-place poster award*
  169. Deesha Bhaumik, Casey D. Wright, Teresa A. Marshall, Katherine Neiswanger, Daniel W. McNeil, Andrew D. Jones, **John R. Shaffer**, Mary L. Marazita, Betsy Foxman. Food insecurity and consumption of cariogenic foods in mothers and their two-year-old children in Appalachia. (2022) *American Association for Dental, Oral, and Craniofacial Research 51<sup>st</sup> General Session and Exhibition and Canadian Association for Dental Research 26<sup>th</sup> Annual Meeting.*
  170. An J, Lilly C, Potter K, Kao EC, Foxman B, **Shaffer JR**, Marazita ML, McNeil DW. Negative Emotion and Oral Self-Care among Women in Appalachia. (2022) *American Association for Dental, Oral, and Craniofacial Research 51<sup>st</sup> General Session and Exhibition and Canadian Association for Dental Research 26<sup>th</sup> Annual Meeting.*
  171. Victory EJ, Tallman T, Sokoto KC, Kao EC, Foxman B, **Shaffer JR**, Alexander L, Marazita ML, McNeil DW. Discrimination based on Social Class in Oral Healthcare in Appalachia. (2022) *American Association for Dental, Oral, and Craniofacial Research 51<sup>st</sup> General Session and Exhibition and Canadian Association for Dental Research 26<sup>th</sup> Annual Meeting.*
  172. Seth M. Weinberg, Myoung Keun Lee, Imani Gilbert, Eshan Schleif, Neda Tahmasebifard, Jamie L. Perry, John R. Shaffer. Genome Scans Identify Variants Impacting Human Velopharyngeal

Morphology. (2022) *American Association of Anatomy 2022 Annual Meeting at Experimental Biology*.

**Presentations**

1. **John R. Shaffer**. *American Society for Bone and Mineral Research 31<sup>st</sup> Annual Meeting. Journal of Bone and Mineral Research*. 2009. 24 Suppl 1:S55.
2. **John R. Shaffer**. Dental caries in the Permanent Dentition, G and GxE. *GENEVA Consortium Steering Committee Meeting*. January 2011.
3. **John R. Shaffer**. Dental caries: New cohort and further analyses of permanent dentition. *GENEVA Consortium Steering Committee Meeting*. May 2011.
4. Kelli Ryckman, **John R. Shaffer**. GWAS of Prematurity and Complications of Prematurity in African-Americans. *GENEVA Consortium Steering Committee Meeting*. May 2011.
5. **John R. Shaffer**. Revisiting GWAS using novel caries phenotypes. *GENEVA Consortium Steering Committee Meeting*. December 2011.
6. **John R. Shaffer**. Keratins discovered in tooth associated with dental defects and tooth decay. Department of Human Genetics Dean’s Annual Review. April 2014.
7. **John R. Shaffer**. Genetic epidemiology of dental caries. Graduate School of Public Health Board of Visitors. November 2015.
8. **John R. Shaffer**. Association of Low-Frequency Variants in Regulatory Regions with Non-Syndromic Orofacial Clefts. *American Cleft Palate-Craniofacial Association 75<sup>th</sup> Annual Meeting*.
9. **John R Shaffer**. (2018) Genome Scans of Early Childhood Caries Phenotypes Identify Novel Associations. *International Association for Dental Research 96<sup>th</sup> General Session and Exhibition*.
10. **John R. Shaffer**. Genetic architecture of human facial morphology. University of Pittsburgh 1<sup>st</sup> Annual Interdepartmental Genetics Retreat. September 2018.
11. **John R. Shaffer**. (2020) Nature and Nurture: Where Dental and Craniofacial Genetics Meets Environment. Symposium: The Social Exposome: Opportunities for Precision Dental and Craniofacial Health. *International Association for Dental Research 96<sup>th</sup> General Session and Exhibition*.
12. **John R. Shaffer**. (2020) Genome-wide scan of dental fear and anxiety nominates novel genes. *International Association for Dental Research/American Association for Dental Research/Canadian Association for Dental Research General Session & Exhibition*.

**Non Peer-reviewed Publications**

1. Seth M. Weinberg, **John R. Shaffer**. We scanned the DNA of 8,000 people to see how facial features are controlled by genes. *The Conversation* (Dec. 7, 2020)  
<https://theconversation.com/we-scanned-the-dna-of-8-000-people-to-see-how-facial-features-are-controlled-by-genes-151539>.

**Service**

**Service to the Department of Human Genetics**

Year	Committee	Position
2007	Strategic Planning Committee	appointed member

<b>Year</b>	<b>Committee</b>	<b>Position</b>
2011-2021	Curriculum Committee	appointed member
2012	Strategic Planning Committee	appointed member
2015-2021	Admissions Committee	elected chair
2018	Reaccreditation Committee, Thesis Review Sub-committee, Genetic Counseling Program	invited member
2018-present	Communications Committee, Website Sub-committee	appointed member
2018-present	Computing Committee	appointed member
2021-present	Curriculum Committee	chair
2021-present	Executive Committee	member

### **Service to the School of Public Health**

<b>Year</b>	<b>Committee</b>	<b>Position</b>
2011-2013	Alumni Panel, Accepted Applicants Day	invited panelist
2012-2021	Dean's Day Faculty Judge	judge
2015	Council on Education for Public Health Site Visit: Session on Research, Service, Workforce Development	invited representative
2015-2020	Educational Policies and Curriculum Committee	elected representative
2018	Internal Scientific Grant Review, Department of Epidemiology	<i>ad hoc</i> grant reviewer
2020-2021	Educational Policies and Curriculum Committee	elected chair
2020-2021	Graduate School of Public Health Council	member
2020-2021	Faculty Senate Executive Committee	member
2021-present	Search Committee, Chair of the Department of Biostatistics, Graduate School of Public Health	appointed member
2021-present	Social justice, equity, diversity, and inclusion (JEDI) Council Culture Working Group, School of Public Health	member

### **Service to the University**

<b>Year</b>	<b>Committee</b>	<b>Position</b>
2012	Center for Research and Evaluation Pilot/Feasibility Study Program, School of Nursing	<i>ad hoc</i> grant reviewer
2014-2016	Research Review Committee, Department of Psychiatry, School of Medicine	<i>ad hoc</i> grant reviewer
2017	Competitive Medical Research Fund review committee, Office of Research, Health Sciences, University of Pittsburgh	<i>ad hoc</i> grant reviewer

### **Service to Field of Scholarship**

#### **Editorial Boards, Editorships**

<b>Date</b>	<b>Position</b>	<b>Journal</b>
2014-present	Associate Editor	BMC Oral Health

Date	Position	Journal
2017-2018	Topic Editor	Frontiers in Genetics

#### Manuscript and Other Document/Publication Review

Year	Journal Title
2009	Diabetes
2009	Diabetes and Metabolism Research and Reviews
2009	Disease Markers
2010	Osteoporosis International
2010-2012	Journal of Bone and Mineral Research
2011	American Journal of Epidemiology
2011	Human Heredity
2011	Human Genetics
2012	Melanoma Research
2012-2019	Journal of Dental Research
2012-2017	Human Molecular Genetics
2012-2015	JAMA: The Journal of the American Medical Association
2013	Molecular Genetics & Genomic Medicine
2014	PLOS ONE
2014	Genes and Immunity
2014	New England Journal of Medicine
2014-2015	Caries Research
2014	International Journal of Oral Science
2015	Biomedical and Environmental Sciences
2015	Oral Diseases
2015	International Journal of Paediatric Dentistry
2016	Australian Dental Journal
2017-2018	Journal of Dental Research: Clinical and Translational Research
2017-2021	Archives of Oral Biology
2018	Molecular Biology Reports
2018	Scientific Reports
2018-2019	PLOS Genetics
2019	Annals of Translational Medicine
2019	Annals of Human Genetics
2020-2021	Clinical Oral Investigations
2021	European Journal of Oral Sciences
2021	American Journal of Medical Genetics: Part A

#### Study Sections, Review Panels, and Advisory Boards

Date	Position	Organization
Feb. 2015	<i>ad hoc</i> grant reviewer	Health Services and Population Health Research Committee, Scottish Government Health and Social Care Directorates
Aug. 2015	<i>ad hoc</i> grant reviewer	Children's Research Foundation of South Australia
Feb. 2018	<i>ad hoc</i> grant reviewer	Health Research Council of New Zealand

<b>Date</b>	<b>Position</b>	<b>Organization</b>
May 2016	Assessor	Evaluated applications for the Academic Promotions Committee, University of Sidney
Jan. 2022	<i>ad hoc</i> member	NIH Study Section, Scientific Review Group 2022/05 (01) M CIDR: Center for Inherited Disease Research Access Committee

#### **Leadership in Scholarly and Professional Organizations and Honorary Societies**

<b>Year</b>	<b>Position</b>	<b>Organization</b>
2010	Platform Moderator	American Society of Human Genetics
2021-present	Representative	Association of Schools and Programs of Public Health, PhD Council