

## Curriculum Vitae

### John R. Shaffer

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## Education and Training

### Undergraduate

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

2003-2008	University of Pittsburgh, Pittsburgh, PA	PhD, 2008	Human Genetics
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### Post-Graduate

2008-2011	University of Pittsburgh, Pittsburgh, PA	Post-doctoral	Human Genetics
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## Appointments and Positions

### Academic

#### Primary

2011-2013	Research Assistant Professor	Department of Human Genetics
2013-2020	Assistant Professor (appointment stream)	School of Public Health
2015-present	Graduate Faculty	University of Pittsburgh
2020-present	Assistant Professor (tenure-stream)	

#### Secondary

2017-present	Assistant Professor	Dept. of Oral and Craniofacial Sciences School of Dental Medicine University of Pittsburgh
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#### Administrative

2021-present	Vice Chair for Education	Department of Human Genetics
2021-present	Director of Graduate Studies	School of Public Health
2021-present	Director, MS and PhD Programs	University of Pittsburgh

## 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, Oral, and Craniofacial 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
2024-present	American Statistical 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
2023	Nomination for the James L. Craig Excellence in Teaching Award, University of Pittsburgh, School of Public Health

## Professional Activities

### Teaching

#### 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
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- 2021	HUGEN 2022: Human Population Genetics University of Pittsburgh	2 hours per week 2 credits 35-60 students	Course director / instructor (100%)
2020- present	HUGEN 2080: Statistical Genetics University of Pittsburgh	3 hours per week 3 credits 10 students	Co-instructor (25%- 33%)
2022- present	HUGEN 2020: Introduction to Population Genetics and Genetic Epidemiology University of Pittsburgh	3 hours per week 3 credits 60 students	Course director / instructor (100%)

#### Other Teaching

Year	Type of Teaching	Title
2006	Guest Facilitator	MS-1 Human Genetics, Department of Medicine, University of Pittsburgh "Problem-based learning: sickle cell disease"
2006	Guest Lecturer	HUGEN 2033: Quantitative Genetics, Department of Human Genetics, University of Pittsburgh "Case study: linkage analysis for hip bone mineral density"
2006	Guest Lecturer	BIOST 2055: Statistical Methods & Data Mining in Microarray Analysis, Department of Biostatistics, University of Pittsburgh "Missing value imputation in microarray data"

Year	Type of Teaching	Title
2007	Guest Lecturer	HUGEN 2022: 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	HUGEN 2022: Human Population Genetics, Department of Human Genetics, University of Pittsburgh “Hardy-Weinberg equilibrium”
2008	Guest Lecturer	HUGEN 2022: 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	HUGEN 2022: Human Population Genetics, Department of Human Genetics, University of Pittsburgh “Hardy-Weinberg equilibrium”
2008	Guest Lecturer	HUGEN 2022: 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	HUGEN 2029: 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>
2021-2022	Guest Lecturer	PUBLTH 100: Fundamentals of Public Health, University of Pittsburgh, “Introduction to Human Genetics”

## Mentorship

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

Student	Degree, Year	Type of Document and Title	Notes
Ekaterina Orlova	MS, April 2015	<ul style="list-style-type: none"> <li>MS Thesis: Putative regulatory function of GWAS-identified markers of susceptibility to acute appendicitis</li> <li>Chairman’s Award for Translational Research, Dept. of Human Genetics, University of Pittsburgh</li> </ul>	<ul style="list-style-type: none"> <li>co-advisor with Michael Morowitz</li> <li>doctoral program, University of Pittsburgh</li> </ul>

<b>Student</b>	<b>Degree, Year</b>	<b>Type of Document and Title</b>	<b>Notes</b>
Kelly Johnson	MS, April 2016	<ul style="list-style-type: none"> <li>• <b>Awards</b></li> <li>MS Thesis: Genome-wide association studies of childhood bone health</li> <li>• Russel Rule Rycheck Award for Public Health, Graduate School of Public Health, University of Pittsburgh</li> <li>• Outstanding Student Award, Graduate School of Public Health, University of Pittsburgh</li> </ul>	<ul style="list-style-type: none"> <li>• <b>Placement</b></li> <li>genetic counseling position</li> </ul>
Yunqi Li	MS, April 2018	<ul style="list-style-type: none"> <li>MS Thesis: “Epigenome-wide association study of recovery outcomes of traumatic brain injury patients”</li> <li>• William T. Green Award for Public Health Studies, Graduate School of Public Health, University of Pittsburgh</li> </ul>	<ul style="list-style-type: none"> <li>co-advisor with Daniel Weeks</li> <li>• doctoral program, University of Washington</li> </ul>
Amanda Everman	MS, April 2018	<ul style="list-style-type: none"> <li>MS Thesis: “Genome-wide association study and meta-analysis of missing teeth and functional dentition”</li> <li>• Dean’s Scholarship Award, Graduate School of Public Health, University of Pittsburgh</li> </ul>	<ul style="list-style-type: none"> <li>• Statistical Programmer/Scientific Affairs Analyst, PRA Health Sciences</li> </ul>
Julia Verbiar	MS, April 2018	<ul style="list-style-type: none"> <li>MS Thesis: “Genetics of the relationship between periodontitis and cardiovascular disease”</li> </ul>	<ul style="list-style-type: none"> <li>• genetic counseling position</li> </ul>
Yuqiao Zhou	PhD, Dec. 2018	<ul style="list-style-type: none"> <li>Dissertation: “Investigating the role of ER stress sensor IRE1<math>\alpha</math> in postnatal tooth development”</li> <li>• Dean’s Scholarship Award, School of Dental Medicine</li> <li>• American Association for Dental Research Pittsburgh Chapter Award</li> </ul>	<ul style="list-style-type: none"> <li>• post-doctoral associate, University of Pittsburgh</li> <li>• DDS program, University of Michigan</li> </ul>
Annie Infancia Arockiaraj	MS, May 2019	<ul style="list-style-type: none"> <li>MS Thesis: “Epigenome-wide association studies in aneurysmal subarachnoid hemorrhage patients”</li> </ul>	<ul style="list-style-type: none"> <li>co-advisor with Daniel Weeks</li> <li>• doctoral program, University of Pittsburgh</li> </ul>

<b>Student</b>	<b>Degree, Year</b>	<b>Type of Document and Title</b> • <b>Awards</b>	<b>Notes</b> • <b>Placement</b>
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 • First Place Poster Award, 2021 International Genetic Epidemiology Society	• post-doctoral fellow, Icahn School of Medicine at Mount Sinai • Scientist, GSK
Tianyu Zou	PhD, June 2022	Dissertation: "Investigating the genetics of dental caries incidence, development over time, and variability" • C.C. and Clara Li Award, Dept. of Human Genetic, University of Pittsburgh	• Scientist, Statistical Genetics, Beam Therapeutics
Kestrel Allawos	MPH, April 2022	MPH Essay: "Impact of Social Deprivation Index on Severe Traumatic Brain Injury Outcomes"	co-advisor with Yvette Conley
Tara Harmony	MPH, April 2022	MPH Essay: "Assessing the relationship between social deprivation and DNA methylation in a traumatic brain injury cohort"	co-advisor with Yvette Conley
Anna MacKinnon	MPH, in April 2023	MPH Essay: "Comparing Oral Health in Urban and Rural Children in North-Central Appalachia" • AADOCR platform presentation	
Ekaterina Orlova	PhD, April 2024	Dissertation: "Genome-wide association studies of early childhood caries"	
Dylan Maher	MS and PhD, in progress	MS Thesis: "Genetic Investigations of Cleft Lip with or without Cleft Palate: A Literature Review and Genome-wide Association Study in a Filipino Population" • C.C. and Clara Li Award, Dept. of Human Genetic, University of Pittsburgh • Fellow, Pittsburgh Craniofacial Sciences Training Program	
Kathryn K Ryan	MPH, in progress	MPH Thesis: TBD	

#### Service on Masters or Doctoral Committees

<b>Year</b>	<b>Student</b>	<b>Degree</b>	<b>Title of Dissertation/Essay</b>
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)

Year	Student	Degree	Title of Dissertation/Essay
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)
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)

<b>Year</b>	<b>Student</b>	<b>Degree</b>	<b>Title of Dissertation/Essay</b>
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)
2018-2021	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)
2019-2020	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)
2020-2021	Revella Raker	MS	Genetic literacy and the acquisition of clinical genetics knowledge in medical students (April 9, 2021)



Year	Student	Degree	Title of Dissertation/Essay
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 (Aug. 4, 2021)
2020-2022	Gillian Goobie	PhD	Environmental impacts of clinical outcomes and the epigenome in patients with interstitial lung disease (Oct. 19, 2022)
2021-2023	Yulong Bai	PhD	Elucidating the role of alternative polyadenylation in cancer by integrated transcriptomic analysis (Jan. 9, 2023)
2021-2022	Ellyn Dunbar	PhD	Psychiatric and Pain Risk Genes that May Worsen Quality of Life in Chronic Pancreatitis Patients (July 13, 2022)
2021-2022	Haley Soller	MS	The Acquisition of Clinical Genetics Knowledge in Medical Students (April 8, 2022)
2022	Laura Stackhouse	MS	<i>CREBRF</i> Missense Variant rs373863828 and Relative Grip Strength (April 13, 2022)
2022-2023	Rebecca Oberschmidt	MS	Correlating clinical findings with genetic testing results in patients with concern for connective tissue disorders - a retrospective chart review (March 31, 2023)
2022-2023	Lauren Spor	MPH	Cancer Risks for Carriers of Germline Pathogenic Variants in Xeroderma Pigmentosum Genes (April 2023)
2022-2023	Rhianna Ericson	MPH	Assessing Community Strengths and Needs within Rural Municipalities in Allegheny County, PA (April 2023)
2022-2023	Amanda Koloskee	MS	Effect of Exercise on DNA Methylation Age Acceleration in Breast Cancer Patients Undergoing Aromatase Inhibitor Therapy (April 11, 2023)
2022-2023	Alexis Cename	MS	A Case-Control Study Investigating the Association of the VMAT2 with a Diagnosis of Schizophrenia (April 18, 2023)
2022-2023	Nauman Ahmad	MS	Genome-wide association study of stature loss (Aug. 2, 2023)
2022-2023	Sandra Deslouches	MS	Evaluating the Genetic Variability of EDIL3 in Aneurysmal Subarachnoid Hemorrhage (Dec. 6, 2023)
2022-2024	Yidi Qin	PhD	Deriving Biological Meaning and Clinical Application for Pediatric Sepsis with Data-driven Analysis (April 5, 2024)
2023-2024	Caitlyn Sibiskie Kandel	MS	hEDS and HSD Patient Educational Materials and Resources: Patient Experiences and Needs, and the Availability and Accessibility of Information. (March 25, 2024)
2023-2024	Gabriela Slizewska	MS	Perceived Value of Non-Diagnostic Genetic Testing in Hypermobile Patients (April 4, 2024)
2023-	Priyamvada Guha Roy	PhD	TBD

#### Service on Comprehensive or Qualifying Examination Committees

Dates Served	Student	Type of Exam
June 1, 2012	Siddhartha Das	PhD Qualifying

<b>Dates Served</b>	<b>Student</b>	<b>Type of Exam</b>
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
June 9, 2021	Tianyu Zou	PhD Comprehensive (chair)
Dec. 13, 2021	Yulong Bai	PhD Comprehensive
Feb. 11, 2022	Laura Stackhouse	MS Comprehensive
March 16, 2022	Christopher Pombar	MS Comprehensive
May 24, 2022	Jie Sun	PhD Qualifying
July 15, 2022	Shuwei Liu	PhD Qualifying
Aug. 12, 2022	Nauman Ahmad	MS Comprehensive
Dec. 8, 2022	Amanda Koloskee	MS Comprehensive
March 3, 2023	Yidi Qin	PhD Comprehensive
March 9, 2023	Tabitha Banks-Tibbs	PhD Qualifying (chair)
March 24, 2023	Priyamvada Guha Roy	PhD Qualifying (chair)
April 20, 2023	Sandra Deslouches	MS Comprehensive (chair)
June 29, 2023	Megan Perez	PhD Qualifying
Aug. 15, 2023	Lauren Spor	PhD Qualifying
Oct. 11, 2023	Aditya Sriram	PhD Qualifying
April 3, 2024	Priyamvada Guha Roy	PhD Comprehensive
May 21, 2024	Dylan Maher	MS Comprehensive (chair)
June 20, 2024	Gracen Mitrick	PhD Qualifying

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

Year	Trainee	Position
		<ul style="list-style-type: none"> <li>Awards/Placement</li> </ul>
2015-2016	Jatinder Singh, PhD	Post-doctoral research associate (secondary mentor)
2019-2021	Yuqiao Zhou, PhD, DDS	Post-doctoral research associate <ul style="list-style-type: none"> <li>IADR Hatton Award Finalist (2020)</li> <li>placement: Internationally Trained Dentist Program DDS program, University of Michigan</li> </ul>
2019-2021	Chenxing Liu, PhD	Post-doctoral research associate
2022-	Zeynep Erdogan-Yildirim, MD, PhD	Post-doctoral fellow (co-mentored)
2023-	Noah Herrick, PhD	Post-doctoral research associate (co-mentored)

### Other Teaching and Training

Year	Teaching Activity	Description
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

Year	Teaching Activity	Description
2017	mentor	rotation project student Yuqiao Zhou
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
2019-2021	co-mentor	research mentor to medical student Kathleen Koesarie
2022-	co-mentor	graduate student researcher Xinyi Zhang (co-mentored with Jenna Carlson)

## Research and Training

### Grants and Contracts Received

#### Principal Investigator

Year	Grant and/or Contract Number and Title	Source	Annual Direct Costs	% Effort
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%
2022-2025	R01 DE032122 Investigating the role of genes, maternal exposures, and interactions on orofacial clefts	NIH/NIDCR	\$466,696	30%

#### Multiple Principal Investigator

Year	Grant and/or Contract Number and Title	Source	Annual Direct Costs	% Effort
2017-2021	X01 HG009878 Genomic Studies of Oral Health and Disease (PIs: Shaffer/Feingold)	NIH / NHGRI	\$0 (\$667,040 genotyping services)	-

Year	Grant and/or Contract Number and Title	Source	Annual Direct Costs	% Effort
2017-2020	R01 DE027023 The Genetic Architecture of Human Facial Morphology (PIs: Weinberg, Shaffer)	NIH/NIDCR	\$436,922	20%
2020-2025	R01 DE027023 The Genetic Architecture of Human Facial Morphology (PIs: Weinberg, Shaffer, Claes, Wysoka)	NIH/NIDCR	\$575,707	10%
2020-2025	X01 HG011437 Genetics of Orofacial Clefts, Sub-types, and Subclinical Phenotypes (PIs: Shaffer, Marazita)	NIH / NHGRI	\$0 (\$624,000 genotyping services)	-
2020-2023	R01 DE014899 Factors contributing to Oral Health Disparities in Appalachia (PIs: Marazita, McNeil, Foxman, Shaffer) *brought on as MPI after NOA	NIH / NIDCR	\$1,566,868	20%
2021-2023	X01 HG012156 Genetic markers of caries risk in diverse underserved children (PIs: Fontana, Shaffer)	NIH / NHGRI	\$0 (\$137,670 genotyping services)	-
2023-2024	X01 HD114124 Epigenomics of orofacial clefts (PIs: Shaffer, Marazita)	NIH / NHGRI	\$0 (\$1,670,000 in omics services)	-

#### 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 (PIs: 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, 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%

Year	Grant and/or Contract Number and Title	Source	Annual Direct Costs	% Effort
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, Feingold)	NIH common fund	\$0 (whole genome sequencing services)	-
2016-2018	R03 DE026469 Analysis of Whole Genome Sequence Data in Orofacial Clefts (PIs: Marazita, Feingold)	NIH / NIDCR	\$100,000	-
2018-2023	R01 CA221882 Epigenomics of Neurocognitive Function in Breast Cancer (PI: Conley) <b>PI of subaward</b>	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) <b>PI of subaward</b>	NIH/NNDS	\$472,560	4.5%
2022-2027	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%
2022-2027	R01-DE032319 Genomic Risk Variants in Orofacial Clefting: Discovery and Functional Validation (PIs: Marazita, Murray) <b>PI of subaward</b>	NIH/NIDCR	\$637,890	5%
2024-2029	U01-DE021412 Predicting Caries Risk in Underserved Children, from Toddlers to the School-Age Years, in Primary Healthcare Settings (PI: Fontana)	NIH/NIDCR	\$45,897	2.5%

#### Invited Lectureships and Major Seminars

Date	Title of Presentation	Venue
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.

<b>Date</b>	<b>Title of Presentation</b>	<b>Venue</b>
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
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**

<b>Date</b>	<b>Position</b>	<b>Description of Activity</b>
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

Date	Position	Description of Activity
2014-2015	consultant to Dr. Lina Moreno-Uribe, 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

### Refereed 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, Tylavsky 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.
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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.
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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 Zerfas, 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-Uribe 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-Uribe 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 Hallgrimsson, 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 Hallgrimsson, 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, Wasii 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, Wasii 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. Freida Blostein, Deesha Bhaumik, Elyse Davis, Elizabeth Salzman, Daniel W. McNeil, **John R. Shaffer**, Mary L. Marazita, Betsy Foxman. Early life assembly of the salivary microbiome is

- associated with maternal microbiome and future dental caries. (2022) *8<sup>th</sup> Conference on Beneficial Microbes*.
173. Franziska Wilke, Noah Herrick, Harold Matthews, Hanne Hoskens, **John R. Shaffer**, Seth M. Weinberg, Mark D. Shriver, Peter Claes, Susan Walsh. Exploring Regional Aspects of Facial Variation in a European Population. (2022) *International Society for Forensic Genetics 29th International Congress*.
  174. Meng Yuan, Sahin Naqvi, Hanne Hoskens, Ahmed Radwan, John R. Shaffer, Mark D. Shriver, Seth M. Weinberg, Susan Walsh, Stefan Sunaert, Hilde Peeters, Rose Bruffaerts, Joanna Wysocka, and Peter Claes. The genetic architecture of cortical brain shape and thickness (2022) *Belgium Society of Human Genetics 22<sup>nd</sup> Annual Meeting*.
  175. Seppe Goovaerts, Hanne Hoskens, Meng Yuan, Noah Herrick, Sahin Naqvi, Myoung Keun Lee, **John R Shaffer**, Susan Walsh, Mark D Shriver, Joanna Wysocka, Seth M Weinberg, Peter Claes. Multivariate GWAS of 3D cranial vault shape in multi-ethnic and admixed children. (2022) *American Society of Human Genetics Annual Meeting*.
  176. Tianyu Zou, Betsy Foxman, Daniel McNeil, Mary L. Marazita, **John R. Shaffer**. Genome-wide analyses of dental caries variability identifies genotype-by-environment interactions. (2022) *American Society of Human Genetics Annual Meeting*.
  177. Shuwei Liu, Dongjing Liu, Catherine M. Bender, Kirk I. Erickson, Susan M. Sereika, Yvette P. Conley, **John R. Shaffer**, Daniel E. Weeks. DNA methylation changes associated with cognitive function in post-surgery breast cancer patients. (2022) *American Society of Human Genetics Annual Meeting*.
  178. Ekaterina Orlova, Tom Dudding, Jonathan M. Chernus, Rasha N. Alotaibi, Simon Haworth, Richard J. Crout, Myoung Keun Lee, Nandita Mukhopadhyay, Eleanor Feingold, Steven M. Levy, Daniel W. McNeil, Betsy Foxman, Robert J. Weyant, Nicholas J. Timpson, Mary L. Marazita, **John R Shaffer**. Genome scans of early childhood caries implicate bitter taste receptors (2022) *American Society of Human Genetics Annual Meeting*.
  179. Simon Haworth, **John R. Shaffer**, A. Esberg, Pernilla Lundberg, Kimon Divaris, Mary L. Marazita, Ingegerd Johansson. Leveraging spatial patterns in the distribution of dental caries to resolve genetic heterogeneity. (2022) *American Society of Human Genetics Annual Meeting*.
  180. Anna H. MacKinnon, Yuqiao Zhou, Daniel W. McNeil, Richard J. Crout, Betsy Foxman, Mary L. Marazita, **John R. Shaffer**. Environment matters: rural-urban differences in oral health among Appalachian children. (2023) *American Association for Dental, Oral, and Craniofacial Research 52<sup>nd</sup> Annual Meeting and Exposition*.
  181. Rasha N. Alotaibi, **John R. Shaffer**, Daniel W. McNeil, Steven E. Reis, Betsy Foxman, Mary L. Marazita. (2023) Association of Salivary Flow Rate with Permanent Dentition Caries. *American Association for Dental, Oral, and Craniofacial Research 52<sup>nd</sup> Annual Meeting and Exposition*.
  182. Elyse Davis, Michael Fiebig, Betsy Salzman, Deesha Bhaumik, Freida Blostein, **John R. Shaffer**, Daniel W. McNeil, Mary L. Marazita, Betsy Foxman. Candida Colonization Interacts with Smoking to Modify Oral Bacteriome Composition (2023) *American Association for Dental, Oral, and Craniofacial Research 52<sup>nd</sup> Annual Meeting and Exposition*.
  183. Eshan Pua Schleif, Myoung Keun Lee, Neda Tahmasebifard, Imani Gilbert, Taylor Snodgrass, **John R. Shaffer**, Seth M. Weinberg, Jamie L. Perry. Sexual dimorphism in velopharyngeal anatomy of nine- and ten-year-old children. (2023) *American Cleft Palate-Craniofacial Association 80<sup>th</sup> Annual Meeting*.
  184. Divaris K, Papananou PN, Lundberg P, Haworth S, **Shaffer JR**, Moss KL, Preisser JS, Marazita ML, Johansson I, Beck JD. Leveraging Screening-Type Periodontal Probing Depth Data for Consortium-Wide Periodontitis Studies. (2023) *American Association for Dental, Oral, and Craniofacial Research 52<sup>nd</sup> Annual Meeting and Exposition*.

185. Meng Yuan, Seppe Goovaerts, Michiel Vanneste, Harold Matthews, Hanne Hoskens, Ophir D Klein, Richard Spritz, Benedikt Hallgrímsson, Susan Walsh, Mark D Shriver, Joanna Wysocka, **John R Shaffer**, Seth M Weinberg, Hilde Peeters, Peter Claes. Assessing univariate facial phenotyping approaches in GWAS. (2023) *Belgium Society of Human Genetics 23<sup>rd</sup> Annual Meeting*.
186. Seppe Goovaerts, Hanne Hoskens, Harold S Matthews, Noah Herrick, Dirk Vandermeulen, Meng Yuan, **John R Shaffer**, Mark D Shriver, Joanna Wysocka, Susan Walsh, Seth M Weinberg, Peter Claes. Joint multi-ancestry and admixed GWAS on 3D facial shape derived from MRI (2023) *European Society of Human Genetics Conference*.
187. Jamey T. Brumbaugh, **John R. Shaffer**, Betsy Foxman, Mary L., Marazita, Daniel W. McNeil. Maternal Depression and Infant Sleeping Arrangements. (2023) *Association for Behavioral and Cognitive Therapies 2023 Convention*.
188. Meng Yuan, Seppe Goovaerts, Hanne Hoskens, Ophir D. Klein, Richard A. Spritz, Benedikt Hallgrímsson, Stephen Richmond, Susan Walsh, Mark D. Shriver, **John R. Shaffer**, Seth M Weinberg, Hilde Peeters, Peter Claes. Data-driven trait heritability-based extraction of human facial phenotypes in genome-wide association studies. (2023) *American Society of Human Genetics 2023 Annual Meeting*.
189. Carolyn S. Harris, Cathy Bender, Bruce A. Cooper, Shuwei Liu, **John R. Shaffer**, Daniel E. Weeks, Yvette P. Conley. Area Deprivation and Epigenomics Influence Psychological Symptom Cluster Severity in Women with Breast Cancer. (2023) *International Society of Nurses in Genetics*.
190. Xinyi Zhang, Terri H. Beaty, Eleanor Feingold, Seth M. Weinberg, Mary L. Marazita, **John R. Shaffer**, Jenna C. Carlson. Reanalyzing legacy GWAS dataset using TOPMed imputation yields novel loci for orofacial clefting. (2023) *American Society of Human Genetics Annual Meeting*.
191. Dylan Maher, Nandita Mukhopadhyay, Sarah W. Curtis, Jenna Carlson, Eleanor Feingold, Seth M. Weinberg, Carmencita D. Padilla, Jeffrey C. Murray, Mary L. Marazita, Elizabeth J. Leslie, **John R. Shaffer**. Genome-wide Association Study of Orofacial Clefts in Filipinos Identifies Novel Locus. (2023) *American Society of Human Genetics Annual Meeting*.
192. Zan Koenig, Isaac Wong, Ying Wang, Lord J.J. Gowans, Mekonen Eshete, W. L. Adeyemo, Azeez A. Alade, Nandita Mukhopadhyay, Carmen J. Buxó-Martinez, Jacqueline T. Hecht, Lina M. Moreno Uribe, Carmencita D. Padilla, Fernando A. Poletta, George L. Wehby, Eleanor Feingold, **John R. Shaffer**, Elizabeth J. Leslie, Jeffrey C. Murray, Seth M. Weinberg, Azeez Butali, Mary L. Marazita, Harrison Brand, Alicia R. Martin. Elucidating genetic susceptibility to orofacial clefting within a multi-ancestry cohort. (2023) *American Society of Human Genetics Annual Meeting*.
193. Zeynep Erdogan-Yildirim, Nandita Mukhopadhyay, Jenna C Carlson, **John R Shaffer**, Seth M Weinberg, Mary L Marazita. Genome-wide investigation reveals significant interaction of *FURIN* with maternal vitamin intake on orofacial cleft risk. (2023) *American Society of Human Genetics Annual Meeting*.
194. Jonathan M. Chernus, Gracen E. Mitrick, Jasmien Roosenboom, Myoung Keun Lee, Azeez Butali, Carmen J. Buxó-Martinez, Kaare Christensen, Frederic W. B. Deleviannis, 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. Genome-wide meta-analysis of orbicularis oris muscle defects as a subclinical phenotype in orofacial clefting. (2023) *American Society of Human Genetics Annual Meeting*.
195. Meng Yuan, Seppe Goovaerts, Hanne Hoskens, Ophir D. Klein, Richard A. Spritz, Benedikt Hallgrímsson, Stephen Richmond, Susan Walsh, Mark D. Shriver, **John R. Shaffer**, Seth M Weinberg, Hilde Peeters, Peter Claes. Data-driven trait heritability-based extraction of human facial phenotypes in genome-wide association studies. (2023) *American Society of Human Genetics Annual Meeting*.

196. Seppe Goovaerts, Meng Yuan, Hanne Hoskens, Joanna Wysocka, Susan Walsh, Mark D Shriver, **John R Shaffer**, Seth M Weinberg, Peter Claes. GWAS conditioned by SNP-specific shape effects identifies 11 loci underlying shape covariation of the cerebral cortex and cranial vault. (2023) *American Society of Human Genetics Annual Meeting*.
197. Myoung Keun Lee, Ahmed M. El Sergani, Mary L. Marazita, **John R. Shaffer**, Seth M. Weinberg. GWAS of Torus Palatinus Reveals Several Suggestive Loci. (2023) *American Society of Human Genetics Annual Meeting*.
198. Shuwei Liu, Dongjing Liu, Catherine M. Bender, Kirk I. Erickson, Susan M. Sereika, Yvette P. Conley, **John R. Shaffer**, Daniel E. Weeks. Associations between DNA methylation and cognitive function in early-stage breast cancer patients. (2023) *American Society of Human Genetics Annual Meeting*.
199. Mary L. Marazita, Nandita Mukhopadhyay, **John R. Shaffer**. Association Study of Orofacial Cleft Risk Variants across All of Us Cancer Diagnoses. (2023) *All of Us Research Program's Extramural Program to Advance Research (EPAR) Virtual Symposium*.
200. Zeynep Erdogan-Yildirim, Nandita Mukhopadhyay, Jenna C Carlson, **John R Shaffer**, Seth M Weinberg, Mary L Marazita. Genome-wide investigation reveals possible interaction of *FURIN* with maternal vitamin intake on orofacial cleft risk. (2023) *Society for Craniofacial Genetics and Developmental Biology*.
201. Antonio J Cardoso, Joel Anderton, Stephanie Brandebura, Chelsea Carver, Youssef Gabrielael, Shivam Khairnar, **John R Shaffer**, Mary L Marazita, Seth M Weinberg, Ahmed M El Sergani for Characterizing Sexual Dimorphism in the Human Palate using Statistical Shape Analysis. (2024) *International Association for Dental Research/American Association for Dental, Oral, and Craniofacial Research/Canadian Association for Dental Research General Session & Exhibition*.
202. Youssef Gabrielael, Joel Anderton, Stephanie Brandebura, Antonio J Cardoso, Chelsea Carver, Shivam Khairnar, **John R Shaffer**, Mary L Marazita, Seth M Weinberg, Ahmed M El Sergani for Statistical Shape Analysis identifies palatal endophenotypes in unaffected parents of children affected with nonsyndromic orofacial clefting. (2024) *International Association for Dental, Oral, and Craniofacial Research/American Association for Dental Research/Canadian Association for Dental Research General Session & Exhibition*.
203. Shivam Khairnar, Joel Anderton, Stephanie Brandebura, Antonio J Cardoso, Chelsea Carver, Youssef Gabrielael, **John R Shaffer**, Mary L Marazita, Seth M Weinberg, Ahmed M El Sergani for Exploring Ancestral Differences in the Human Palate using Geometric Morphometric Analysis. (2024) *International Association for Dental Research/American Association for Dental, Oral, and Craniofacial Research/Canadian Association for Dental Research General Session & Exhibition*.
204. D. Baxter, M. Bezamat, N. Mukhopadhyay, A. Saeed, L. Liu, L. de las Fuentes, B. Foxman, **J. R. Shaffer**, D. W. McNeil, S. Reis, M. L. Marazita. Genome-wide Association Studies in Oral and Atherosclerotic Cardiovascular Diseases (2024). *International Association for Dental Research/American Association for Dental, Oral, and Craniofacial Research/Canadian Association for Dental Research General Session & Exhibition*.
205. Meng Yuan, Seppe Goovaerts, Michiel Vanneste, Myoung Keun Lee, Susan Walsh, Mark D Shriver, **John R Shaffer**, Seth M Weinberg, Hilde Peeters, Peter Claes. Commingling analysis combined phenotyping for identification of 3D facial traits with potential major gene effects. (2024) *Belgium Society of Human Genetics 24<sup>th</sup> Annual Meeting*.
206. Seppe Goovaerts, Hanne Hoskens, Noah Herrick, Meng Yuan, Mark D Shriver, **John R Shaffer**, Susan Walsh, Seth M Weinberg, Peter Claes. Enhanced insights into the genetic architecture of 3D cranial vault morphology using pleiotropy-informed conditional GWAS (2024) *Belgium Society of Human Genetics 24<sup>th</sup> Annual Meeting*.



207. Alba Sanchis-Juan, Sarah Curtis, Eren Shin, Arthur Lee, Kelsey Robinson, Azeez A. Alade, Xuefang Zhao, Jack Fu, Kimberly-Diaz-Perez, Lord J. J. Gowans, Mekonen A. Eshete, Wasu L. Adeyemo, Carmen J. Buxó-Martinez, Carmencita D. Padilla, Eleanor Feingold, Fernando A. Poletta, George L. Wehby, Jacqueline T. Hecht, **John R. Shaffer**, Lina M. Moreno Uribe, Nandita Mukhopadhyay, Seth M. Weinberg, Jeffrey C. Murray, Terri H. Beaty, Azeez Butali, Michael Talkowski, Mary L. Marazita, Elizabeth J. Leslie, Harrison Brand. Application of Integrative Analyses to Decipher the Genetic Architecture of Orofacial Clefting. (2024) *Society for Birth Defects Research and Prevention 2024 Annual Meeting*.
208. Noah Herrick, Nandita Mukhopadhyay, Zeynep Erdogan-Yildirim, Myoung Keun Lee, Xinyi Zhang, Jenna C. Carlson, Seth M. Weinberg, Mary L. Marazita, **John R. Shaffer**. Insights of local ancestry in an orofacial cleft genome-wide association study in a Filipino cohort. (2024) *Society for Birth Defects Research and Prevention 2024 Annual Meeting*.
209. Gabriela Slizewska, Christine D. Munro, Mylynda B. Massart, **John R. Shaffer**, Natasha K. Robin Berman. Hypermobile Patient Experiences with Negative Genetic Testing Results. (2024) Pennsylvania Association of Genetic Counselors Annual Conference.
210. Meng Yuan, Seppe Goovaerts, Myoung Keun Lee, Hanne Hoskens, Stephen Richmond, Susan Walsh, Mark D. Shriver, **John R. Shaffer**, Mary L. Marazita, Seth M. Weinberg, Hilde Peeters, Peter Claes. Data-driven phenotyping to optimize human facial traits in GWAS and WES. (2024) *American Society of Human Genetics Annual Meeting*.
211. Nandita Mukhopadhyay, Alexis Cennamo, **John R. Shaffer**, Mary L. Marazita. Association of orofacial cleft risk loci with elevated risk of prostate cancer. (2024) *American Society of Human Genetics Annual Meeting*.
212. Xinyi Zhang, Zeynep Erdogan-Yildirim, Terri H. Beaty, Azeez Butali, Carmen J Buxó, Lord J.J. Gowans, Jacqueline T Hecht, Lina Moreno, Jeffrey C Murray, Ieda M Orioli, Carmencita Padilla, Alexandre R Vieira, George L Wehby, Eleanor Feingold, Elizabeth J. Leslie, Seth M. Weinberg, Mary L. Marazita, **John R. Shaffer**, Jenna C. Carlson. Novel contributors to orofacial cleft risk identified at 7q33 and 10q24.32 through multi-ancestry meta-analysis. (2024) *American Society of Human Genetics Annual Meeting*.
213. Nehir E. Kurtas, Alba Sanchis-Juan, Sarah Curtis, Vahid Jalili, Ben Weisburd, Arthur Lee, Kelsey Robinson, Azeez A. Alade, Xuefang Zhao, Jack Fu, Kimberly-Diaz-Perez, Lord J. J. Gowans, Mekonen A. Eshete, Wasu L. Adeyemo, Carmen J. Buxó-Martinez, Carmencita D. Padilla, Fernando A. Poletta, George L. Wehby, Jacqueline T. Hecht, **John R. Shaffer**, Lina M. Moreno Uribe, Nandita Mukhopadhyay, Seth M. Weinberg, Jeffrey C. Murray, Terri H. Beaty, Azeez Butali, Michael Talkowski, Mary L. Marazita, Elizabeth J. Leslie, Harrison Brand. A comprehensive analysis of structural variations in 2,733 Orofacial Clefts (OFC) trios reveals novel genetic drivers. (2024) *American Society of Human Genetics Annual Meeting*.
214. Kathryn K. Ryan, **John R. Shaffer**, Nandita Mukhopadhyay, Emily Yanca, George J. Eckert, Jie Ren, Peter F. Castelluccio, Barry P. Katz, Anderson Hara, Martha Ann Keels, Steven Levy, Mary L. Marazita, Margherita Fontana. Genome-wide association study of childhood dental caries in a multi-ethnic sample. (2024) *American Society of Human Genetics Annual Meeting*.
215. Zeynep Erdogan-Yildirim, Jenna C Carlson, Nandita Mukhopadhyay, Seth M Weinberg, Mary L Marazita, **John R Shaffer**. Understanding the role of gene-environment interactions in CL/P etiology by the example of identified potential GxE loci. (2024) *American Society of Human Genetics Annual Meeting*.
216. Jay Devine, Seppe Goovaerts, Alex Pugner, Nina Claessens, Meng Yuan, **John R. Shaffer**, Susan Walsh, Mark D. Shriver, Seth M. Weinberg, Joanna Wysocka, Peter Claes. Revealing the developmental-genetic basis for the human cranial vault. (2024) *American Society of Human Genetics Annual Meeting*.

217. Noah Herrick, Seppe Goovaerts, Myoung Keun Lee, Mary L Marazita, Peter Claes, Seth M Weinberg, **John R Shaffer**. Identifying loci with pleiotropic effects on orofacial clefts and facial morphology using conditional false discovery rate analyses. (2024) *American Society of Human Genetics Annual Meeting*.

### 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*.

### Other Publications: 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

<b>Year</b>	<b>Committee</b>	<b>Position</b>
2007	Strategic Planning Committee	appointed member
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-present	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 (EPCC)	elected representative
2018	Internal Scientific Grant Review, Department of Epidemiology	<i>ad hoc</i> grant reviewer
2020-2021	Educational Policies and Curriculum Committee (EPCC)	elected chair
2020-2021	Graduate School of Public Health Council	member
2020-2021	Faculty Senate Executive Committee	member
2021-2022	Search Committee, Chair of the Department of Biostatistics, School of Public Health	appointed member
2021-present	Social justice, equity, diversity, and inclusion (JEDI) Council Culture Working Group, School of Public Health	member
2022-present	Search Committee, Chair of the Department of Human Genetics, School of Public Health	co-chair (2022- 2023), chair (2023)
2022-present	Bachelor of Science in Public Health (BSPH) Education Committee (BEC)	appointed member
2023-2024	Search Committee, BSPH teaching faculty, School of Public Health	appointed member
2023-2024	Accreditation Self-Study Committee	appointed 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

<b>Year</b>	<b>Committee</b>	<b>Position</b>
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### Service to Field of Scholarship

### Editorial Boards, Editorships

<b>Date</b>	<b>Position</b>	<b>Journal</b>
2014-present	Associate Editor	BMC Oral Health
2017-2018	Topic Editor	Frontiers in Genetics

### Manuscript and Other Document/Publication Review

<b>Year</b>	<b>Journal Title</b>
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
2012-2024	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
2022	Cleft Palate Craniofacial Journal

### 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
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 CIDR 2022/05 (01) M National Human Genome Research Institute Center for Inherited Disease Research Access Committee
Feb. 2022	<i>ad hoc</i> member	Department of Defense (DOD), Congressionally Directed Medical Research Programs (CDMRP), Traumatic Brain Injury and Psychological Health Research Program (TBIPHRP), Focused Program – Detection, Diagnosis, and Prognosis (FP-DDP) review panel.
May 2022	<i>ad hoc</i> member	NIH Study Section, Scientific Review Group ZHG1 HGR-T (02) National Human Genome Research Institute Special Emphasis Panel Gabriella Miller Kids First
July 2023	permanent member	NIH Study Section, Scientific Review Group CIDR 2023/07 (01) National Human Genome Research Institute Center for Inherited Disease Research Access Committee
Nov. 2023	permanent member	NIH Study Section, Scientific Review Group CIDR 2024/1 (01) 1 National Human Genome Research Institute Center for Inherited Disease Research Access Committee
Jan. 2024	permanent member	NIH Study Section, Scientific Review Group CIDR Special Review Session; National Human Genome Research Institute Center for Inherited Disease Research Access Committee
Jan. 2024	external reviewer	evaluated a promotion dossier for the Committee on Appointments and Promotions and the Advisory Board, Bloomberg School of Public Health, Johns Hopkins University

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

Year	Position	Organization
2010	Platform Moderator	American Society of Human Genetics
2021-present	Representative	Association of Schools and Programs of Public Health, PhD Council
2023-2024	Session organizer and co-chair	“Genetic and Environmental Factors Driving Facial Morphology and Dysmorphology”, Society for Birth Defects Research and Prevention, Annual Meeting
2023-2024	Local organizing committee member	2024 Statistical Genomics and Genetics (SGG) Conference, American Statistical Association (ASA)