

Elizabeth J Leslie

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/5140985/publications.pdf>

Version: 2024-02-01

75
papers

3,680
citations

201385

27
h-index

149479

56
g-index

93
all docs

93
docs citations

93
times ranked

3294
citing authors

#	ARTICLE	IF	CITATIONS
1	A genome-wide association study of cleft lip with and without cleft palate identifies risk variants near MAFB and ABCA4. <i>Nature Genetics</i> , 2010, 42, 525-529.	9.4	518
2	Genetics of cleft lip and cleft palate. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2013, 163, 246-258.	0.7	336
3	Genome-wide mapping of global-to-local genetic effects on human facial shape. <i>Nature Genetics</i> , 2018, 50, 414-423.	9.4	205
4	Dominant Mutations in GRHL3 Cause Van der Woude Syndrome and Disrupt Oral Periderm Development. <i>American Journal of Human Genetics</i> , 2014, 94, 23-32.	2.6	195
5	A multi-ethnic genome-wide association study identifies novel loci for non-syndromic cleft lip with or without cleft palate on 2p24.2, 17q23 and 19q13. <i>Human Molecular Genetics</i> , 2016, 25, ddw104.	1.4	163
6	Genetic factors influencing risk to orofacial clefts: today's challenges and tomorrow's opportunities. <i>F1000Research</i> , 2016, 5, 2800.	0.8	155
7	Identification of Functional Variants for Cleft Lip with or without Cleft Palate in or near PAX7, FGFR2, and NOG by Targeted Sequencing of GWAS Loci. <i>American Journal of Human Genetics</i> , 2015, 96, 397-411.	2.6	150
8	A Genome-wide Association Study of Nonsyndromic Cleft Palate Identifies an Etiologic Missense Variant in GRHL3. <i>American Journal of Human Genetics</i> , 2016, 98, 744-754.	2.6	146
9	Genome-Wide Association Study Reveals Multiple Loci Influencing Normal Human Facial Morphology. <i>PLoS Genetics</i> , 2016, 12, e1006149.	1.5	140
10	Genome-wide meta-analyses of nonsyndromic orofacial clefts identify novel associations between FOXE1 and all orofacial clefts, and TP63 and cleft lip with or without cleft palate. <i>Human Genetics</i> , 2017, 136, 275-286.	1.8	139
11	Expression and mutation analyses implicate ARHGAP29 as the etiologic gene for the cleft lip with or without cleft palate locus identified by genome-wide association on chromosome 1p22. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2012, 94, 934-942.	1.6	85
12	Genome-Wide Association Studies in Dogs and Humans Identify ADAMTS20 as a Risk Variant for Cleft Lip and Palate. <i>PLoS Genetics</i> , 2015, 11, e1005059.	1.5	82
13	Genome-wide association study of facial morphology reveals novel associations with FREM1 and PARK2. <i>PLoS ONE</i> , 2017, 12, e0176566.	1.1	68
14	Replication of Genome Wide Association Identified Candidate Genes Confirm the Role of Common and Rare Variants in <i>PAX7</i> and <i>VAX1</i> in the Etiology of Nonsyndromic CL(P). <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 965-972.	0.7	60
15	An etiologic regulatory mutation in IRF6 with loss- and gain-of-function effects. <i>Human Molecular Genetics</i> , 2014, 23, 2711-2720.	1.4	55
16	Sonic Hedgehog regulation of <i>Foxf2</i> promotes cranial neural crest mesenchyme proliferation and is disrupted in cleft lip morphogenesis. <i>Development (Cambridge)</i> , 2017, 144, 2082-2091.	1.2	55
17	Rare and Common Variants Conferring Risk of Tooth Agenesis. <i>Journal of Dental Research</i> , 2018, 97, 515-522.	2.5	52
18	<i>IRF6</i> mutation screening in non-syndromic orofacial clefting: analysis of 1521 families. <i>Clinical Genetics</i> , 2016, 90, 28-34.	1.0	50

#	ARTICLE	IF	CITATIONS
19	A LINE-1 Insertion in DLX6 Is Responsible for Cleft Palate and Mandibular Abnormalities in a Canine Model of Pierre Robin Sequence. <i>PLoS Genetics</i> , 2014, 10, e1004257.	1.5	49
20	Irf6 directly regulates Klf17 in zebrafish periderm and Klf4 in murine oral epithelium, and dominant-negative KLF4 variants are present in patients with cleft lip and palate. <i>Human Molecular Genetics</i> , 2016, 25, 766-776.	1.4	48
21	Identification of common non-coding variants at 1p22 that are functional for non-syndromic orofacial clefting. <i>Nature Communications</i> , 2017, 8, 14759.	5.8	48
22	Genome-wide Enrichment of De Novo Coding Mutations in Orofacial Cleft Trios. <i>American Journal of Human Genetics</i> , 2020, 107, 124-136.	2.6	48
23	Comparative analysis of IRF6 variants in families with Van der Woude syndrome and popliteal pterygium syndrome using public whole-exome databases. <i>Genetics in Medicine</i> , 2013, 15, 338-344.	1.1	47
24	Identifying Genetic Sources of Phenotypic Heterogeneity in Orofacial Clefts by Targeted Sequencing. <i>Birth Defects Research</i> , 2017, 109, 1030-1038.	0.8	41
25	Expanding the genetic and phenotypic spectrum of popliteal pterygium disorders. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 545-552.	0.7	38
26	Association studies of low-frequency coding variants in nonsyndromic cleft lip with or without cleft palate. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1531-1538.	0.7	36
27	A systematic genetic analysis and visualization of phenotypic heterogeneity among orofacial cleft GWAS signals. <i>Genetic Epidemiology</i> , 2019, 43, 704-716.	0.6	36
28	Evaluating rare coding variants as contributing causes to non-syndromic cleft lip and palate. <i>Clinical Genetics</i> , 2013, 84, 496-500.	1.0	34
29	Craniofacial genetics: Where have we been and where are we going?. <i>PLoS Genetics</i> , 2018, 14, e1007438.	1.5	32
30	Deep phenotyping in 3q29 deletion syndrome: recommendations for clinical care. <i>Genetics in Medicine</i> , 2021, 23, 872-880.	1.1	32
31	Replication of 13q31.1 association in nonsyndromic cleft lip with cleft palate in Europeans. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1054-1060.	0.7	31
32	Caries Experience Differs between Females and Males across Age Groups in Northern Appalachia. <i>International Journal of Dentistry</i> , 2015, 2015, 1-8.	0.5	30
33	The TFAP2A-IRF6-GRHL3 genetic pathway is conserved in neurulation. <i>Human Molecular Genetics</i> , 2019, 28, 1726-1737.	1.4	30
34	Multiethnic GWAS Reveals Polygenic Architecture of Earlobe Attachment. <i>American Journal of Human Genetics</i> , 2017, 101, 913-924.	2.6	29
35	FaceBase 3: analytical tools and FAIR resources for craniofacial and dental research. <i>Development (Cambridge)</i> , 2020, 147, .	1.2	25
36	Evidence for SNP-SNP interaction identified through targeted sequencing of cleft case-parent trios. <i>Genetic Epidemiology</i> , 2017, 41, 244-250.	0.6	24

#	ARTICLE	IF	CITATIONS
37	Identification of 16q21 as a modifier of nonsyndromic orofacial cleft phenotypes. <i>Genetic Epidemiology</i> , 2017, 41, 887-897.	0.6	24
38	Analysis of zebrafish periderm enhancers facilitates identification of a regulatory variant near human KRT8/18. <i>ELife</i> , 2020, 9, .	2.8	23
39	Search for genetic modifiers of IRF6 and genotype-phenotype correlations in Van der Woude and popliteal pterygium syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2535-2544.	0.7	21
40	Whole genome sequencing of orofacial cleft trios from the Gabriella Miller Kids First Pediatric Research Consortium identifies a new locus on chromosome 21. <i>Human Genetics</i> , 2020, 139, 215-226.	1.8	19
41	Association of low-frequency genetic variants in regulatory regions with nonsyndromic orofacial clefts. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 467-474.	0.7	18
42	Pleiotropy method reveals genetic overlap between orofacial clefts at multiple novel loci from GWAS of multi-ethnic trios. <i>PLoS Genetics</i> , 2021, 17, e1009584.	1.5	18
43	Genome-Wide Association Study of Non-syndromic Orofacial Clefts in a Multiethnic Sample of Families and Controls Identifies Novel Regions. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 621482.	1.8	16
44	A novel hereditary spastic paraplegia with dystonia linked to chromosome 2q24-q31. <i>Movement Disorders</i> , 2009, 24, 364-370.	2.2	15
45	Genome-wide interaction studies identify sex-specific risk alleles for nonsyndromic orofacial clefts. <i>Genetic Epidemiology</i> , 2018, 42, 664-672.	0.6	15
46	Presence of Epilepsy-Associated Variants in Large Exome Databases. <i>Journal of Neurogenetics</i> , 2013, 27, 1-4.	0.6	14
47	<i>FAT4</i> identified as a potential modifier of orofacial cleft laterality. <i>Genetic Epidemiology</i> , 2021, 45, 721-735.	0.6	14
48	Relationship of Genetic Variants With Procedural Pain, Anxiety, and Distress in Children. <i>Biological Research for Nursing</i> , 2017, 19, 339-349.	1.0	13
49	Craniofacial features of 3q29 deletion syndrome: Application of next-generation phenotyping technology. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2094-2101.	0.7	13
50	Genetic models and approaches to study orofacial clefts. <i>Oral Diseases</i> , 2022, 28, 1327-1338.	1.5	13
51	Convergent and distributed effects of the 3q29 deletion on the human neural transcriptome. <i>Translational Psychiatry</i> , 2021, 11, 357.	2.4	12
52	Hypertelorism and Orofacial Clefting Revisited: An Anthropometric Investigation. <i>Cleft Palate-Craniofacial Journal</i> , 2017, 54, 631-638.	0.5	11
53	Exploring Subclinical Phenotypic Features in Twin Pairs Discordant for Cleft Lip and Palate. <i>Cleft Palate-Craniofacial Journal</i> , 2017, 54, 90-93.	0.5	10
54	A Novel Variant of <i>ATP5MC3</i> Associated with Both Dystonia and Spastic Paraplegia. <i>Movement Disorders</i> , 2022, 37, 375-383.	2.2	10

#	ARTICLE	IF	CITATIONS
55	The PAX1 locus at 20p11 is a potential genetic modifier for bilateral cleft lip. Human Genetics and Genomics Advances, 2021, 2, 100025.	1.0	9
56	GWAS reveals loci associated with velopharyngeal dysfunction. Scientific Reports, 2018, 8, 8470.	1.6	8
57	Integrative approaches generate insights into the architecture of non-syndromic cleft lip ± cleft palate. Human Genetics and Genomics Advances, 2021, 2, 100038.	1.0	8
58	Testing the face shape hypothesis in twins discordant for nonsyndromic orofacial clefting. American Journal of Medical Genetics, Part A, 2017, 173, 2886-2892.	0.7	7
59	Genetics of Orofacial Cleft Birth Defects. Current Genetic Medicine Reports, 2015, 3, 118-126.	1.9	6
60	Ear Infection in Isolated Cleft Lip: Etiological Implications. Cleft Palate-Craniofacial Journal, 2017, 54, 189-192.	0.5	6
61	Individuals with nonsyndromic orofacial clefts have increased asymmetry of fingerprint patterns. PLoS ONE, 2020, 15, e0230534.	1.1	6
62	The Effect of Ethylene Glycol, Glycine Betaine, and Urea on Lysozyme Thermal Stability. Journal of Chemical Education, 2010, 87, 1393-1395.	1.1	5
63	Genome-wide association study of multiethnic nonsyndromic orofacial cleft families identifies novel loci specific to family and phenotypic subtypes. Genetic Epidemiology, 2022, , .	0.6	4
64	Embracing human genetics: a primer for developmental biologists. Development (Cambridge), 2020, 147, .	1.2	3
65	Genome-wide Interaction Study Implicates VGLL2 and Alcohol Exposure and PRL and Smoking in Orofacial Cleft Risk. Frontiers in Cell and Developmental Biology, 2022, 10, 621261.	1.8	3
66	Detection of de novo copy number deletions from targeted sequencing of trios. Bioinformatics, 2019, 35, 571-578.	1.8	2
67	Detecting Gene-Environment Interaction for Maternal Exposures Using Case-Parent Trios Ascertained Through a Case With Non-Syndromic Orofacial Cleft. Frontiers in Cell and Developmental Biology, 2021, 9, 621018.	1.8	2
68	Efficient estimation of indirect effects in case-control studies using a unified likelihood framework. Statistics in Medicine, 2022, 41, 2879-2893.	0.8	2
69	Feasibility of Social Media Recruitment for Orofacial Cleft Genetic Research. Cleft Palate-Craniofacial Journal, 2022, 59, 701-707.	0.5	1
70	Genetic Variants and the Cortisol Response in Children: An Exploratory Study. Biological Research for Nursing, 2019, 21, 157-165.	1.0	1
71	Heritability Analysis in Twins Indicates a Genetic Basis for Velopharyngeal Morphology. Cleft Palate-Craniofacial Journal, 2022, 59, 1340-1345.	0.5	1
72	Leveraging Family History in Case-Control Analyses of Rare Variation. Genetics, 2020, 214, 295-303.	1.2	0

#	ARTICLE	IF	CITATIONS
73	Identification of a non-coding SNP associated with risk for non-syndromic orofacial clefting with allele-specific effects on IRF6 expression in vitro. FASEB Journal, 2021, 35, .	0.2	0
74	Developmental Anomalies “ Clefts. , 2015, , 91-116.		0
75	Identifying Genetic Sources of Phenotypic Heterogeneity in Orofacial Clefts by Targeted Sequencing. Birth Defects Research, 2017, , .	0.8	0