Elizabeth J Leslie

List of Publications by Year
in descending order

75 papers	3,680 citations	201674 27 h-index	149698 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. Nature Genetics, 2010, 42, 525-529.	21.4	518
2	Genetics of cleft lip and cleft palate. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 246-258.	1.6	336
3	Genome-wide mapping of global-to-local genetic effects on human facial shape. Nature Genetics, 2018, 50, 414-423.	21.4	205
4	Dominant Mutations in GRHL3 Cause Van der Woude Syndrome and Disrupt Oral Periderm Development. American Journal of Human Genetics, 2014, 94, 23-32.	6.2	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. Human Molecular Genetics, 2016, 25, ddw104.	2.9	163
6	Genetic factors influencing risk to orofacial clefts: todayâ€™s challenges and tomorrowâ€™s opportunities. F1000Research, 2016, 5, 2800.	1.6	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. American Journal of Human Genetics, 2015, 96, 397-411.	6.2	150
8	A Genome-wide Association Study of Nonsyndromic Cleft Palate Identifies an Etiologic Missense Variant in GRHL3. American Journal of Human Genetics, 2016, 98, 744-754.	6.2	146
9	Genome-Wide Association Study Reveals Multiple Loci Influencing Normal Human Facial Morphology. PLoS Genetics, 2016, 12, e1006149.	3.5	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. Human Genetics, 2017, 136, 275-286.	3.8	139
11	Expression and mutation analyses implicate ARHGAP29 as the etiologic gene for the cleft lip with or without cleft palate locus identified by genomeâ€wide association on chromosome 1p22. Birth Defects Research Part A: Clinical and Molecular Teratology, 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. PLoS Genetics, 2015, 11, e1005059.	3.5	82
13	Genome-wide association study of facial morphology reveals novel associations with FREM1 and PARK2. PLoS ONE, 2017, 12, e0176566.	2.5	68
14	Replication of Genome Wide Association Identified Candidate Genes Confirm the Role of Common and Rare Variants in <scp>PAX</scp>7 and <scp>VAX</scp>1 in the Etiology of Nonsyndromic CL(P). American Journal of Medical Genetics, Part A, 2013, 161, 965-972.	1.2	60
15	An etiologic regulatory mutation in IRF6 with loss- and gain-of-function effects. Human Molecular Genetics, 2014, 23, 2711-2720.	2.9	55
16	Sonic Hedgehog regulation of Foxf2 promotes cranial neural crest mesenchyme proliferation and is disrupted in cleft lip morphogenesis. Development (Cambridge), 2017, 144, 2082-2091.	2.5	55
17	Rare and Common Variants Conferring Risk of Tooth Agenesis. Journal of Dental Research, 2018, 97, 515-522.	5.2	52
18	IRF6 mutation screening in nonâ€syndromic orofacial clefting: analysis of 1521 families. Clinical Genetics, 2016, 90, 28-34.	2.0	50

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19	A LINE-1 Insertion in DLX6 Is Responsible for Cleft Palate and Mandibular Abnormalities in a Canine Model of Pierre Robin Sequence. PLoS Genetics, 2014, 10, e1004257.	3.5	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. Human Molecular Genetics, 2016, 25, 766-776.	2.9	48
21	Identification of common non-coding variants at 1p22 that are functional for non-syndromic orofacial clefting. Nature Communications, 2017, 8, 14759.	12.8	48
22	Genome-wide Enrichment of De Novo Coding Mutations in Orofacial Cleft Trios. American Journal of Human Genetics, 2020, 107, 124-136.	6.2	48
23	Comparative analysis of IRF6 variants in families with Van der Woude syndrome and popliteal pterygium syndrome using public whole-exome databases. Genetics in Medicine, 2013, 15, 338-344.	2.4	47
24	Identifying Genetic Sources of Phenotypic Heterogeneity in Orofacial Clefts by Targeted Sequencing. Birth Defects Research, 2017, 109, 1030-1038.	1.5	41
25	Expanding the genetic and phenotypic spectrum of popliteal pterygium disorders. American Journal of Medical Genetics, Part A, 2015, 167, 545-552.	1.2	38
26	Association studies of lowâ€frequency coding variants in nonsyndromic cleft lip with or without cleft palate. American Journal of Medical Genetics, Part A, 2017, 173, 1531-1538.	1.2	36
27	A systematic genetic analysis and visualization of phenotypic heterogeneity among orofacial cleft GWAS signals. Genetic Epidemiology, 2019, 43, 704-716.	1.3	36
28	Evaluating rare coding variants as contributing causes to nonâ€syndromic cleft lip and palate. Clinical Genetics, 2013, 84, 496-500.	2.0	34
29	Craniofacial genetics: Where have we been and where are we going?. PLoS Genetics, 2018, 14, e1007438.	3.5	32
30	Deep phenotyping in 3q29 deletion syndrome: recommendations for clinical care. Genetics in Medicine, 2021, 23, 872-880.	2.4	32
31	Replication of 13q31.1 association in nonsyndromic cleft lip with cleft palate in Europeans. American Journal of Medical Genetics, Part A, 2015, 167, 1054-1060.	1.2	31
32	Caries Experience Differs between Females and Males across Age Groups in Northern Appalachia. International Journal of Dentistry, 2015, 2015, 1-8.	1.5	30
33	The TFAP2Aâ€“IRF6â€“GRHL3 genetic pathway is conserved in neurulation. Human Molecular Genetics, 2019, 28, 1726-1737.	2.9	30
34	Multiethnic GWAS Reveals Polygenic Architecture of Earlobe Attachment. American Journal of Human Genetics, 2017, 101, 913-924.	6.2	29
35	FaceBase 3: analytical tools and FAIR resources for craniofacial and dental research. Development (Cambridge), 2020, 147, .	2.5	25
36	Evidence for SNP-SNP interaction identified through targeted sequencing of cleft case-parent trios. Genetic Epidemiology, 2017, 41, 244-250.	1.3	24

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37	Identification of 16q21 as a modifier of nonsyndromic orofacial cleft phenotypes. Genetic Epidemiology, 2017, 41, 887-897.	1.3	24
38	Analysis of zebrafish periderm enhancers facilitates identification of a regulatory variant near human KRT8/18. ELife, 2020, 9, .	6.0	23
39	Search for genetic modifiers of IRF6 and genotypeâ€“phenotype correlations in Van der Woude and popliteal pterygium syndromes. American Journal of Medical Genetics, Part A, 2013, 161, 2535-2544.	1.2	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. Human Genetics, 2020, 139, 215-226.	3.8	19
41	Association of lowâ€frequency genetic variants in regulatory regions with nonsyndromic orofacial clefts. American Journal of Medical Genetics, Part A, 2019, 179, 467-474.	1.2	18
42	Pleiotropy method reveals genetic overlap between orofacial clefts at multiple novel loci from GWAS of multi-ethnic trios. PLoS Genetics, 2021, 17, e1009584.	3.5	18
43	Genome-Wide Association Study of Non-syndromic Orofacial Clefts in a Multiethnic Sample of Families and Controls Identifies Novel Regions. Frontiers in Cell and Developmental Biology, 2021, 9, 621482.	3.7	16
44	A novel hereditary spastic paraplegia with dystonia linked to chromosome 2q24â€2q31. Movement Disorders, 2009, 24, 364-370.	3.9	15
45	Genomeâ€wide interaction studies identify sexâ€specific risk alleles for nonsyndromic orofacial clefts. Genetic Epidemiology, 2018, 42, 664-672.	1.3	15
46	Presence of Epilepsy-Associated Variants in Large Exome Databases. Journal of Neurogenetics, 2013, 27, 1-4.	1.4	14
47	FAT4 identified as a potential modifier of orofacial cleft laterality. Genetic Epidemiology, 2021, 45, 721-735.	1.3	14
48	Relationship of Genetic Variants With Procedural Pain, Anxiety, and Distress in Children. Biological Research for Nursing, 2017, 19, 339-349.	1.9	13
49	Craniofacial features of 3q29 deletion syndrome: Application of nextâ€generation phenotyping technology. American Journal of Medical Genetics, Part A, 2021, 185, 2094-2101.	1.2	13
50	Genetic models and approaches to study orofacial clefts. Oral Diseases, 2022, 28, 1327-1338.	3.0	13
51	Convergent and distributed effects of the 3q29 deletion on the human neural transcriptome. Translational Psychiatry, 2021, 11, 357.	4.8	12
52	Hypertelorism and Orofacial Clefting Revisited: An Anthropometric Investigation. Cleft Palate-Craniofacial Journal, 2017, 54, 631-638.	0.9	11
53	Exploring Subclinical Phenotypic Features in Twin Pairs Discordant for Cleft Lip and Palate. Cleft Palate-Craniofacial Journal, 2017, 54, 90-93.	0.9	10
54	A Novel Variant of <scp>ATP5MC3</scp> Associated with Both Dystonia and Spastic Paraplegia. Movement Disorders, 2022, 37, 375-383.	3.9	10

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55	The PAX1 locus at 20p11 is a potential genetic modifier for bilateral cleft lip. Human Genetics and Genomics Advances, 2021, 2, 100025.	1.7	9
56	GWAS reveals loci associated with velopharyngeal dysfunction. Scientific Reports, 2018, 8, 8470.	3.3	8
57	Integrative approaches generate insights into the architecture of non-syndromic cleft lip Â± cleft palate. Human Genetics and Genomics Advances, 2021, 2, 100038.	1.7	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.	1.2	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.	0.9	6
61	Individuals with nonsyndromic orofacial clefts have increased asymmetry of fingerprint patterns. PLoS ONE, 2020, 15, e0230534.	2.5	6
62	The Effect of Ethylene Glycol, Glycine Betaine, and Urea on Lysozyme Thermal Stability. Journal of Chemical Education, 2010, 87, 1393-1395.	2.3	5
63	Genomeâ€wide association study of multiethnic nonsyndromic orofacial cleft families identifies novel loci specific to family and phenotypic subtypes. Genetic Epidemiology, 2022, , .	1.3	4
64	Embracing human genetics: a primer for developmental biologists. Development (Cambridge), 2020, 147, .	2.5	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.	3.7	3
66	Detection of de novo copy number deletions from targeted sequencing of trios. Bioinformatics, 2019, 35, 571-578.	4.1	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.	3.7	2
68	Efficient estimation of indirect effects in caseâ€control studies using a unified likelihood framework. Statistics in Medicine, 2022, 41, 2879-2893.	1.6	2
69	Feasibility of Social Media Recruitment for Orofacial Cleft Genetic Research. Cleft Palate-Craniofacial Journal, 2022, 59, 701-707.	0.9	1
70	Genetic Variants and the Cortisol Response in Children: An Exploratory Study. Biological Research for Nursing, 2019, 21, 157-165.	1.9	1
71	Heritability Analysis in Twins Indicates a Genetic Basis for Velopharyngeal Morphology. Cleft Palate-Craniofacial Journal, 2022, 59, 1340-1345.	0.9	1
72	Leveraging Family History in Case-Control Analyses of Rare Variation. Genetics, 2020, 214, 295-303.	2.9	0

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73	Identification of a nonâ€coding SNP associated with risk for nonâ€syndromic orofacial clefting with alleleâ€specific effects on IRF6 expression in vitro. FASEB Journal, 2021, 35, .	0.5	0
74	Developmental Anomalies â€“ Clefts. , 2015, , 91-116.		0
75	Identifying Genetic Sources of Phenotypic Heterogeneity in Orofacial Clefts by Targeted Sequencing. Birth Defects Research, 2017, , .	1.5	0