

Laura Southgate

List of Publications by Year in descending order

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Version: 2024-02-01

48
papers

3,686
citations

236612

25
h-index

264894

42
g-index

56
all docs

56
docs citations

56
times ranked

6697
citing authors

#	ARTICLE	IF	CITATIONS
1	Elevated Levels of Inflammatory Cytokines Predict Survival in Idiopathic and Familial Pulmonary Arterial Hypertension. <i>Circulation</i> , 2010, 122, 920-927.	1.6	661
2	Common variants near CAV1 and CAV2 are associated with primary open-angle glaucoma. <i>Nature Genetics</i> , 2010, 42, 906-909.	9.4	357
3	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 75-90.	2.6	343
4	Identification of rare sequence variation underlying heritable pulmonary arterial hypertension. <i>Nature Communications</i> , 2018, 9, 1416.	5.8	279
5	Health and population effects of rare gene knockouts in adult humans with related parents. <i>Science</i> , 2016, 352, 474-477.	6.0	272
6	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. <i>Human Mutation</i> , 2015, 36, 1113-1127.	1.1	185
7	Molecular genetic framework underlying pulmonary arterial hypertension. <i>Nature Reviews Cardiology</i> , 2020, 17, 85-95.	6.1	181
8	Molecular genetic characterization of SMAD signaling molecules in pulmonary arterial hypertension. <i>Human Mutation</i> , 2011, 32, 1385-1389.	1.1	152
9	Genetic determinants of risk in pulmonary arterial hypertension: international genome-wide association studies and meta-analysis. <i>Lancet Respiratory Medicine</i> , 2019, 7, 227-238.	5.2	122
10	Phenotypic Characterization of <i>EIF2AK4</i> Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. <i>Circulation</i> , 2017, 136, 2022-2033.	1.6	111
11	Gain-of-Function Mutations of ARHGAP31, a Cdc42/Rac1 GTPase Regulator, Cause Syndromic Cutis Aplasia and Limb Anomalies. <i>American Journal of Human Genetics</i> , 2011, 88, 574-585.	2.6	100
12	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adams-Oliver Syndrome With Variable Cardiac Anomalies. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 572-581.	5.1	84
13	Characterization of <i>GDF2</i> Mutations and Levels of BMP9 and BMP10 in Pulmonary Arterial Hypertension. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2020, 201, 575-585.	2.5	80
14	Information processing bias in anorexia nervosa. <i>Psychiatry Research</i> , 2008, 160, 221-227.	1.7	74
15	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 475-482.	2.6	73
16	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 103, 3-18.	2.6	46
17	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. <i>American Journal of Human Genetics</i> , 2019, 104, 948-956.	2.6	45
18	Whole-Blood RNA Profiles Associated with Pulmonary Arterial Hypertension and Clinical Outcome. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2020, 202, 586-594.	2.5	45

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19	Rare variant analysis of 4241 pulmonary arterial hypertension cases from an international consortium implicates FBLN2, PDGFD, and rare de novo variants in PAH. <i>Genome Medicine</i> , 2021, 13, 80.	3.6	43
20	A restricted spectrum of missense KMT2D variants cause a multiple malformations disorder distinct from Kabuki syndrome. <i>Genetics in Medicine</i> , 2020, 22, 867-877.	1.1	41
21	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. <i>American Journal of Human Genetics</i> , 2018, 103, 144-153.	2.6	36
22	<i>DOCK6</i> Mutations Are Responsible for a Distinct Autosomal-Recessive Variant of Adams-Oliver Syndrome Associated with Brain and Eye Anomalies. <i>Human Mutation</i> , 2015, 36, 593-598.	1.1	32
23	Genome-Wide Association Study Identifies Risk Loci for Cluster Headache. <i>Annals of Neurology</i> , 2021, 90, 193-202.	2.8	31
24	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. <i>Human Mutation</i> , 2018, 39, 1246-1261.	1.1	31
25	Bayesian Inference Associates Rare <i>KDR</i> Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, .	1.6	29
26	Novel SPG11 mutations in Asian kindreds and disruption of spatascin function in the zebrafish. <i>Neurogenetics</i> , 2010, 11, 379-389.	0.7	26
27	Dymeclin, the gene underlying Dyggve-Melchior-Clausen syndrome, encodes a protein integral to extracellular matrix and golgi organization and is associated with protein secretion pathways critical in bone development. <i>Human Mutation</i> , 2011, 32, 231-239.	1.1	26
28	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. <i>American Journal of Human Genetics</i> , 2017, 100, 334-342.	2.6	26
29	Mendelian randomisation analysis of red cell distribution width in pulmonary arterial hypertension. <i>European Respiratory Journal</i> , 2020, 55, 1901486.	3.1	26
30	Biallelic variants of <i>ATP13A3</i> cause dose-dependent childhood-onset pulmonary arterial hypertension characterised by extreme morbidity and mortality. <i>Journal of Medical Genetics</i> , 2022, 59, 906-911.	1.5	22
31	Length of uninterrupted repeats determines instability at the unstable mouse expanded simple tandem repeat family MMS10 derived from independent SINE B1 elements. <i>Mammalian Genome</i> , 2001, 12, 104-111.	1.0	18
32	The development of the childhood retrospective perfectionism questionnaire (CHIRP) in an eating disorder sample. <i>European Eating Disorders Review</i> , 2008, 16, 451-462.	2.3	18
33	The DOCK protein family in vascular development and disease. <i>Angiogenesis</i> , 2021, 24, 417-433.	3.7	18
34	Whole Exome Sequence Analysis Provides Novel Insights into the Genetic Framework of Childhood-Onset Pulmonary Arterial Hypertension. <i>Genes</i> , 2020, 11, 1328.	1.0	14
35	Assessment of a Pulmonary Origin for Blood Outgrowth Endothelial Cells by Examination of Identical Twins Harboring a BMPR2 Mutation. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2013, 188, 258-260.	2.5	7
36	Network-Informed Gene Ranking Tackles Genetic Heterogeneity in Exome-Sequencing Studies of Monogenic Disease. <i>Human Mutation</i> , 2015, 36, 1135-1144.	1.1	7

#	ARTICLE	IF	CITATIONS
37	Molecular genetics of pulmonary hypertension in children. <i>Current Opinion in Genetics and Development</i> , 2022, 75, 101936.	1.5	2
38	Response to Letter Regarding Article, "Elevated Levels of Inflammatory Cytokines Predict Survival in Idiopathic and Familial Pulmonary Arterial Hypertension" <i>Circulation</i> , 2011, 123, .	1.6	1
39	Current opinion in the molecular genetics of Adams-Oliver syndrome. <i>Expert Opinion on Orphan Drugs</i> , 2019, 7, 21-26.	0.5	1
40	ARHGAP31, DOCK6, RBPJ, EOGT, and Adams-Oliver Syndrome. , 2016, , 1203-1209.		1
41	Extraction and high-throughput sequencing of oak heartwood DNA: Assessing the feasibility of genome-wide DNA methylation profiling. <i>PLoS ONE</i> , 2021, 16, e0254971.	1.1	1
42	Elevated Levels Of Inflammatory Cytokines Are Strong Predictors Of Survival In Idiopathic And Familial Pulmonary Arterial Hypertension. , 2010, , .		0
43	Exome Sequencing Identifies Mutations Of A GTPase Regulator In Adams-Oliver Syndrome, A Rare Cause Of Pulmonary Hypertension. , 2011, , .		0
44	Elucidating the molecular genetic basis of cluster headache: delineation of the genetic architecture by exome sequencing. <i>Journal of Headache and Pain</i> , 2013, 14, .	2.5	0
45	Letter regarding "Distal Limb Defects and Aplasia Cutis: Adams' Oliver Syndrome" <i>Journal of Hand Surgery</i> , 2016, 41, e327.	0.7	0
46	S107...Genotype-phenotype associations in pulmonary arterial hypertension caused by BMPR2 and EIF2AK4 variants. <i>Thorax</i> , 2016, 71, A63-A64.	2.7	0
47	Pulmonary Arterial Hypertension: A Deeper Evaluation of Genetic Risk in the -Omics Era. <i>Genes</i> , 2021, 12, 1798.	1.0	0
48	Multi-omic profiling in pulmonary arterial hypertension. , 2020, , .		0