Laura Southgate

List of Publications by Year
in descending order

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48 papers	3,686 citations	236833 25 h-index	265120 42 g-index
56 all docs	56 docs citations	56 times ranked	6697 citing authors

#	Article	IF	Citations
1	Biallelic variants of ATP13A3 cause dose-dependent childhood-onset pulmonary arterial hypertension characterised by extreme morbidity and mortality. Journal of Medical Genetics, 2022, 59, 906-911.	1.5	22
2	Molecular genetics of pulmonary hypertension in children. Current Opinion in Genetics and Development, 2022, 75, 101936.	1.5	2
3	Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	1.6	29
4	The DOCK protein family in vascular development and disease. Angiogenesis, 2021, 24, 417-433.	3.7	18
5	Rare variant analysis of 4241 pulmonary arterial hypertension cases from an international consortium implicates FBLN2, PDGFD, and rare de novo variants in PAH. Genome Medicine, 2021, 13, 80.	3.6	43
6	Genomeâ€Wide Association Study Identifies Risk Loci for Cluster Headache. Annals of Neurology, 2021, 90, 193-202.	2.8	31
7	Extraction and high-throughput sequencing of oak heartwood DNA: Assessing the feasibility of genome-wide DNA methylation profiling. PLoS ONE, 2021, 16, e0254971.	1.1	1
8	Pulmonary Arterial Hypertension: A Deeper Evaluation of Genetic Risk in the -Omics Era. Genes, 2021, 12, 1798.	1.0	0
9	Molecular genetic framework underlying pulmonary arterial hypertension. Nature Reviews Cardiology, 2020, 17, 85-95.	6.1	181
10	Characterization of GDF2 Mutations and Levels of BMP9 and BMP10 in Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2020, 201, 575-585.	2.5	80
11	Whole Exome Sequence Analysis Provides Novel Insights into the Genetic Framework of Childhood-Onset Pulmonary Arterial Hypertension. Genes, 2020, 11, 1328.	1.0	14
12	Whole-Blood RNA Profiles Associated with Pulmonary Arterial Hypertension and Clinical Outcome. American Journal of Respiratory and Critical Care Medicine, 2020, 202, 586-594.	2.5	45
13	A restricted spectrum of missense KMT2D variants cause a multiple malformations disorder distinct fromKabuki syndrome. Genetics in Medicine, 2020, 22, 867-877.	1.1	41
14	Mendelian randomisation analysis of red cell distribution width in pulmonary arterial hypertension. European Respiratory Journal, 2020, 55, 1901486.	3.1	26
15	Multi-omic profiling in pulmonary arterial hypertension. , 2020, , .		0
16	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	2.6	45
17	Genetic determinants of risk in pulmonary arterial hypertension: international genome-wide association studies and meta-analysis. Lancet Respiratory Medicine,the, 2019, 7, 227-238.	5.2	122
18	Current opinion in the molecular genetics of Adams-Oliver syndrome. Expert Opinion on Orphan Drugs, 2019, 7, 21-26.	0.5	1

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19	Identification of rare sequence variation underlying heritable pulmonary arterial hypertension. Nature Communications, 2018, 9, 1416.	5.8	279
20	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	2.6	36
21	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. American Journal of Human Genetics, 2018, 103, 3-18.	2.6	46
22	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	1.1	31
23	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342.	2.6	26
24	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	2.6	343
25	Phenotypic Characterization of EIF2AK4 Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. Circulation, 2017, 136, 2022-2033.	1.6	111
26	Letter regarding â€œDistal Limb Defects and Aplasia Cutis: Adamsâ€“Oliver Syndromeâ€• Journal of Hand Surgery, 2016, 41, e327.	0.7	0
27	S107â€…Genotype-phenotype associations in pulmonary arterial hypertension caused by BMPR2 and EIF2AK4 variants. Thorax, 2016, 71, A63-A64.	2.7	0
28	Health and population effects of rare gene knockouts in adult humans with related parents. Science, 2016, 352, 474-477.	6.0	272
29	ARHGAP31, DOCK6, RBPJ, EOGT, and Adams-Oliver Syndrome. , 2016, , 1203-1209.		1
30	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. Human Mutation, 2015, 36, 1113-1127.	1.1	185
31	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adamsâ€“Oliver Syndrome With Variable Cardiac Anomalies. Circulation: Cardiovascular Genetics, 2015, 8, 572-581.	5.1	84
32	Network-Informed Gene Ranking Tackles Genetic Heterogeneity in Exome-Sequencing Studies of Monogenic Disease. Human Mutation, 2015, 36, 1135-1144.	1.1	7
33	DOCK6Mutations Are Responsible for a Distinct Autosomal-Recessive Variant of Adams-Oliver Syndrome Associated with Brain and Eye Anomalies. Human Mutation, 2015, 36, 593-598.	1.1	32
34	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2015, 97, 475-482.	2.6	73
35	Elucidating the molecular genetic basis of cluster headache: delineation of the genetic architecture by exome sequencing. Journal of Headache and Pain, 2013, 14, .	2.5	0
36	Assessment of a Pulmonary Origin for Blood Outgrowth Endothelial Cells by Examination of Identical Twins Harboring aBMPR2Mutation. American Journal of Respiratory and Critical Care Medicine, 2013, 188, 258-260.	2.5	7

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37	Exome Sequencing Identifies Mutations Of A GTPase Regulator In Adams-Oliver Syndrome, A Rare Cause Of Pulmonary Hypertension. , 2011, , .		0
38	Gain-of-Function Mutations of ARHGAP31, a Cdc42/Rac1 GTPase Regulator, Cause Syndromic Cutis Aplasia and Limb Anomalies. American Journal of Human Genetics, 2011, 88, 574-585.	2.6	100
39	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. Human Mutation, 2011, 32, 231-239.	1.1	26
40	Molecular genetic characterization of SMAD signaling molecules in pulmonary arterial hypertension. Human Mutation, 2011, 32, 1385-1389.	1.1	152
41	Response to Letter Regarding Article, â€œElevated Levels of Inflammatory Cytokines Predict Survival in Idiopathic and Familial Pulmonary Arterial Hypertensionâ€• Circulation, 2011, 123, .	1.6	1
42	Elevated Levels Of Inflammatory Cytokines Are Strong Predictors Of Survival In Idiopathic And Familial Pulmonary Arterial Hypertension. , 2010, , .		0
43	Novel SPG11 mutations in Asian kindreds and disruption of spatacsin function in the zebrafish. Neurogenetics, 2010, 11, 379-389.	0.7	26
44	Common variants near CAV1 and CAV2 are associated with primary open-angle glaucoma. Nature Genetics, 2010, 42, 906-909.	9.4	357
45	Elevated Levels of Inflammatory Cytokines Predict Survival in Idiopathic and Familial Pulmonary Arterial Hypertension. Circulation, 2010, 122, 920-927.	1.6	661
46	The development of the childhood retrospective perfectionism questionnaire (CHIRP) in an eating disorder sample. European Eating Disorders Review, 2008, 16, 451-462.	2.3	18
47	Information processing bias in anorexia nervosa. Psychiatry Research, 2008, 160, 221-227.	1.7	74
48	Length of uninterrupted repeats determines instability at the unstable mouse expanded simple tandem repeat family MMS10 derived from independent SINE B1 elements. Mammalian Genome, 2001, 12, 104-111.	1.0	18