David A Piccoli

List of Publications by Year
in descending order

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Version: 2024-02-01

67 papers	7,857 citations	81900 39 h-index	144013 57 g-index
68 all docs	68 docs citations	68 times ranked	4958 citing authors

#	Article	IF	Citations
1	Alagille syndrome is caused by mutations in human Jagged1, which encodes a ligand for Notch1. Nature Genetics, 1997, 16, 243-251.	21.4	1,184
2	Mutations in the human Jagged1 gene are responsible for Alagille syndrome. Nature Genetics, 1997, 16, 235-242.	21.4	1,072
3	NOTCH2 Mutations Cause Alagille Syndrome, a Heterogeneous Disorder of the Notch Signaling Pathway. American Journal of Human Genetics, 2006, 79, 169-173.	6.2	663
4	Features of alagille syndrome in 92 patients: Frequency and relation to prognosis. Hepatology, 1999, 29, 822-829.	7.3	591
5	Vascular Anomalies in Alagille Syndrome. Circulation, 2004, 109, 1354-1358.	1.6	333
6	Analysis of Cardiovascular Phenotype and Genotype-Phenotype Correlation in Individuals With a JAG1 Mutation and/or Alagille Syndrome. Circulation, 2002, 106, 2567-2574.	1.6	273
7	Spectrum and Frequency of Jagged1 (JAG1) Mutations in Alagille Syndrome Patients and Their Families. American Journal of Human Genetics, 1998, 62, 1361-1369.	6.2	218
8	NOTCH2mutations in Alagille syndrome. Journal of Medical Genetics, 2012, 49, 138-144.	3.2	197
9	Inflammatory bowel disease in children 5 years of age and younger. American Journal of Gastroenterology, 2002, 97, 2005-2010.	0.4	189
10	INFLAMMATORY BOWEL DISEASE IN PEDIATRIC AND ADOLESCENT PATIENTS. Gastroenterology Clinics of North America, 1999, 28, 445-458.	2.2	183
11	Vitamin D status in children, adolescents, and young adults with Crohn disease,,. American Journal of Clinical Nutrition, 2002, 76, 1077-1081.	4.7	172
12	Risk factors for low bone mineral density in children and young adults with Crohnâ€™s disease. Journal of Pediatrics, 1999, 135, 593-600.	1.8	147
13	Jagged1 mutations in patients ascertained with isolated congenital heart defects. , 1999, 84, 56-60.		137
14	Protein-Losing Enteropathy in Patients With Congenital Heart Disease. Journal of the American College of Cardiology, 2017, 69, 2929-2937.	2.8	136
15	Growth, Body Composition, and Nutritional Status in Children and Adolescents With Crohn's Disease. Journal of Pediatric Gastroenterology and Nutrition, 2000, 31, 33-40.	1.8	130
16	Alagille Syndrome and the Jagged1 Gene. Seminars in Liver Disease, 2001, 21, 525-534.	3.6	127
17	Characterization of Notch receptor expression in the developing mammalian heart and liver. American Journal of Medical Genetics Part A, 2002, 112, 181-189.	2.4	113
18	Renal anomalies in Alagille syndrome: A diseaseâ€defining feature. American Journal of Medical Genetics, Part A, 2012, 158A, 85-89.	1.2	102

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19	Facial features in Alagille syndrome: Specific or cholestasis facies?. American Journal of Medical Genetics Part A, 2002, 112, 163-170.	2.4	101
20	Intracranial Vascular Abnormalities in Patients with Alagille Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2005, 41, 99-107.	1.8	101
21	Clinical and molecular genetics of Alagille syndrome. Current Opinion in Pediatrics, 1999, 11, 558-564.	2.0	96
22	Frequency and Clinical Correlations of Granulomas in Children With Crohn Disease. Journal of Pediatric Gastroenterology and Nutrition, 2008, 46, 392-398.	1.8	91
23	Alagille syndrome mutation update: Comprehensive overview ofJAG1andNOTCH2mutation frequencies and insight into missense variant classification. Human Mutation, 2019, 40, 2197-2220.	2.5	84
24	Mutation analysis of Jagged1 (JAG1) in Alagille syndrome patients. Human Mutation, 2001, 17, 151-152.	2.5	76
25	Deletions of 20p12 in Alagille syndrome: Frequency and molecular characterization. , 1997, 70, 80-86.		71
26	Autoimmune Pancreatitis in Children: Characteristic Features, Diagnosis, and Management. American Journal of Gastroenterology, 2017, 112, 1604-1611.	0.4	70
27	Pathologic Lower Extremity Fractures in Children With Alagille Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2010, 51, 66-70.	1.8	69
28	Heritable disorders of the bile ducts. Gastroenterology Clinics of North America, 2003, 32, 857-875.	2.2	63
29	SNP array mapping of chromosome 20p deletions: Genotypes, phenotypes, and copy number variation. Human Mutation, 2009, 30, 371-378.	2.5	61
30	Medical Management of Alagille Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2010, 50, 580-586.	1.8	61
31	Bone Mineral Density in Children and Young Adults with CrohnÊ¼s Disease. Inflammatory Bowel Diseases, 1999, 5, 161-166.	1.9	60
32	Experiences with 6-Mercaptopurine and Azathioprine Therapy in Pediatric Patients with Severe Ulcerative Colitis. Journal of Pediatric Gastroenterology and Nutrition, 1999, 28, 54-58.	1.8	60
33	Outcomes of Childhood Cholestasis in Alagille Syndrome: Results of a Multicenter Observational Study. Hepatology Communications, 2020, 4, 387-398.	4.3	52
34	Intrahepatic dynamic contrast MR lymphangiography: initial experience with a new technique for the assessment of liver lymphatics. European Radiology, 2019, 29, 5190-5196.	4.5	51
35	Single toxin detection is inadequate to diagnose Clostridium difficile diarrhea in pediatric patients. Gastroenterology, 1998, 115, 1329-1334.	1.3	49
36	Monozygotic twins with a severe form of Alagille syndrome and phenotypic discordance. American Journal of Medical Genetics Part A, 2002, 112, 194-197.	2.4	49

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37	A Longitudinal Study to Identify Laboratory Predictors of Liver Disease Outcome in Alagille Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2010, 50, 526-530.	1.8	47
38	Craniosynostosis in Alagille syndrome. American Journal of Medical Genetics Part A, 2002, 112, 176-180.	2.4	46
39	Severe Eosinophilic Gastroenteritis in a Crohnâ€™s Disease Patient Treated With Infliximab and Adalimumab. American Journal of Gastroenterology, 2016, 111, 437-438.	0.4	44
40	Deficits in Size-Adjusted Bone Mass in Children with Alagille Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2005, 40, 76-82.	1.8	41
41	Growth, nutritional status, body composition, and energy expenditure in prepubertal children with Alagille syndrome. Journal of Pediatrics, 1999, 134, 172-177.	1.8	39
42	A de novo whole gene deletion of XIAP detected by exome sequencing analysis in very early onset inflammatory bowel disease: a case report. BMC Gastroenterology, 2015, 15, 160.	2.0	38
43	Early life predictive markers of liver disease outcome in an International, Multicentre Cohort of children with Alagille syndrome. Liver International, 2016, 36, 755-760.	3.9	37
44	Rethinking Growth Failure in Alagille Syndrome: The Role of Dietary Intake and Steatorrhea. Journal of Pediatric Gastroenterology and Nutrition, 2002, 35, 495-502.	1.8	36
45	Recommendations for Diagnosis and Management of Autoimmune Pancreatitis in Childhood. Journal of Pediatric Gastroenterology and Nutrition, 2018, 67, 232-236.	1.8	35
46	Moyamoya Syndrome Associated with Alagille Syndrome: Outcome after Surgical Revascularization. Journal of Pediatrics, 2015, 166, 470-473.	1.8	29
47	Alagille syndrome inherited from a phenotypically normal mother with a mosaic 20p microdeletion. American Journal of Medical Genetics Part A, 2002, 112, 190-193.	2.4	26
48	Inflammatory bowel disease in children. Medical Clinics of North America, 1994, 78, 1281-1302.	2.5	24
49	Compound heterozygous mutations in NEK8 in siblings with endâ€stage renal disease with hepatic and cardiac anomalies. American Journal of Medical Genetics, Part A, 2016, 170, 750-753.	1.2	22
50	Bone mineral density in children and young adults with Crohn's disease. Inflammatory Bowel Diseases, 0, 5, 161-166.	1.9	21
51	Pancreatic Insufficiency Is Not a Prevalent Problem in Alagille Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2012, 55, 612-614.	1.8	20
52	Proteinâ€elongating mutations inMYH11are implicated in a dominantly inherited smooth muscle dysmotility syndrome with severe esophageal, gastric, and intestinal disease. Human Mutation, 2020, 41, 973-982.	2.5	18
53	Genome sequencing increases diagnostic yield in clinically diagnosed Alagille syndrome patients with previously negative test results. Genetics in Medicine, 2021, 23, 323-330.	2.4	17
54	Alagille Syndrome. , 2007, , 326-345.		15

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55	Alagille syndrome and risk for hepatocellular carcinoma: Need for increased surveillance in adults with mild liver phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 719-731.	1.2	12
56	Bile canalicular morphometry in arteriohepatic dysplasia. Hepatology, 1987, 7, 1262-1266.	7.3	11
57	Histological features of ileitis differentiating pediatric Crohn disease from ulcerative colitis with backwash ileitis. Digestive and Liver Disease, 2018, 50, 147-153.	0.9	11
58	Alagille syndrome. , 2014, , 216-233.		10
59	Bone geometry and microarchitecture deficits in children with Alagille syndrome. Bone, 2020, 141, 115576.	2.9	9
60	Anterior Chamber Pathology in Alagille Syndrome. Ocular Oncology and Pathology, 2016, 2, 270-275.	1.0	6
61	Chromosome 10q23 Deletion Syndrome: An Overlap of Bannayanâ€“Rileyâ€“Ruvalcaba Syndrome and Juvenile Polyposis Syndrome. Journal of Paediatrics and Child Health, 2016, 52, 852-852.	0.8	3
62	Alagille Syndrome. , 2021, , 222-241.		2
63	Alagille Syndrome and JAGGED1/NOTCH Sequence. , 2010, , 159-178.		1
64	Alagille Syndrome. , 2008, , 227-232.		1
65	Alagille Syndrome: Overview and Introduction. , 2018, , 1-9.		0
66	Back Cover, Volume 40, Issue 12. Human Mutation, 2019, 40, iii.	2.5	0
67	Alagille Syndrome. , 2004, , 20-24.		0