Nancy B Spinner

List of Publications by Year
in descending order

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120	15,432	44069 48	24258 110
papers	citations	h-index	g-index

127	127	127	15627
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies. American Journal of Human Genetics, 2010, 86, 749-764.	6.2	2,325
2	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
3	Mutations in the human Jagged1 gene are responsible for Alagille syndrome. Nature Genetics, 1997, 16, 235-242.	21.4	1,072
4	KILLER/DR5 is a DNA damageâ€“inducible p53â€“regulated death receptor gene. Nature Genetics, 1997, 17, 141-143.	21.4	1,005
5	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
6	Features of alagille syndrome in 92 patients: Frequency and relation to prognosis. Hepatology, 1999, 29, 822-829.	7.3	591
7	A genomic view of mosaicism and human disease. Nature Reviews Genetics, 2013, 14, 307-320.	16.3	527
8	Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents. American Journal of Human Genetics, 2015, 97, 6-21.	6.2	453
9	Deletions and microdeletions of 22q11.2 in veloâ€cardioâ€facial syndrome. American Journal of Medical Genetics Part A, 1992, 44, 261-268.	2.4	387
10	Mechanisms of mosaicism, chimerism and uniparental disomy identified by single nucleotide polymorphism array analysis. Human Molecular Genetics, 2010, 19, 1263-1275.	2.9	373
11	GLUT-1 deficiency syndrome caused by haploinsufficiency of the blood-brain barrier hexose carrier. Nature Genetics, 1998, 18, 188-191.	21.4	349
12	Return of Genomic Results to Research Participants: The Floor, the Ceiling, and the Choices In Between. American Journal of Human Genetics, 2014, 94, 818-826.	6.2	342
13	Vascular Anomalies in Alagille Syndrome. Circulation, 2004, 109, 1354-1358.	1.6	333
14	High-resolution mapping and analysis of copy number variations in the human genome: A data resource for clinical and research applications. Genome Research, 2009, 19, 1682-1690.	5.5	313
15	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. Genome Research, 2015, 25, 305-315.	5.5	313
16	Notch signaling in human development and disease. Seminars in Cell and Developmental Biology, 2012, 23, 450-457.	5.0	286
17	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
18	Jagged1 mutations in Alagille syndrome. Human Mutation, 2001, 17, 18-33.	2.5	224

#	Article	IF	Citations
19	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
20	Down syndrome congenital heart disease: A narrowed region and a candidate gene. Genetics in Medicine, 2001, 3, 91-101.	2.4	168
21	Jagged1 mutations in patients ascertained with isolated congenital heart defects. , 1999, 84, 56-60.		137
22	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	6.2	137
23	Alagille Syndrome and the Jagged1 Gene. Seminars in Liver Disease, 2001, 21, 525-534.	3.6	127
24	Evidence From Human and Zebrafish That GPC1 Is a Biliary Atresia Susceptibility Gene. Gastroenterology, 2013, 144, 1107-1115.e3.	1.3	125
25	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	2.4	125
26	Jagged1 (JAG1) mutations in patients with tetralogy of fallot or pulmonic stenosis. Human Mutation, 2010, 31, 594-601.	2.5	113
27	Jagged1 (JAG1): Structure, expression, and disease associations. Gene, 2016, 576, 381-384.	2.2	104
28	Renal anomalies in Alagille syndrome: A diseaseâ€defining feature. American Journal of Medical Genetics, Part A, 2012, 158A, 85-89.	1.2	102
29	Facial features in Alagille syndrome: Specific or cholestasis facies?. American Journal of Medical Genetics Part A, 2002, 112, 163-170.	2.4	101
30	Intracranial Vascular Abnormalities in Patients with Alagille Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2005, 41, 99-107.	1.8	101
31	Processes and preliminary outputs for identification of actionable genes as incidental findings in genomic sequence data in the Clinical Sequencing Exploratory Research Consortium. Genetics in Medicine, 2013, 15, 860-867.	2.4	99
32	Clinical and molecular genetics of Alagille syndrome. Current Opinion in Pediatrics, 1999, 11, 558-564.	2.0	96
33	Alagille syndrome mutation update: Comprehensive overview ofJAG1andNOTCH2mutation frequencies and insight into missense variant classification. Human Mutation, 2019, 40, 2197-2220.	2.5	84
34	Mutation analysis of Jagged1 (JAG1) in Alagille syndrome patients. Human Mutation, 2001, 17, 151-152.	2.5	76
35	Defective intracellular transport and processing of JAG1 missense mutations in Alagille syndrome. Human Molecular Genetics, 2001, 10, 405-413.	2.9	76
36	Renal involvement and the role of Notch signalling in Alagille syndrome. Nature Reviews Nephrology, 2013, 9, 409-418.	9.6	75

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37	Deletions of 20p12 in Alagille syndrome: Frequency and molecular characterization. , 1997, 70, 80-86.		71
38	Bile duct proliferation in Jag1/fringe heterozygous mice identifies candidate modifiers of the alagille syndrome hepatic phenotype. Hepatology, 2008, 48, 1989-1997.	7.3	69
39	Pathologic Lower Extremity Fractures in Children With Alagille Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2010, 51, 66-70.	1.8	69
40	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315.	2.4	69
41	A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. Genetics in Medicine, 2017, 19, 575-582.	2.4	68
42	Parentsâ€™ Perceptions of the Usefulness of Chromosomal Microarray Analysis for Children with Autism Spectrum Disorders. Journal of Autism and Developmental Disorders, 2015, 45, 3262-3275.	2.7	67
43	Genomic alterations in biliary atresia suggest region of potential disease susceptibility in 2q37.3. American Journal of Medical Genetics, Part A, 2010, 152A, 886-895.	1.2	64
44	SNP array mapping of chromosome 20p deletions: Genotypes, phenotypes, and copy number variation. Human Mutation, 2009, 30, 371-378.	2.5	61
45	Molecular analysis of ring chromosome 20 syndrome reveals two distinct groups of patients. Journal of Medical Genetics, 2011, 48, 1-9.	3.2	61
46	Replication of a GWAS signal in a Caucasian population implicates ADD3 in susceptibility to biliary atresia. Human Genetics, 2014, 133, 235-243.	3.8	59
47	Peripheral Bile Duct Paucity and Cholestasis in the Liver of a Patient With Alagille Syndrome. American Journal of Surgical Pathology, 2005, 29, 820-826.	3.7	58
48	Identification of Polycystic Kidney Disease 1 Like 1 Gene Variants in Children With Biliary Atresia Splenic Malformation Syndrome. Hepatology, 2019, 70, 899-910.	7.3	58
49	Conditional JAG1 Mutation Shows the Developing Heart Is More Sensitive Than Developing Liver to JAG1 Dosage. American Journal of Human Genetics, 2003, 72, 1065-1070.	6.2	55
50	Outcomes of Childhood Cholestasis in Alagille Syndrome: Results of a Multicenter Observational Study. Hepatology Communications, 2020, 4, 387-398.	4.3	52
51	A genome-wide association study identifies a susceptibility locus for biliary atresia on 2p16.1 within the gene EFEMP1. PLoS Genetics, 2018, 14, e1007532.	3.5	51
52	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
53	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
54	Further evidence for the possible role of MEIS2 in the development of cleft palate and cardiac septum. American Journal of Medical Genetics, Part A, 2010, 152A, 1326-1327.	1.2	47

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55	Prenatal DNA Sequencing: Clinical, Counseling, and Diagnostic Laboratory Considerations. Prenatal Diagnosis, 2018, 38, 26-32.	2.3	47
56	Use of a Dynamic Genetic Testing Approach for Childhood-Onset Epilepsy. JAMA Network Open, 2019, 2, e192129.	5.9	45
57	Heterozygous Deletion ofFOXA2Segregates with Disease in a Family with Heterotaxy, Panhypopituitarism, and Biliary Atresia. Human Mutation, 2015, 36, 631-637.	2.5	43
58	Ring chromosome 20. European Journal of Medical Genetics, 2012, 55, 381-387.	1.3	40
59	CADASIL: Notch signaling defect or protein accumulation problem?. Journal of Clinical Investigation, 2000, 105, 561-562.	8.2	38
60	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
61	Jagged1 (JAG1) mutation detection in an Australian Alagille syndrome population. Human Mutation, 2000, 16, 408-416.	2.5	35
62	THBS2 Is a Candidate Modifier of Liver Disease Severity in Alagille Syndrome. Cellular and Molecular Gastroenterology and Hepatology, 2016, 2, 663-675.e2.	4.5	35
63	Ablepharon macrostomia syndrome with associated cutis laxa: Possible localization to 18q. Human Genetics, 1996, 97, 532-536.	3.8	32
64	Panhandle Polymerase Chain Reaction Amplifies MLL Genomic Translocation Breakpoint Involving Unknown Partner Gene. Blood, 1997, 90, 4679-4686.	1.4	32
65	Alagille Syndrome: Genetics and Functional Models. Current Pathobiology Reports, 2017, 5, 233-241.	3.4	32
66	Mosaic loss of 15q11q13 in a patient with hypomelanosis of Ito: is there a role for the P gene?. Human Genetics, 1995, 96, 485-9.	3.8	30
67	Mosaic paternal uniparental (iso)disomy for chromosome 20 associated with multiple anomalies. American Journal of Medical Genetics Part A, 2004, 124A, 274-279.	2.4	30
68	Clinical utility gene card for: Alagille Syndrome (ALGS). European Journal of Human Genetics, 2014, 22, 435-435.	2.8	30
69	Genomic Diagnosis for Pediatric Disorders: Revolution and Evolution. Frontiers in Pediatrics, 2020, 8, 373.	1.9	30
70	Moyamoya Syndrome Associated with Alagille Syndrome: Outcome after Surgical Revascularization. Journal of Pediatrics, 2015, 166, 470-473.	1.8	29
71	Sequencing-based diagnostics for pediatric genetic diseases: progress and potential. Expert Review of Molecular Diagnostics, 2016, 16, 987-999.	3.1	29
72	Supernumerary inv dup(15) in a patient with Angelman syndrome and a deletion of 15q11-q13. American Journal of Medical Genetics Part A, 1995, 57, 61-65.	2.4	28

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73	Novel findings with reassessment of exome data: implications for validation testing and interpretation of genomic data. Genetics in Medicine, 2018, 20, 329-336.	2.4	28
74	AUDIOME: a tiered exome sequencingâ€“based comprehensive gene panel for the diagnosis of heterogeneous nonsyndromic sensorineural hearing loss. Genetics in Medicine, 2018, 20, 1600-1608.	2.4	27
75	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
76	Utility and limitations of exome sequencing as a genetic diagnostic tool for children with hearing loss. Genetics in Medicine, 2018, 20, 1663-1676.	2.4	26
77	Complex management of a patient with a contiguous Xp11.4 gene deletion involving ornithine transcarbamylase: A role for detailed molecular analysis in complex presentations of classical diseases. Molecular Genetics and Metabolism, 2008, 94, 498-502.	1.1	25
78	Alagille syndrome in a Vietnamese cohort: Mutation analysis and assessment of facial features. American Journal of Medical Genetics, Part A, 2012, 158A, 1005-1013.	1.2	24
79	Spectrum of JAG1 gene mutations in Polish patients with Alagille syndrome. Journal of Applied Genetics, 2014, 55, 329-336.	1.9	24
80	Duplication 9q34â†'qter identified by chromosome painting. American Journal of Medical Genetics Part A, 1993, 45, 609-613.	2.4	23
81	Mosaicism for a chromosome 8â€derived minute marker chromosome in a patient with manifestations of trisomy 8 mosaicism. American Journal of Medical Genetics Part A, 1995, 56, 22-24.	2.4	23
82	Boy with bilateral retinoblastoma due to an unusual ring chromosome 13 with activation of a latent centromere. American Journal of Medical Genetics Part A, 2001, 99, 21-28.	2.4	22
83	Discordant clinical phenotype in monozygotic twins with Alagille syndrome: Possible influence of nonâ€genetic factors. American Journal of Medical Genetics, Part A, 2016, 170, 471-475.	1.2	22
84	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
85	Utility and limitations of exome sequencing in the molecular diagnosis of pediatric inherited platelet disorders. American Journal of Hematology, 2018, 93, 8-16.	4.1	22
86	Mosaic trisomy 17: Variable clinical and cytogenetic presentation. American Journal of Medical Genetics, Part A, 2011, 155, 2489-2495.	1.2	19
87	â€œSet in Stoneâ€• or â€œRay of Hopeâ€: Parentsâ€™ Beliefs About Cause and Prognosis After Genomic Testing of Children Diagnosed with ASD. Journal of Autism and Developmental Disorders, 2017, 47, 1453-1463.	2.7	18
88	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
89	46, XX, 15p+ documented as dup (17p) by fluorescence in situ hybridization. American Journal of Medical Genetics Part A, 1993, 46, 95-97.	2.4	17
90	Rapid and accurate interpretation of clinical exomes using Phenoxome: a computational phenotype-driven approach. European Journal of Human Genetics, 2019, 27, 612-620.	2.8	17

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91	Exome Sequencing in Individuals with Isolated Biliary Atresia. Scientific Reports, 2020, 10, 2709.	3.3	17
92	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
93	Diploid/triploid mixoploidy: A consequence of asymmetric zygotic segregation of parental genomes. American Journal of Medical Genetics, Part A, 2018, 176, 2720-2732.	1.2	16
94	Alagille Syndrome. , 2007, , 326-345.		15
95	Genome-Wide Expression Analysis in Fibroblast Cell Lines from Probands with Pallister Killian Syndrome. PLoS ONE, 2014, 9, e108853.	2.5	14
96	Intracytoplasmic sperm injection (ICSI) with transmission of a ring(Y) chromosome and ovotesticular disorder of sex development in offspring. American Journal of Medical Genetics, Part A, 2008, 146A, 1828-1831.	1.2	13
97	Developmental profile in a patient with monosomy 10q and dup(17p) associated with a peripheral neuropathy. , 1996, 61, 377-381.		12
98	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
99	Complete androgen insensitivity syndrome due to X chromosome inversion: A clinical report. American Journal of Medical Genetics Part A, 2003, 120A, 434-436.	2.4	10
100	Patient with trisomy 9p and a hypoplastic left heart with a tricentric chromosome 9. American Journal of Medical Genetics Part A, 2003, 123A, 279-284.	2.4	10
101	Alagille syndrome. , 2014, , 216-233.		10
102	Exome sequencing reveals compound heterozygous mutations in ATP8B1 in a JAG1/NOTCH2 mutationâ€negative patient with clinically diagnosed Alagille syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 891-893.	1.2	9
103	Impaired Redox and Protein Homeostasis as Risk Factors and Therapeutic Targets in Toxin-Induced Biliary Atresia. Gastroenterology, 2020, 159, 1068-1084.e2.	1.3	9
104	De novo interstitial deletion of the long arm of chromosome 3:46, XX, del(3) (q25.1q26.1). Clinical Genetics, 1993, 44, 335-337.	2.0	8
105	Placental mosaicism in a case of 46,XY, â˜22, +t(22;22)(p11;q11) or i(22q) diagnosed at amniocentesis. Prenatal Diagnosis, 1992, 12, 47-51.	2.3	7
106	A patient with mosaic partial trisomy 18 resulting from dicentric chromosome breakage. American Journal of Medical Genetics, Part A, 2005, 137A, 208-212.	1.2	7
107	CMIP haploinsufficiency in two patients with autism spectrum disorder and coâ€occurring gastrointestinal issues. American Journal of Medical Genetics, Part A, 2017, 173, 2101-2107.	1.2	6
108	Genetics of Alagille syndrome. Progress in Pediatric Cardiology, 2005, 20, 169-176.	0.4	5

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109	Utility and limitations of exome sequencing as a genetic diagnostic tool for conditions associated with pediatric sudden cardiac arrest/sudden cardiac death. Human Genomics, 2015, 9, 15.	2.9	5
110	Advanced approach for comprehensive mtDNA genome testing in mitochondrial disease. Molecular Genetics and Metabolism, 2022, 135, 93-101.	1.1	5
111	Detection of mutually exclusive mosaicism in a girl with genotypeâ€phenotype discrepancies. American Journal of Medical Genetics, Part A, 2015, 167, 3091-3095.	1.2	4
112	The Cardiovascular Manifestations of Alagille Syndrome and JAG1 Mutations. , 2006, 126, 217-232.		3
113	Genetics of Alagille Syndrome. , 2018, , 33-48.		3
114	Ablepharon macrostomia syndrome with associated cutis laxa: possible localization to 18q. Human Genetics, 1996, 97, 532-536.	3.8	3
115	Expanded non-invasive prenatal diagnostics. Nature Medicine, 2019, 25, 361-362.	30.7	2
116	Alagille Syndrome. , 2021, , 222-241.		2
117	Alagille Syndrome. , 2015, , 155-165.		1
118	How real are our data? Copy number variation in lymphoblastoid and other cell lines. Human Mutation, 2012, 33, v-v.	2.5	0
119	Cytogenetic highlights and transitions. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 87-91.	1.6	0
120	Back Cover, Volume 40, Issue 12. Human Mutation, 2019, 40, iii.	2.5	0