Nancy B Spinner

List of Publications by Year
in descending order

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	Advanced approach for comprehensive mtDNA genome testing in mitochondrial disease. Molecular Genetics and Metabolism, 2022, 135, 93-101.	1.1	5
2	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
3	Alagille Syndrome. , 2021, , 222-241.		2
4	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
5	Genomic Diagnosis for Pediatric Disorders: Revolution and Evolution. Frontiers in Pediatrics, 2020, 8, 373.	1.9	30
6	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
7	Exome Sequencing in Individuals with Isolated Biliary Atresia. Scientific Reports, 2020, 10, 2709.	3.3	17
8	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
9	Outcomes of Childhood Cholestasis in Alagille Syndrome: Results of a Multicenter Observational Study. Hepatology Communications, 2020, 4, 387-398.	4.3	52
10	Alagille syndrome mutation update: Comprehensive overview ofJAG1andNOTCH2mutation frequencies and insight into missense variant classification. Human Mutation, 2019, 40, 2197-2220.	2.5	84
11	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
12	Use of a Dynamic Genetic Testing Approach for Childhood-Onset Epilepsy. JAMA Network Open, 2019, 2, e192129.	5.9	45
13	Expanded non-invasive prenatal diagnostics. Nature Medicine, 2019, 25, 361-362.	30.7	2
14	Back Cover, Volume 40, Issue 12. Human Mutation, 2019, 40, iii.	2.5	0
15	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
16	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
17	Prenatal DNA Sequencing: Clinical, Counseling, and Diagnostic Laboratory Considerations. Prenatal Diagnosis, 2018, 38, 26-32.	2.3	47
18	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

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19	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
20	Genetics of Alagille Syndrome. , 2018, , 33-48.		3
21	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
22	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
23	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
24	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
25	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
26	Alagille Syndrome: Genetics and Functional Models. Current Pathobiology Reports, 2017, 5, 233-241.	3.4	32
27	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
28	Sequencing-based diagnostics for pediatric genetic diseases: progress and potential. Expert Review of Molecular Diagnostics, 2016, 16, 987-999.	3.1	29
29	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
30	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
31	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
32	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
33	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	6.2	137
34	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	2.4	125
35	Cytogenetic highlights and transitions. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 87-91.	1.6	0
36	Jagged1 (JAG1): Structure, expression, and disease associations. Gene, 2016, 576, 381-384.	2.2	104

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37	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315.	2.4	69
38	Alagille Syndrome. , 2015, , 155-165.		1
39	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
40	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
41	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
42	Moyamoya Syndrome Associated with Alagille Syndrome: Outcome after Surgical Revascularization. Journal of Pediatrics, 2015, 166, 470-473.	1.8	29
43	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
44	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
45	Heterozygous Deletion ofFOXA2Segregates with Disease in a Family with Heterotaxy, Panhypopituitarism, and Biliary Atresia. Human Mutation, 2015, 36, 631-637.	2.5	43
46	Actionable exomic incidental findings in 6503 participants: challenges of variant classification. Genome Research, 2015, 25, 305-315.	5.5	313
47	Alagille syndrome. , 2014, , 216-233.		10
48	Clinical utility gene card for: Alagille Syndrome (ALGS). European Journal of Human Genetics, 2014, 22, 435-435.	2.8	30
49	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
50	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
51	Spectrum of JAG1 gene mutations in Polish patients with Alagille syndrome. Journal of Applied Genetics, 2014, 55, 329-336.	1.9	24
52	Genome-Wide Expression Analysis in Fibroblast Cell Lines from Probands with Pallister Killian Syndrome. PLoS ONE, 2014, 9, e108853.	2.5	14
53	Evidence From Human and Zebrafish That GPC1 Is a Biliary Atresia Susceptibility Gene. Gastroenterology, 2013, 144, 1107-1115.e3.	1.3	125
54	A genomic view of mosaicism and human disease. Nature Reviews Genetics, 2013, 14, 307-320.	16.3	527

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55	Renal involvement and the role of Notch signalling in Alagille syndrome. Nature Reviews Nephrology, 2013, 9, 409-418.	9.6	75
56	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
57	Notch signaling in human development and disease. Seminars in Cell and Developmental Biology, 2012, 23, 450-457.	5.0	286
58	Ring chromosome 20. European Journal of Medical Genetics, 2012, 55, 381-387.	1.3	40
59	How real are our data? Copy number variation in lymphoblastoid and other cell lines. Human Mutation, 2012, 33, v-v.	2.5	0
60	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
61	Renal anomalies in Alagille syndrome: A diseaseâ€defining feature. American Journal of Medical Genetics, Part A, 2012, 158A, 85-89.	1.2	102
62	Mosaic trisomy 17: Variable clinical and cytogenetic presentation. American Journal of Medical Genetics, Part A, 2011, 155, 2489-2495.	1.2	19
63	Molecular analysis of ring chromosome 20 syndrome reveals two distinct groups of patients. Journal of Medical Genetics, 2011, 48, 1-9.	3.2	61
64	Pathologic Lower Extremity Fractures in Children With Alagille Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2010, 51, 66-70.	1.8	69
65	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
66	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
67	Jagged1 (JAG1) mutations in patients with tetralogy of fallot or pulmonic stenosis. Human Mutation, 2010, 31, 594-601.	2.5	113
68	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
69	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
70	Mechanisms of mosaicism, chimerism and uniparental disomy identified by single nucleotide polymorphism array analysis. Human Molecular Genetics, 2010, 19, 1263-1275.	2.9	373
71	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
72	SNP array mapping of chromosome 20p deletions: Genotypes, phenotypes, and copy number variation. Human Mutation, 2009, 30, 371-378.	2.5	61

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73	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
74	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
75	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
76	Alagille Syndrome. , 2007, , 326-345.		15
77	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
78	The Cardiovascular Manifestations of Alagille Syndrome and JAG1 Mutations. , 2006, 126, 217-232.		3
79	Intracranial Vascular Abnormalities in Patients with Alagille Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2005, 41, 99-107.	1.8	101
80	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
81	Genetics of Alagille syndrome. Progress in Pediatric Cardiology, 2005, 20, 169-176.	0.4	5
82	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
83	Vascular Anomalies in Alagille Syndrome. Circulation, 2004, 109, 1354-1358.	1.6	333
84	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
85	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
86	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
87	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
88	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
89	Facial features in Alagille syndrome: Specific or cholestasis facies?. American Journal of Medical Genetics Part A, 2002, 112, 163-170.	2.4	101
90	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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91	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
92	Down syndrome congenital heart disease: A narrowed region and a candidate gene. Genetics in Medicine, 2001, 3, 91-101.	2.4	168
93	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
94	Jagged1 mutations in Alagille syndrome. Human Mutation, 2001, 17, 18-33.	2.5	224
95	Mutation analysis of Jagged1 (JAG1) in Alagille syndrome patients. Human Mutation, 2001, 17, 151-152.	2.5	76
96	Defective intracellular transport and processing of JAG1 missense mutations in Alagille syndrome. Human Molecular Genetics, 2001, 10, 405-413.	2.9	76
97	Alagille Syndrome and the Jagged1 Gene. Seminars in Liver Disease, 2001, 21, 525-534.	3.6	127
98	Jagged1 (JAG1) mutation detection in an Australian Alagille syndrome population. Human Mutation, 2000, 16, 408-416.	2.5	35
99	CADASIL: Notch signaling defect or protein accumulation problem?. Journal of Clinical Investigation, 2000, 105, 561-562.	8.2	38
100	Features of alagille syndrome in 92 patients: Frequency and relation to prognosis. Hepatology, 1999, 29, 822-829.	7.3	591
101	Jagged1 mutations in patients ascertained with isolated congenital heart defects. , 1999, 84, 56-60.		137
102	Clinical and molecular genetics of Alagille syndrome. Current Opinion in Pediatrics, 1999, 11, 558-564.	2.0	96
103	GLUT-1 deficiency syndrome caused by haploinsufficiency of the blood-brain barrier hexose carrier. Nature Genetics, 1998, 18, 188-191.	21.4	349
104	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
105	Panhandle Polymerase Chain Reaction Amplifies MLL Genomic Translocation Breakpoint Involving Unknown Partner Gene. Blood, 1997, 90, 4679-4686.	1.4	32
106	Mutations in the human Jagged1 gene are responsible for Alagille syndrome. Nature Genetics, 1997, 16, 235-242.	21.4	1,072
107	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
108	KILLER/DR5 is a DNA damageâ€“inducible p53â€“regulated death receptor gene. Nature Genetics, 1997, 17, 141-143.	21.4	1,005

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109	Deletions of 20p12 in Alagille syndrome: Frequency and molecular characterization. , 1997, 70, 80-86.		71
110	Ablepharon macrostomia syndrome with associated cutis laxa: Possible localization to 18q. Human Genetics, 1996, 97, 532-536.	3.8	32
111	Developmental profile in a patient with monosomy 10q and dup(17p) associated with a peripheral neuropathy. , 1996, 61, 377-381.		12
112	Ablepharon macrostomia syndrome with associated cutis laxa: possible localization to 18q. Human Genetics, 1996, 97, 532-536.	3.8	3
113	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
114	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
115	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
116	Duplication 9q34â†'qter identified by chromosome painting. American Journal of Medical Genetics Part A, 1993, 45, 609-613.	2.4	23
117	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
118	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
119	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
120	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