

# Jennifer M Kalish

## List of Publications by Year in descending order

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Version: 2024-02-01

65  
papers

1,987  
citations

304743

22  
h-index

265206

42  
g-index

66  
all docs

66  
docs citations

66  
times ranked

2239  
citing authors

#	ARTICLE	IF	CITATIONS
1	Molecular networks of hepatoblastoma predisposition and oncogenesis in Beckwith-Wiedemann syndrome. <i>Hepatology Communications</i> , 2022, 6, 2132-2146.	4.3	8
2	Clinical Effectiveness of Telemedicine-Based Pediatric Genetics Care. <i>Pediatrics</i> , 2022, 150, .	2.1	5
3	Derivation and investigation of the first human cell-based model of Beckwith-Wiedemann syndrome. <i>Epigenetics</i> , 2021, 16, 1295-1305.	2.7	4
4	Improved molecular detection of mosaicism in Beckwith-Wiedemann Syndrome. <i>Journal of Medical Genetics</i> , 2021, 58, 178-184.	3.2	17
5	A dyadic approach to the delineation of diagnostic entities in clinical genomics. <i>American Journal of Human Genetics</i> , 2021, 108, 8-15.	6.2	71
6	The Prevalence of Difficult Airway in Children With Beckwith-Wiedemann Syndrome: A Retrospective Cohort Study. <i>Anesthesia and Analgesia</i> , 2021, 133, 1559-1567.	2.2	5
7	Prenatal molecular testing and diagnosis of Beckwith-Wiedemann syndrome. <i>Prenatal Diagnosis</i> , 2021, 41, 817-822.	2.3	8
8	A case of macrosomia and macroglossia, likely Beckwith-Wiedemann syndrome from 1628. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2021, 187, 173-175.	1.6	2
9	The role of CTCF in the organization of the centromeric 11p15 imprinted domain interactome. <i>Nucleic Acids Research</i> , 2021, 49, 6315-6330.	14.5	11
10	Expanding the genetic landscape of oral-facial-digital syndrome with two novel genes. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2409-2416.	1.2	9
11	Understanding Syndromic Leg Length Discrepancy. <i>Journal of Pediatrics</i> , 2021, 234, 16-18.	1.8	0
12	Case Report: Two Distinct Focal Congenital Hyperinsulinism Lesions Resulting From Separate Genetic Events. <i>Frontiers in Pediatrics</i> , 2021, 9, 699129.	1.9	3
13	The following 3 cases were presented at the 2020 virtual PUOWG conference: Late Presentation of Wilms Tumor in a Patient with Hemihypertrophy after Normal Screening. <i>Urology</i> , 2021, 154, 271-274.	1.0	1
14	Response to Hamosh et al.. <i>American Journal of Human Genetics</i> , 2021, 108, 1809-1810.	6.2	0
15	Epigenetic mosaicism and cell burden in Beckwith-Wiedemann Syndrome due to loss of methylation at imprinting control region 2. <i>Journal of Physical Education and Sports Management</i> , 2021, 7, mcs.a006115.	1.2	7
16	Characteristics Associated with Tumor Development in Individuals Diagnosed with Beckwith-Wiedemann Spectrum: Novel Tumor-(epi)Genotype-Phenotype Associations in the BWS Population. <i>Genes</i> , 2021, 12, 1839.	2.4	9
17	Results From the WAGR Syndrome Patient Registry: Characterization of WAGR Spectrum and Recommendations for Care Management. <i>Frontiers in Pediatrics</i> , 2021, 9, 733018.	1.9	13
18	Reply. <i>Journal of Pediatrics</i> , 2020, 216, 249-250.	1.8	0

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19	Novel variants in <i>CDH2</i> are associated with a new syndrome including Peters anomaly. <i>Clinical Genetics</i> , 2020, 97, 502-508.	2.0	13
20	Coexistence of paternally-inherited ABCC8 mutation and mosaic paternal uniparental disomy 11p hyperinsulinism. <i>International Journal of Pediatric Endocrinology (Springer)</i> , 2020, 2020, 13.	1.6	4
21	The Utility of Early Tongue Reduction Surgery for Macroglossia in Beckwith-Wiedemann Syndrome. <i>Plastic and Reconstructive Surgery</i> , 2020, 145, 803e-813e.	1.4	22
22	Tatton-Brown-Rahman syndrome: Six individuals with novel features. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 673-680.	1.2	11
23	11p15.5 epimutations in children with Wilms tumor and hepatoblastoma detected in peripheral blood. <i>Cancer</i> , 2020, 126, 3114-3121.	4.1	23
24	Characterization and Childhood Tumor Risk Assessment of Genetic and Epigenetic Syndromes Associated With Lateralized Overgrowth. <i>Frontiers in Pediatrics</i> , 2020, 8, 613260.	1.9	14
25	Development of the Serum $\alpha$ -Fetoprotein Reference Range in Patients with Beckwith-Wiedemann Spectrum. <i>Journal of Pediatrics</i> , 2019, 212, 195-200.e2.	1.8	11
26	Characterization of the Beckwith-Wiedemann spectrum: Diagnosis and management. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2019, 181, 693-708.	1.6	62
27	The effectiveness of Wilms tumor screening in Beckwith-Wiedemann spectrum. <i>Journal of Cancer Research and Clinical Oncology</i> , 2019, 145, 3115-3123.	2.5	25
28	Androgenetic chimerism as an etiology for Beckwith-Wiedemann syndrome: diagnosis and management. <i>Genetics in Medicine</i> , 2019, 21, 2644-2649.	2.4	15
29	Altered microRNA expression profiles in large offspring syndrome and Beckwith-Wiedemann syndrome. <i>Epigenetics</i> , 2019, 14, 850-876.	2.7	32
30	Diagnosis and management of the phenotypic spectrum of twins with Beckwith-Wiedemann syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1139-1147.	1.2	17
31	Cover Image, Volume 179A, Number 4, April 2019. , 2019, 179, i-i.		4
32	Hyperinsulinemic hypoglycemia in seven patients with de novo <i>NSD1</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 542-551.	1.2	16
33	Pediatric chondrodermatitis nodularis helioides ( <i>CNH</i> ) in a child with Beckwith-Wiedemann syndrome ( <i>BWS</i> ). <i>Pediatric Dermatology</i> , 2019, 36, 388-390.	0.9	2
34	Beckwith-Wiedemann syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 525-533.	1.2	18
35	Molecular diagnosis of somatic overgrowth conditions: A single-center experience. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e536.	1.2	28
36	Obstructive Sleep Apnea in Children With Beckwith-Wiedemann Syndrome. <i>Journal of Clinical Sleep Medicine</i> , 2019, 15, 375-381.	2.6	15

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37	Allele-specific RNA imaging shows that allelic imbalances can arise in tissues through transcriptional bursting. <i>PLoS Genetics</i> , 2019, 15, e1007874.	3.5	52
38	Longitudinal Monitoring of Alpha-Fetoprotein by Dried Blood Spot for Hepatoblastoma Screening in Beckwith-Wiedemann Syndrome. <i>Cancers</i> , 2019, 11, 86.	3.7	7
39	Syndromic Causes of Congenital Hyperinsulinism. <i>Contemporary Endocrinology</i> , 2019, , 49-59.	0.1	1
40	Defining an optimal time window to screen for hepatoblastoma in children with Beckwith-Wiedemann syndrome. <i>Pediatric Blood and Cancer</i> , 2019, 66, e27492.	1.5	23
41	Diagnosis and Management of Beckwith-Wiedemann Syndrome. <i>Frontiers in Pediatrics</i> , 2019, 7, 562.	1.9	73
42	Beckwith-Wiedemann Syndrome: Partnership in the Diagnostic Journey of a Rare Disorder. <i>Pediatrics</i> , 2018, 141, e20170475.	2.1	2
43	Clinical and molecular diagnosis, screening and management of Beckwith-Wiedemann syndrome: an international consensus statement. <i>Nature Reviews Endocrinology</i> , 2018, 14, 229-249.	9.6	388
44	Tumor Screening in Beckwith-Wiedemann Syndrome: Parental Perspectives. <i>Journal of Genetic Counseling</i> , 2018, 27, 844-853.	1.6	19
45	Obstructive sleep apnoea and the role of tongue reduction surgery in children with Beckwith-Wiedemann syndrome. <i>Paediatric Respiratory Reviews</i> , 2018, 25, 58-63.	1.8	14
46	Diagnosis of Beckwith-Wiedemann syndrome in children presenting with Wilms tumor. <i>Pediatric Blood and Cancer</i> , 2018, 65, e27296.	1.5	32
47	Nonreentrant atrial tachycardia occurs independently of hypertrophic cardiomyopathy in RASopathy patients. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1711-1722.	1.2	21
48	Management of adrenal masses in patients with Beckwith-Wiedemann syndrome. <i>Pediatric Blood and Cancer</i> , 2017, 64, e26432.	1.5	22
49	The utility of alpha-fetoprotein screening in Beckwith-Wiedemann syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 581-584.	1.2	14
50	Nomenclature and definition in asymmetric regional body overgrowth. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1735-1738.	1.2	36
51	Recommendations for Cancer Surveillance in Individuals with RASopathies and Other Rare Genetic Conditions with Increased Cancer Risk. <i>Clinical Cancer Research</i> , 2017, 23, e83-e90.	7.0	122
52	Cover Image, Volume 173A, Number 7, July 2017. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, i.	1.2	0
53	Surveillance Recommendations for Children with Overgrowth Syndromes and Predisposition to Wilms Tumors and Hepatoblastoma. <i>Clinical Cancer Research</i> , 2017, 23, e115-e122.	7.0	140
54	Urological Findings in Beckwith-Wiedemann Syndrome With Chromosomal Duplications of 11p15.5: Evaluation and Management. <i>Urology</i> , 2017, 100, 224-227.	1.0	5

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55	An Atypical Presentation of a Male with Oral-Facial-Digital Syndrome Type 1 Related Ciliopathy. Case Reports in Nephrology, 2016, 2016, 1-4.	0.4	8
56	Congenital hyperinsulinism in children with paternal 11p uniparental isodisomy and Beckwithâ€™s Wiedemann syndrome. Journal of Medical Genetics, 2016, 53, 53-61.	3.2	76
57	Tumor screening in Beckwithâ€™s Wiedemann syndromeâ€™To screen or not to screen?. American Journal of Medical Genetics, Part A, 2016, 170, 2261-2264.	1.2	28
58	Visualizing allele-specific expression in single cells reveals epigenetic mosaicism in an <i>H19</i> loss-of-imprinting mutant. Genes and Development, 2016, 30, 567-578.	5.9	38
59	Overgrowth Syndromes. Journal of Pediatric Genetics, 2015, 04, 136-143.	0.7	57
60	Epigenetics and imprinting in human disease. International Journal of Developmental Biology, 2014, 58, 291-298.	0.6	103
61	Clinical features of three girls with mosaic genome-wide paternal uniparental isodisomy. American Journal of Medical Genetics, Part A, 2013, 161, 1929-1939.	1.2	63
62	Bilateral Pheochromocytomas, Hemihyperplasia, and Subtle Somatic Mosaicism: The Importance of Detecting Low-level Uniparental Disomy. American Journal of Medical Genetics, Part A, 2013, 161, 993-1001.	1.2	21
63	Triplex-Stimulated Intermolecular Recombination at a Single-Copy Genomic Target. Molecular Therapy, 2006, 14, 392-400.	8.2	37
64	Targeted Genome Modification via Triple Helix Formation. Annals of the New York Academy of Sciences, 2005, 1058, 151-161.	3.8	41
65	Triplex-induced recombination and repair in the pyrimidine motif. Nucleic Acids Research, 2005, 33, 3492-3502.	14.5	39