

# Frederic Brioude

## List of Publications by Year in descending order

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Version: 2024-02-01

51  
papers

2,151  
citations

346980

22  
h-index

263392

45  
g-index

54  
all docs

54  
docs citations

54  
times ranked

2685  
citing authors

#	ARTICLE	IF	CITATIONS
1	Low Maternal DLK1 Levels at 26 Weeks Is Associated With Small for Gestational Age at Birth. <i>Frontiers in Endocrinology</i> , 2022, 13, 836731.	1.5	1
2	IGF2: Development, Genetic and Epigenetic Abnormalities. <i>Cells</i> , 2022, 11, 1886.	1.8	18
3	Screening of patients born small for gestational age with the Silver-Russell syndrome phenotype for DLK1 variants. <i>European Journal of Human Genetics</i> , 2021, 29, 1756-1761.	1.4	2
4	Fertility preservation in young men with Klinefelter syndrome: A systematic review. <i>Journal of Gynecology Obstetrics and Human Reproduction</i> , 2021, 50, 102177.	0.6	9
5	Unilateral nephrocalcinosis. <i>Kidney International</i> , 2021, 100, 1145.	2.6	0
6	Increasing knowledge in <i>IGF1R</i> defects: lessons from 35 new patients. <i>Journal of Medical Genetics</i> , 2020, 57, 160-168.	1.5	20
7	Roles of Type 1 Insulin-Like Growth Factor (IGF) Receptor and IGF-II in Growth Regulation: Evidence From a Patient Carrying Both an 11p Paternal Duplication and 15q Deletion. <i>Frontiers in Endocrinology</i> , 2019, 10, 263.	1.5	10
8	CUGC for Simpson-Golabi-Behmel syndrome (SGBS). <i>European Journal of Human Genetics</i> , 2019, 27, 663-668.	1.4	9
9	Sleep disordered breathing in Silver-Russell syndrome patients: a new outcome. <i>Sleep Medicine</i> , 2019, 64, 23-29.	0.8	5
10	Contribution of functionally assessed <i>GHRHR</i> mutations to idiopathic isolated growth hormone deficiency in patients without <i>GH1</i> mutations. <i>Human Mutation</i> , 2019, 40, 2033-2043.	1.1	9
11	Overgrowth syndromes – clinical and molecular aspects and tumour risk. <i>Nature Reviews Endocrinology</i> , 2019, 15, 299-311.	4.3	59
12	Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. <i>Genetical Research</i> , 2019, 101, e3.	0.3	17
13	Transcriptional profiling at the <i>DLK1/MEG3</i> domain explains clinical overlap between imprinting disorders. <i>Science Advances</i> , 2019, 5, eaau9425.	4.7	29
14	Revisiting Wilms tumour surveillance in Beckwith-Wiedemann syndrome with IC2 methylation loss, reply. <i>European Journal of Human Genetics</i> , 2018, 26, 471-472.	1.4	13
15	Diagnosis and management of postnatal fetal growth restriction. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2018, 32, 523-534.	2.2	23
16	Mutation update for the <i>GPC3</i> gene involved in Simpson-Golabi-Behmel syndrome and review of the literature. <i>Human Mutation</i> , 2018, 39, 790-805.	1.1	24
17	Clinical and molecular diagnosis, screening and management of Beckwith-Wiedemann syndrome: an international consensus statement. <i>Nature Reviews Endocrinology</i> , 2018, 14, 229-249.	4.3	388
18	CHARGE syndrome: a recurrent hotspot of mutations in CHD7 IVS25 analyzed by bioinformatic tools and minigene assays. <i>European Journal of Human Genetics</i> , 2018, 26, 287-292.	1.4	7

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19	Prediction of Neonatal Hyperthyroidism. <i>Journal of Pediatrics</i> , 2018, 197, 249-254.e1.	0.9	21
20	Genetic disruption of the oncogenic HMGA2-PLAG1-IGF2 pathway causes fetal growth restriction. <i>Genetics in Medicine</i> , 2018, 20, 250-258.	1.1	99
21	Chromosomal rearrangements in the 11p15 imprinted region: 17 new 11p15.5 duplications with associated phenotypes and putative functional consequences. <i>Journal of Medical Genetics</i> , 2018, 55, 205-213.	1.5	36
22	Placental Pathology in Beckwith-Wiedemann Syndrome According to Genotype/Epigenotype Subgroups. <i>Fetal and Pediatric Pathology</i> , 2018, 37, 387-399.	0.4	11
23	Corpus Callosum Abnormalities and Short Femurs in Beckwith-Wiedemann Syndrome: A Report of Two Fetal Cases. <i>Fetal and Pediatric Pathology</i> , 2018, 37, 411-417.	0.4	2
24	Chromosome 14q32.2 Imprinted Region Disruption as an Alternative Molecular Diagnosis of Silver-Russell Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 2436-2446.	1.8	48
25	Comment on: Juvenile granulosa cell ovarian tumor in a child with Beckwith-Wiedemann syndrome. <i>Pediatric Blood and Cancer</i> , 2017, 64, e26452.	0.8	1
26	Imprinted disorders and growth. <i>Annales D'Endocrinologie</i> , 2017, 78, 112-113.	0.6	9
27	Nomenclature and definition in asymmetric regional body overgrowth. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1735-1738.	0.7	36
28	Study of the Factors Leading to Fetal and Neonatal Dysthyroidism in Children of Patients With Graves Disease. <i>Journal of the Endocrine Society</i> , 2017, 1, 751-761.	0.1	30
29	Contribution of LHX4 Mutations to Pituitary Deficits in a Cohort of 417 Unrelated Patients. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 290-301.	1.8	19
30	11p15 ICR1 Partial Deletions Associated with IGF2/H19 DMR Hypomethylation and Silver-Russell Syndrome. <i>Human Mutation</i> , 2017, 38, 105-111.	1.1	28
31	Diagnosis and management of Silver-Russell syndrome: first international consensus statement. <i>Nature Reviews Endocrinology</i> , 2017, 13, 105-124.	4.3	336
32	Formation of upd(7)mat by trisomic rescue: SNP array typing provides new insights in chromosomal nondisjunction. <i>Molecular Cytogenetics</i> , 2017, 10, 28.	0.4	10
33	Is Nephron Sparing Surgery Justified in Wilms Tumor With Beckwith-Wiedemann Syndrome or Isolated Hemihypertrophy?. <i>Pediatric Blood and Cancer</i> , 2016, 63, 1571-1577.	0.8	19
34	EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silver-Russell and Beckwith-Wiedemann syndrome. <i>European Journal of Human Genetics</i> , 2016, 24, 1377-1387.	1.4	68
35	Hypercortisolism due to a Pituitary Adenoma Associated with Beckwith-Wiedemann Syndrome. <i>Hormone Research in Paediatrics</i> , 2016, 86, 206-211.	0.8	11
36	New clinical and molecular insights into Silver-Russell syndrome. <i>Current Opinion in Pediatrics</i> , 2016, 28, 529-535.	1.0	14

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37	Prenatal molecular testing for Beckwith-Wiedemann and Silver-Russell syndromes: a challenge for molecular analysis and genetic counseling. <i>European Journal of Human Genetics</i> , 2016, 24, 784-793.	1.4	44
38	IGF2, H19, CDKN1C, KCNQ1OT1, and the Beckwith-Wiedemann Syndrome. , 2016, , 965-970.		0
39	Sleep disordered breathing in patients with Prader-Willi syndrome: A multicenter study. <i>Pediatric Pulmonology</i> , 2015, 50, 1354-1359.	1.0	68
40	Mutations of the Imprinted CDKN1C Gene as a Cause of the Overgrowth Beckwith-Wiedemann Syndrome: Clinical Spectrum and Functional Characterization. <i>Human Mutation</i> , 2015, 36, 894-902.	1.1	62
41	Extensive investigation of the IGF2/H19 imprinting control region reveals novel OCT4/SOX2 binding site defects associated with specific methylation patterns in Beckwith-Wiedemann syndrome. <i>Human Molecular Genetics</i> , 2014, 23, 5763-5773.	1.4	58
42	CDKN1C mutations: two sides of the same coin. <i>Trends in Molecular Medicine</i> , 2014, 20, 614-622.	3.5	89
43	Complex Tissue-Specific Epigenotypes in Russell-Silver Syndrome Associated with 11p15 ICR1 Hypomethylation. <i>Human Mutation</i> , 2014, 35, 1211-1220.	1.1	34
44	Human Imprinting Anomalies in Fetal and Childhood Growth Disorders: Clinical Implications and Molecular Mechanisms. <i>Current Pharmaceutical Design</i> , 2014, 20, 1751-1763.	0.9	12
45	SNP arrays in Beckwith-Wiedemann syndrome: An improved diagnostic strategy. <i>European Journal of Medical Genetics</i> , 2013, 56, 546-550.	0.7	34
46	Two Families with Normosmic Congenital Hypogonadotropic Hypogonadism and Biallelic Mutations in KISS1R (KISS1 Receptor): Clinical Evaluation and Molecular Characterization of a Novel Mutation. <i>PLoS ONE</i> , 2013, 8, e53896.	1.1	38
47	SEMA3A deletion in a family with Kallmann syndrome validates the role of semaphorin 3A in human puberty and olfactory system development. <i>Human Reproduction</i> , 2012, 27, 1460-1465.	0.4	133
48	Imprinted Anomalies in Fetal and Childhood Growth Disorders: The Model of Russell-Silver and Beckwith-Wiedemann Syndromes. <i>Endocrine Development</i> , 2012, 23, 60-70.	1.3	18
49	Existe-t-il une relation entre la dose d'hormone de croissance et d'éventuelles complications tumorales ou cardiovasculaires ?. <i>Bulletin De L'Academie Nationale De Medecine</i> , 2012, 196, 127-137.	0.0	0
50	Non-syndromic congenital hypogonadotropic hypogonadism: clinical presentation and genotype-phenotype relationships. <i>European Journal of Endocrinology</i> , 2010, 162, 835-851.	1.9	104
51	Anomalies épigénétiques et de l'empreinte parentale dans les maladies du développement humain. <i>Bulletin De L'Academie Nationale De Medecine</i> , 2010, 194, 287-300.	0.0	2