

Deborah J G Mackay

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

80
papers

5,670
citations

38
h-index

75
g-index

84
ext. papers

6,527
ext. citations

7.5
avg, IF

5.09
L-index

#	Paper	IF	Citations
80	Activating mutations in the gene encoding the ATP-sensitive potassium-channel subunit Kir6.2 and permanent neonatal diabetes. <i>New England Journal of Medicine</i> , 2004 , 350, 1838-49	59.2	930
79	Hypomethylation of multiple imprinted loci in individuals with transient neonatal diabetes is associated with mutations in ZFP57. <i>Nature Genetics</i> , 2008 , 40, 949-51	36.3	417
78	Mutations in ATP-sensitive K ⁺ channel genes cause transient neonatal diabetes and permanent diabetes in childhood or adulthood. <i>Diabetes</i> , 2007 , 56, 1930-7	0.9	273
77	Expert consensus document: Clinical and molecular diagnosis, screening and management of Beckwith-Wiedemann syndrome: an international consensus statement. <i>Nature Reviews Endocrinology</i> , 2018 , 14, 229-249	15.2	234
76	Diagnosis and management of Silver-Russell syndrome: first international consensus statement. <i>Nature Reviews Endocrinology</i> , 2017 , 13, 105-124	15.2	224
75	Transient neonatal diabetes: widening the understanding of the etiopathogenesis of diabetes. <i>Diabetes</i> , 2000 , 49, 1359-66	0.9	214
74	The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. <i>Lancet, The</i> , 2015 , 386, 957-63	40	186
73	Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwith-Wiedemann syndrome. <i>European Journal of Human Genetics</i> , 2009 , 17, 611-9	5.3	176
72	Relapsing diabetes can result from moderately activating mutations in KCNJ11. <i>Human Molecular Genetics</i> , 2005 , 14, 925-34	5.6	165
71	Permanent neonatal diabetes caused by dominant, recessive, or compound heterozygous SUR1 mutations with opposite functional effects. <i>American Journal of Human Genetics</i> , 2007 , 81, 375-82	11	161
70	Genomic imprinting disorders: lessons on how genome, epigenome and environment interact. <i>Nature Reviews Genetics</i> , 2019 , 20, 235-248	30.1	151
69	A maternal hypomethylation syndrome presenting as transient neonatal diabetes mellitus. <i>Human Genetics</i> , 2006 , 120, 262-9	6.3	139
68	Temple syndrome: improving the recognition of an underdiagnosed chromosome 14 imprinting disorder: an analysis of 51 published cases. <i>Journal of Medical Genetics</i> , 2014 , 51, 495-501	5.8	136
67	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. <i>Clinical Epigenetics</i> , 2015 , 7, 123	7.7	115
66	The RhoS progress: a potential role during neuritogenesis for the Rho family of GTPases. <i>Trends in Neurosciences</i> , 1995 , 18, 496-501	13.3	110
65	Mutations in NLRP5 are associated with reproductive wastage and multilocus imprinting disorders in humans. <i>Nature Communications</i> , 2015 , 6, 8086	17.4	102
64	Epigenotype-phenotype correlations in Silver-Russell syndrome. <i>Journal of Medical Genetics</i> , 2010 , 47, 760-8	5.8	98

63	Clinical presentation of 6q24 transient neonatal diabetes mellitus (6q24 TNDM) and genotype-phenotype correlation in an international cohort of patients. <i>Diabetologia</i> , 2013 , 56, 758-62	10.3	90
62	Insights into the molecular mechanism for type 2 diabetes susceptibility at the KCNQ1 locus from temporal changes in imprinting status in human islets. <i>Diabetes</i> , 2013 , 62, 987-92	0.9	87
61	Transient neonatal diabetes mellitus type 1. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2010 , 154C, 335-42	3.1	81
60	Methylation analysis of 79 patients with growth restriction reveals novel patterns of methylation change at imprinted loci. <i>European Journal of Human Genetics</i> , 2010 , 18, 648-55	5.3	80
59	Genome-wide DNA methylation analysis of patients with imprinting disorders identifies differentially methylated regions associated with novel candidate imprinted genes. <i>Journal of Medical Genetics</i> , 2014 , 51, 229-38	5.8	70
58	Relaxation of imprinted expression of ZAC and HYMAI in a patient with transient neonatal diabetes mellitus. <i>Human Genetics</i> , 2002 , 110, 139-44	6.3	70
57	Maternal variants in and other maternal effect proteins are associated with multilocus imprinting disturbance in offspring. <i>Journal of Medical Genetics</i> , 2018 , 55, 497-504	5.8	66
56	Targeted methylation testing of a patient cohort broadens the epigenetic and clinical description of imprinting disorders. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 2174-82	2.5	65
55	Recent Advances in Imprinting Disorders. <i>Clinical Genetics</i> , 2017 , 91, 3-13	4	62
54	Isolated imprinting mutation of the DLK1/GTL2 locus associated with a clinical presentation of maternal uniparental disomy of chromosome 14. <i>Journal of Medical Genetics</i> , 2007 , 44, 637-40	5.8	54
53	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, 1377-87	5.3	54
52	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. <i>Cell</i> , 2019 , 177, 32-37	56.2	53
51	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. <i>Genetics in Medicine</i> , 2016 , 18, 309-15	8.1	52
50	Characterization of the methylation-sensitive promoter of the imprinted ZAC gene supports its role in transient neonatal diabetes mellitus. <i>Journal of Biological Chemistry</i> , 2001 , 276, 18653-6	5.4	52
49	Beckwith-Wiedemann syndrome caused by maternally inherited mutation of an OCT-binding motif in the IGF2/H19-imprinting control region, ICR1. <i>European Journal of Human Genetics</i> , 2012 , 20, 240-3	5.3	51
48	Epimutation of the TNDM locus and the Beckwith-Wiedemann syndrome centromeric locus in individuals with transient neonatal diabetes mellitus. <i>Human Genetics</i> , 2006 , 119, 179-84	6.3	51
47	Bisulphite sequencing of the transient neonatal diabetes mellitus DMR facilitates a novel diagnostic test but reveals no methylation anomalies in patients of unknown aetiology. <i>Human Genetics</i> , 2005 , 116, 255-61	6.3	51
46	Mutation of HERC2 causes developmental delay with Angelman-like features. <i>Journal of Medical Genetics</i> , 2013 , 50, 65-73	5.8	50

45	Transient neonatal diabetes, ZFP57, and hypomethylation of multiple imprinted loci: a detailed follow-up. <i>Diabetes Care</i> , 2013 , 36, 505-12	14.6	48
44	Clinical characterisation of the multiple maternal hypomethylation syndrome in siblings. <i>European Journal of Human Genetics</i> , 2008 , 16, 453-61	5.3	46
43	Incidence of neonatal diabetes in Austria-calculation based on the Austrian Diabetes Register. <i>Pediatric Diabetes</i> , 2010 , 11, 18-23	3.6	42
42	Human imprinting disorders: Principles, practice, problems and progress. <i>European Journal of Medical Genetics</i> , 2017 , 60, 618-626	2.6	35
41	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, 784-93	5.3	34
40	Deletions and rearrangements of the H19/IGF2 enhancer region in patients with Silver-Russell syndrome and growth retardation. <i>Journal of Medical Genetics</i> , 2011 , 48, 308-11	5.8	33
39	An atypical case of hypomethylation at multiple imprinted loci. <i>European Journal of Human Genetics</i> , 2011 , 19, 360-2	5.3	33
38	Multilocus methylation defects in imprinting disorders. <i>Biomolecular Concepts</i> , 2015 , 6, 47-57	3.7	31
37	Further refinement of the critical minimal genetic region for the imprinting disorder 6q24 transient neonatal diabetes. <i>Diabetologia</i> , 2010 , 53, 2347-51	10.3	27
36	Neonatal diabetes in Ukraine: incidence, genetics, clinical phenotype and treatment. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2015 , 28, 1279-86	1.6	25
35	Evidence for anticipation in Beckwith-Wiedemann syndrome. <i>European Journal of Human Genetics</i> , 2013 , 21, 1344-8	5.3	23
34	Temple syndrome as a result of isolated hypomethylation of the 14q32 imprinted DLK1/MEG3 region. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 170-5	2.5	22
33	A familial disorder of altered DNA-methylation. <i>Journal of Medical Genetics</i> , 2014 , 51, 407-12	5.8	22
32	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. <i>Genome Research</i> , 2019 , 29, 1057-1066	9.7	20
31	Zellweger syndrome resulting from maternal isodisomy of chromosome 1. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 2172-7	2.5	20
30	DNA hypomethylation, transient neonatal diabetes, and prune belly sequence in one of two identical twins. <i>European Journal of Pediatrics</i> , 2010 , 169, 207-13	4.1	19
29	Role of noninsulin therapies alone or in combination in chromosome 6q24-related transient neonatal diabetes: sulfonylurea improves but does not always normalize insulin secretion. <i>Diabetes Care</i> , 2015 , 38, e86-7	14.6	18
28	Very small deletions within the NESP55 gene in pseudohypoparathyroidism type 1b. <i>European Journal of Human Genetics</i> , 2015 , 23, 494-9	5.3	18

27	Hepatoblastoma in a child with a paternally-inherited ABCC8 mutation and mosaic paternal uniparental disomy 11p causing focal congenital hyperinsulinism. <i>European Journal of Medical Genetics</i> , 2013 , 56, 114-7	2.6	16
26	Familial occurrence of neonatal diabetes with duplications in chromosome 6q24: treatment with sulfonylurea and 40-yr follow-up. <i>Pediatric Diabetes</i> , 2012 , 13, 155-62	3.6	15
25	The phenotypic variations of multi-locus imprinting disturbances associated with maternal-effect variants of NLRP5 range from overt imprinting disorder to apparently healthy phenotype. <i>Clinical Epigenetics</i> , 2019 , 11, 190	7.7	13
24	A statistical method for single sample analysis of HumanMethylation450 array data: genome-wide methylation analysis of patients with imprinting disorders. <i>Clinical Epigenetics</i> , 2015 , 7, 48	7.7	12
23	Mosaic paternal uniparental isodisomy and an ABCC8 gene mutation in a patient with permanent neonatal diabetes and hemihypertrophy. <i>Diabetes</i> , 2008 , 57, 255-8	0.9	12
22	Monozygous triplets discordant for transient neonatal diabetes mellitus and for imprinting of the TNDM differentially methylated region. <i>Human Genetics</i> , 2005 , 117, 398-401	6.3	12
21	Genetic diagnosis of subfertility: the impact of meiosis and maternal effects. <i>Journal of Medical Genetics</i> , 2019 , 56, 271-282	5.8	10
20	Assessment of the role of common genetic variation in the transient neonatal diabetes mellitus (TNDM) region in type 2 diabetes: a comparative genomic and tagging single nucleotide polymorphism approach. <i>Diabetes</i> , 2006 , 55, 2272-6	0.9	10
19	Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. <i>Genetical Research</i> , 2019 , 101, e3	1.1	9
18	Revisiting Wilms tumour surveillance in Beckwith-Wiedemann syndrome with IC2 methylation loss, reply. <i>European Journal of Human Genetics</i> , 2018 , 26, 471-472	5.3	7
17	Lived experience of Silver-Russell syndrome: implications for management during childhood and into adulthood. <i>Archives of Disease in Childhood</i> , 2019 , 104, 76-82	2.2	7
16	Transient Neonatal Diabetes Mellitus followed by recurrent asymptomatic hypoglycaemia: a case report. <i>BMC Pediatrics</i> , 2015 , 15, 200	2.6	7
15	Growth Hormone Improves Short-Term Growth in Patients with Temple Syndrome. <i>Hormone Research in Paediatrics</i> , 2018 , 90, 407-413	3.3	7
14	Report and review of described associations of Mayer-Rokitansky-Küster-Hauser syndrome and Silver-Russell syndrome. <i>Journal of Paediatrics and Child Health</i> , 2015 , 51, 555-560	1.3	6
13	Concurrent course of transient neonatal diabetes with cholestasis and paucity of interlobular bile ducts: a case report. <i>Pediatric and Developmental Pathology</i> , 2009 , 12, 417-20	2.2	6
12	Investigation of 90 patients referred for molecular cytogenetic analysis using aCGH uncovers previously unsuspected anomalies of imprinting. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 1990-3	2.5	6
11	Phenotype of genetically confirmed Silver-Russell syndrome beyond childhood. <i>Journal of Medical Genetics</i> , 2020 , 57, 683-691	5.8	6
10	Transient neonatal diabetes mellitus in a Turkish patient with three novel homozygous variants in the ZFP57 gene. <i>JCRPE Journal of Clinical Research in Pediatric Endocrinology</i> , 2013 , 5, 125-8	1.9	5

9	Maturity onset diabetes of the young (MODY) and early onset Type II diabetes are not caused by loss of imprinting at the transient neonatal diabetes (TNDM) locus. <i>Diabetologia</i> , 2001 , 44, 924	10.3	5
8	Whole-genome analysis as a diagnostic tool for patients referred for diagnosis of Silver-Russell syndrome: a real-world study. <i>Journal of Medical Genetics</i> , 2021 ,	5.8	4
7	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging		2
6	Beyond the gene roundtable discussion. <i>Textual Practice</i> , 2015 , 29, 415-432	0.1	1
5	Genetics, molar pregnancies and medieval ideas of monstrous births: the lump of flesh in. <i>Medical Humanities</i> , 2019 , 45, 2-9	1.4	1
4	Chromosome 6q24 transient neonatal diabetes mellitus and protein sensitive hyperinsulinaemic hypoglycaemia. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2014 , 27, 1065-9	1.6	1
3	Imprinting Disorders of Early Childhood 2012 , 137-160		1
2	Experiences of adolescents living with Silver-Russell syndrome. <i>Archives of Disease in Childhood</i> , 2021 , 106, 1195-1201	2.2	1
1	A patient with multilocus imprinting disturbance involving hypomethylation at 11p15 and 14q32, and phenotypic features of Beckwith-Wiedemann and Temple syndromes.. <i>American Journal of Medical Genetics, Part A</i> , 2022 ,	2.5	0