Deborah J G Mackay

List of Publications by Year in Descending Order

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	A patient with multilocus imprinting disturbance involving hypomethylation at 11p15 and 14q32, and phenotypic features of Beckwith-Wiedemann and Temple syndromes.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	0
79	Experiences of adolescents living with Silver-Russell syndrome. *Archives of Disease in Childhood*, **2021**, 106, 1195-1201	2.2	1
78	Whole-genome analysis as a diagnostic tool for patients referred for diagnosis of Silver-Russell syndrome: a real-world study. *Journal of Medical Genetics*, **2021**,	5.8	4
77	Phenotype of genetically confirmed Silver-Russell syndrome beyond childhood. *Journal of Medical Genetics*, **2020**, 57, 683-691	5.8	6
76	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. *Genome Research*, **2019**, 29, 1057-1066	9.7	20
75	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. *Cell*, **2019**, 177, 32-37	56.2	53
74	Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. *Genetical Research*, **2019**, 101, e3	1.1	9
73	Genetic diagnosis of subfertility: the impact of meiosis and maternal effects. *Journal of Medical Genetics*, **2019**, 56, 271-282	5.8	10
72	Lived experience of Silver-Russell syndrome: implications for management during childhood and into adulthood. *Archives of Disease in Childhood*, **2019**, 104, 76-82	2.2	7
71	Genetics, molar pregnancies and medieval ideas of monstrous births: the lump of flesh in. *Medical Humanities*, **2019**, 45, 2-9	1.4	1
70	The phenotypic variations of multi-locus imprinting disturbances associated with maternal-effect variants of NLRP5 range from overt imprinting disorder to apparently healthy phenotype. *Clinical Epigenetics*, **2019**, 11, 190	7.7	13
69	Genomic imprinting disorders: lessons on how genome, epigenome and environment interact. *Nature Reviews Genetics*, **2019**, 20, 235-248	30.1	151
68	Revisiting Wilms tumour surveillance in Beckwith-Wiedemann syndrome with IC2 methylation loss, reply. *European Journal of Human Genetics*, **2018**, 26, 471-472	5.3	7
67	Expert consensus document: Clinical and molecular diagnosis, screening and management of Beckwith-Wiedemann syndrome: an international consensus statement. *Nature Reviews Endocrinology*, **2018**, 14, 229-249	15.2	234
66	Maternal variants in and other maternal effect proteins are associated with multilocus imprinting disturbance in offspring. *Journal of Medical Genetics*, **2018**, 55, 497-504	5.8	66
65	Growth Hormone Improves Short-Term Growth in Patients with Temple Syndrome. *Hormone Research in Paediatrics*, **2018**, 90, 407-413	3.3	7
64	Human imprinting disorders: Principles, practice, problems and progress. *European Journal of Medical Genetics*, **2017**, 60, 618-626	2.6	35

63	Diagnosis and management of Silver-Russell syndrome: first international consensus statement. *Nature Reviews Endocrinology*, **2017**, 13, 105-124	15.2	224
62	Recent Advances in Imprinting Disorders. *Clinical Genetics*, **2017**, 91, 3-13	4	62
61	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. *Genetics in Medicine*, **2016**, 18, 309-15	8.1	52
60	Temple syndrome as a result of isolated hypomethylation of the 14q32 imprinted DLK1/MEG3 region. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 170-5	2.5	22
59	Prenatal molecular testing for Beckwith-Wiedemann and Silver-Russell syndromes: a challenge for molecular analysis and genetic counseling. *European Journal of Human Genetics*, **2016**, 24, 784-93	5.3	34
58	EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silver-Russell and Beckwith-Wiedemann syndrome. *European Journal of Human Genetics*, **2016**, 24, 1377-87	5.3	54
57	The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. *Lancet, The*, **2015**, 386, 957-63	40	186
56	Role of noninsulin therapies alone or in combination in chromosome 6q24-related transient neonatal diabetes: sulfonylurea improves but does not always normalize insulin secretion. *Diabetes Care*, **2015**, 38, e86-7	14.6	18
55	Beyond the gene roundtable discussion. *Textual Practice*, **2015**, 29, 415-432	0.1	1
54	Very small deletions within the NESP55 gene in pseudohypoparathyroidism type 1b. *European Journal of Human Genetics*, **2015**, 23, 494-9	5.3	18
53	Mutations in NLRP5 are associated with reproductive wastage and multilocus imprinting disorders in humans. *Nature Communications*, **2015**, 6, 8086	17.4	102
52	Multilocus methylation defects in imprinting disorders. *Biomolecular Concepts*, **2015**, 6, 47-57	3.7	31
51	Neonatal diabetes in Ukraine: incidence, genetics, clinical phenotype and treatment. *Journal of Pediatric Endocrinology and Metabolism*, **2015**, 28, 1279-86	1.6	25
50	Transient Neonatal Diabetes Mellitus followed by recurrent asymptomatic hypoglycaemia: a case report. *BMC Pediatrics*, **2015**, 15, 200	2.6	7
49	A statistical method for single sample analysis of HumanMethylation450 array data: genome-wide methylation analysis of patients with imprinting disorders. *Clinical Epigenetics*, **2015**, 7, 48	7.7	12
48	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. *Clinical Epigenetics*, **2015**, 7, 123	7.7	115
47	Report and review of described associations of Mayer-Rokitansky-K□ter-Hauser syndrome and Silver-Russell syndrome. *Journal of Paediatrics and Child Health*, **2015**, 51, 555-560	1.3	6
46	Genome-wide DNA methylation analysis of patients with imprinting disorders identifies differentially methylated regions associated with novel candidate imprinted genes. *Journal of Medical Genetics*, **2014**, 51, 229-38	5.8	70

45	A familial disorder of altered DNA-methylation. *Journal of Medical Genetics*, **2014**, 51, 407-12	5.8	22
44	Chromosome 6q24 transient neonatal diabetes mellitus and protein sensitive hyperinsulinaemic hypoglycaemia. *Journal of Pediatric Endocrinology and Metabolism*, **2014**, 27, 1065-9	1.6	1
43	Temple syndrome: improving the recognition of an underdiagnosed chromosome 14 imprinting disorder: an analysis of 51 published cases. *Journal of Medical Genetics*, **2014**, 51, 495-501	5.8	136
42	Clinical presentation of 6q24 transient neonatal diabetes mellitus (6q24 TNDM) and genotype-phenotype correlation in an international cohort of patients. *Diabetologia*, **2013**, 56, 758-62	10.3	90
41	Targeted methylation testing of a patient cohort broadens the epigenetic and clinical description of imprinting disorders. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2174-82	2.5	65
40	Transient neonatal diabetes, ZFP57, and hypomethylation of multiple imprinted loci: a detailed follow-up. *Diabetes Care*, **2013**, 36, 505-12	14.6	48
39	Insights into the molecular mechanism for type 2 diabetes susceptibility at the KCNQ1 locus from temporal changes in imprinting status in human islets. *Diabetes*, **2013**, 62, 987-92	0.9	87
38	Hepatoblastoma in a child with a paternally-inherited ABCC8 mutation and mosaic paternal uniparental disomy 11p causing focal congenital hyperinsulinism. *European Journal of Medical Genetics*, **2013**, 56, 114-7	2.6	16
37	Transient neonatal diabetes mellitus in a Turkish patient with three novel homozygous variants in the ZFP57 gene. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2013**, 5, 125-8	1.9	5
36	Mutation of HERC2 causes developmental delay with Angelman-like features. *Journal of Medical Genetics*, **2013**, 50, 65-73	5.8	50
35	Evidence for anticipation in Beckwith-Wiedemann syndrome. *European Journal of Human Genetics*, **2013**, 21, 1344-8	5.3	23
34	Familial occurrence of neonatal diabetes with duplications in chromosome 6q24: treatment with sulfonylurea and 40-yr follow-up. *Pediatric Diabetes*, **2012**, 13, 155-62	3.6	15
33	Imprinting Disorders of Early Childhood **2012**, 137-160		1
32	Beckwith-Wiedemann syndrome caused by maternally inherited mutation of an OCT-binding motif in the IGF2/H19-imprinting control region, ICR1. *European Journal of Human Genetics*, **2012**, 20, 240-3	5.3	51
31	Deletions and rearrangements of the H19/IGF2 enhancer region in patients with Silver-Russell syndrome and growth retardation. *Journal of Medical Genetics*, **2011**, 48, 308-11	5.8	33
30	An atypical case of hypomethylation at multiple imprinted loci. *European Journal of Human Genetics* , **2011**, 19, 360-2	5.3	33
29	Methylation analysis of 79 patients with growth restriction reveals novel patterns of methylation change at imprinted loci. *European Journal of Human Genetics*, **2010**, 18, 648-55	5.3	80
28	Incidence of neonatal diabetes in Austria-calculation based on the Austrian Diabetes Register. *Pediatric Diabetes*, **2010**, 11, 18-23	3.6	42

27	Epigenotype-phenotype correlations in Silver-Russell syndrome. *Journal of Medical Genetics*, **2010**, 47, 760-8	5.8	98
26	Further refinement of the critical minimal genetic region for the imprinting disorder 6q24 transient neonatal diabetes. *Diabetologia*, **2010**, 53, 2347-51	10.3	27
25	DNA hypomethylation, transient neonatal diabetes, and prune belly sequence in one of two identical twins. *European Journal of Pediatrics*, **2010**, 169, 207-13	4.1	19
24	Investigation of 90 patients referred for molecular cytogenetic analysis using aCGH uncovers previously unsuspected anomalies of imprinting. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1990-3	2.5	6
23	Transient neonatal diabetes mellitus type 1. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2010**, 154C, 335-42	3.1	81
22	Concurrent course of transient neonatal diabetes with cholestasis and paucity of interlobular bile ducts: a case report. *Pediatric and Developmental Pathology*, **2009**, 12, 417-20	2.2	6
21	Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwith-Wiedemann syndrome. *European Journal of Human Genetics*, **2009**, 17, 611-9	5.3	176
20	Hypomethylation of multiple imprinted loci in individuals with transient neonatal diabetes is associated with mutations in ZFP57. *Nature Genetics*, **2008**, 40, 949-51	36.3	417
19	Clinical characterisation of the multiple maternal hypomethylation syndrome in siblings. *European Journal of Human Genetics*, **2008**, 16, 453-61	5.3	46
18	Mosaic paternal uniparental isodisomy and an ABCC8 gene mutation in a patient with permanent neonatal diabetes and hemihypertrophy. *Diabetes*, **2008**, 57, 255-8	0.9	12
17	Zellweger syndrome resulting from maternal isodisomy of chromosome 1. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2172-7	2.5	20
16	Mutations in ATP-sensitive K+ channel genes cause transient neonatal diabetes and permanent diabetes in childhood or adulthood. *Diabetes*, **2007**, 56, 1930-7	0.9	273
15	Isolated imprinting mutation of the DLK1/GTL2 locus associated with a clinical presentation of maternal uniparental disomy of chromosome 14. *Journal of Medical Genetics*, **2007**, 44, 637-40	5.8	54
14	Permanent neonatal diabetes caused by dominant, recessive, or compound heterozygous SUR1 mutations with opposite functional effects. *American Journal of Human Genetics*, **2007**, 81, 375-82	11	161
13	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. *Diabetes*, **2006**, 55, 2272-6	0.9	10
12	Epimutation of the TNDM locus and the Beckwith-Wiedemann syndrome centromeric locus in individuals with transient neonatal diabetes mellitus. *Human Genetics*, **2006**, 119, 179-84	6.3	51
11	A maternal hypomethylation syndrome presenting as transient neonatal diabetes mellitus. *Human Genetics*, **2006**, 120, 262-9	6.3	139
10	Bisulphite sequencing of the transient neonatal diabetes mellitus DMR facilitates a novel diagnostic test but reveals no methylation anomalies in patients of unknown aetiology. *Human Genetics*, **2005**, 116, 255-61	6.3	51

9	Monozygous triplets discordant for transient neonatal diabetes mellitus and for imprinting of the TNDM differentially methylated region. *Human Genetics*, **2005**, 117, 398-401	6.3	12
8	Relapsing diabetes can result from moderately activating mutations in KCNJ11. *Human Molecular Genetics*, **2005**, 14, 925-34	5.6	165
7	Activating mutations in the gene encoding the ATP-sensitive potassium-channel subunit Kir6.2 and permanent neonatal diabetes. *New England Journal of Medicine*, **2004**, 350, 1838-49	59.2	930
6	Relaxation of imprinted expression of ZAC and HYMAI in a patient with transient neonatal diabetes mellitus. *Human Genetics*, **2002**, 110, 139-44	6.3	70
5	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. *Diabetologia*, **2001**, 44, 924	10.3	5
4	Characterization of the methylation-sensitive promoter of the imprinted ZAC gene supports its role in transient neonatal diabetes mellitus. *Journal of Biological Chemistry*, **2001**, 276, 18653-6	5.4	52
3	Transient neonatal diabetes: widening the understanding of the etiopathogenesis of diabetes. *Diabetes*, **2000**, 49, 1359-66	0.9	214
2	The Rho's progress: a potential role during neuritogenesis for the Rho family of GTPases. *Trends in Neurosciences*, **1995**, 18, 496-501	13.3	110
1	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging		2