# Deborah J G Mackay

List of Publications by Year
in descending order

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| 81 | 7,214 | 76322 40 | 62593 80 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 84 | 84 | 84 | 6390 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | 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, 1838-1849. | 27.0 | 1,077 |
| 2 | Hypomethylation of multiple imprinted loci in individuals with transient neonatal diabetes is associated with mutations in ZFP57. Nature Genetics, 2008, 40, 949-951. | 21.4 | 460 |
| 3 | Clinical and molecular diagnosis, screening and management of Beckwithâ€“Wiedemann syndrome: an international consensus statement. Nature Reviews Endocrinology, 2018, 14, 229-249. | 9.6 | 388 |
| 4 | Diagnosis and management of Silverâ€“Russell syndrome: first international consensus statement. Nature Reviews Endocrinology, 2017, 13, 105-124. | 9.6 | 336 |
| 5 | Mutations in ATP-Sensitive K+ Channel Genes Cause Transient Neonatal Diabetes and Permanent Diabetes in Childhood or Adulthood. Diabetes, 2007, 56, 1930-1937. | 0.6 | 320 |
| 6 | Genomic imprinting disorders: lessons on how genome, epigenome and environment interact. Nature Reviews Genetics, 2019, 20, 235-248. | 16.3 | 291 |
| 7 | The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. Lancet, The, 2015, 386, 957-963. | 13.7 | 250 |
| 8 | Transient neonatal diabetes: widening the understanding of the etiopathogenesis of diabetes.. Diabetes, 2000, 49, 1359-1366. | 0.6 | 249 |
| 9 | Permanent Neonatal Diabetes Caused by Dominant, Recessive, or Compound Heterozygous SUR1 Mutations with Opposite Functional Effects. American Journal of Human Genetics, 2007, 81, 375-382. | 6.2 | 194 |
| 10 | Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2009, 17, 611-619. | 2.8 | 194 |
| 11 | Relapsing diabetes can result from moderately activating mutations in KCNJ11. Human Molecular Genetics, 2005, 14, 925-934. | 2.9 | 184 |
| 12 | 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. | 3.2 | 182 |
| 13 | Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. Clinical Epigenetics, 2015, 7, 123. | 4.1 | 174 |
| 14 | A maternal hypomethylation syndrome presenting as transient neonatal diabetes mellitus. Human Genetics, 2006, 120, 262-269. | 3.8 | 147 |
| 15 | Mutations in NLRP5 are associated with reproductive wastage and multilocus imprinting disorders in humans. Nature Communications, 2015, 6, 8086. | 12.8 | 134 |
| 16 | Maternal variants in NLRP and other maternal effect proteins are associated with multilocus imprinting disturbance in offspring. Journal of Medical Genetics, 2018, 55, 497-504. | 3.2 | 126 |
| 17 | Epigenotype-phenotype correlations in Silver-Russell syndrome. Journal of Medical Genetics, 2010, 47, 760-768. | 3.2 | 123 |
| 18 | The Rho's progress: a potential role during neuritogenesis for the Rho family of GTPases. Trends in Neurosciences, 1995, 18, 496-501. | 8.6 | 119 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Clinical presentation of 6q24 transient neonatal diabetes mellitus (6q24 TNDM) and genotypeâ€“phenotype correlation in an international cohort of patients. Diabetologia, 2013, 56, 758-762. | 6.3 | 113 |
| 20 | A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37. | 28.9 | 113 |
| 21 | 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, 987-992. | 0.6 | 112 |
| 22 | Recent Advances in Imprinting Disorders. Clinical Genetics, 2017, 91, 3-13. | 2.0 | 101 |
| 23 | Transient neonatal diabetes mellitus type 1. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 335-342. | 1.6 | 99 |
| 24 | Methylation analysis of 79 patients with growth restriction reveals novel patterns of methylation change at imprinted loci. European Journal of Human Genetics, 2010, 18, 648-655. | 2.8 | 91 |
| 25 | 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, 229-238. | 3.2 | 91 |
| 26 | Relaxation of imprinted expression of ZAC and HYMAI in a patient with transient neonatal diabetes mellitus. Human Genetics, 2002, 110, 139-144. | 3.8 | 83 |
| 27 | Targeted methylation testing of a patient cohort broadens the epigenetic and clinical description of imprinting disorders. American Journal of Medical Genetics, Part A, 2013, 161, 2174-2182. | 1.2 | 69 |
| 28 | Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315. | 2.4 | 69 |
| 29 | EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silverâ€“Russell and Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 1377-1387. | 2.8 | 68 |
| 30 | 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, 18653-18656. | 3.4 | 65 |
| 31 | Beckwithâ€“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, 240-243. | 2.8 | 60 |
| 32 | 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, 637-640. | 3.2 | 59 |
| 33 | Mutation of HERC2 causes developmental delay with Angelman-like features. Journal of Medical Genetics, 2013, 50, 65-73. | 3.2 | 57 |
| 34 | Epimutation of the TNDM locus and the Beckwithâ€“Wiedemann syndrome centromeric locus in individuals with transient neonatal diabetes mellitus. Human Genetics, 2006, 119, 179-184. | 3.8 | 56 |
| 35 | Transient Neonatal Diabetes, ZFP57, and Hypomethylation of Multiple Imprinted Loci. Diabetes Care, 2013, 36, 505-512. | 8.6 | 56 |
| 36 | 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, 255-261. | 3.8 | 54 |

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|---|---|---|---|
| 37 | Incidence of neonatal diabetes in Austria-calculation based on the Austrian Diabetes Register. Pediatric Diabetes, 2010, 11, 18-23. | 2.9 | 52 |
| 38 | Clinical characterisation of the multiple maternal hypomethylation syndrome in siblings. European Journal of Human Genetics, 2008, 16, 453-461. | 2.8 | 51 |
| 39 | Human imprinting disorders: Principles, practice, problems and progress. European Journal of Medical Genetics, 2017, 60, 618-626. | 1.3 | 49 |
| 40 | Prenatal molecular testing for Beckwithâ€“Wiedemann and Silverâ€“Russell syndromes: a challenge for molecular analysis and genetic counseling. European Journal of Human Genetics, 2016, 24, 784-793. | 2.8 | 44 |
| 41 | Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. Genome Research, 2019, 29, 1057-1066. | 5.5 | 38 |
| 42 | Deletions and rearrangements of the H19/IGF2 enhancer region in patients with Silver-Russell syndrome and growth retardation. Journal of Medical Genetics, 2011, 48, 308-311. | 3.2 | 35 |
| 43 | An atypical case of hypomethylation at multiple imprinted loci. European Journal of Human Genetics, 2011, 19, 360-362. | 2.8 | 35 |
| 44 | Multilocus methylation defects in imprinting disorders. Biomolecular Concepts, 2015, 6, 47-57. | 2.2 | 34 |
| 45 | Neonatal diabetes in Ukraine: incidence, genetics, clinical phenotype and treatment. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 1279-86. | 0.9 | 34 |
| 46 | Evidence for anticipation in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2013, 21, 1344-1348. | 2.8 | 30 |
| 47 | Further refinement of the critical minimal genetic region for the imprinting disorder 6q24 transient neonatal diabetes. Diabetologia, 2010, 53, 2347-2351. | 6.3 | 29 |
| 48 | 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-e87. | 8.6 | 25 |
| 49 | Temple syndrome as a result of isolated hypomethylation of the 14q32 imprinted DLK1/MEG3 region. American Journal of Medical Genetics, Part A, 2016, 170, 170-175. | 1.2 | 25 |
| 50 | A familial disorder of altered DNA-methylation. Journal of Medical Genetics, 2014, 51, 407-412. | 3.2 | 24 |
| 51 | Zellweger syndrome resulting from maternal isodisomy of chromosome 1. American Journal of Medical Genetics, Part A, 2007, 143A, 2172-2177. | 1.2 | 23 |
| 52 | 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. | 4.1 | 22 |
| 53 | DNA hypomethylation, transient neonatal diabetes, and prune belly sequence in one of two identical twins. European Journal of Pediatrics, 2010, 169, 207-213. | 2.7 | 21 |
| 54 | Very small deletions within the NESP55 gene in pseudohypoparathyroidism type 1b. European Journal of Human Genetics, 2015, 23, 494-499. | 2.8 | 20 |

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|---|---|---|---|
| 55 | Familial occurrence of neonatal diabetes with duplications in chromosome 6q24: treatment with sulfonylurea and 40-yr follow-up. Pediatric Diabetes, 2012, 13, 155-162. | 2.9 | 19 |
| 56 | 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, 114-117. | 1.3 | 19 |
| 57 | 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. | 4.1 | 18 |
| 58 | Phenotype of genetically confirmed Silver-Russell syndrome beyond childhood. Journal of Medical Genetics, 2020, 57, 683-691. | 3.2 | 18 |
| 59 | Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. Genetical Research, 2019, 101, e3. | 0.9 | 17 |
| 60 | Mosaic Paternal Uniparental Isodisomy and an ABCC8 Gene Mutation in a Patient With Permanent Neonatal Diabetes and Hemihypertrophy. Diabetes, 2008, 57, 255-258. | 0.6 | 15 |
| 61 | 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, 2272-2276. | 0.6 | 13 |
| 62 | Revisiting Wilms tumour surveillance in Beckwithâ€“Wiedemann syndrome with IC2 methylation loss, reply. European Journal of Human Genetics, 2018, 26, 471-472. | 2.8 | 13 |
| 63 | Lived experience of Silver-Russell syndrome: implications for management during childhood and into adulthood. Archives of Disease in Childhood, 2019, 104, 76-82. | 1.9 | 13 |
| 64 | Monozygous triplets discordant for transient neonatal diabetes mellitus and for imprinting of the TNDM differentially methylated region. Human Genetics, 2005, 117, 398-401. | 3.8 | 12 |
| 65 | Genetic diagnosis of subfertility: the impact of meiosis and maternal effects. Journal of Medical Genetics, 2019, 56, 271-282. | 3.2 | 11 |
| 66 | Report and review of described associations of <scp>M</scp>ayerâ€<scp>R</scp>okitanskyâ€<scp>K</scp>Ã¼sterâ€<scp>H</scp>auser syndrome and <scp>S</scp>ilverâ€“<scp>R</scp>ussell syndrome. Journal of Paediatrics and Child Health, 2015, 51, 555-560. | 0.8 | 10 |
| 67 | Growth Hormone Improves Short-Term Growth in Patients with Temple Syndrome. Hormone Research in Paediatrics, 2018, 90, 407-413. | 1.8 | 10 |
| 68 | Observationsï¿½Maturity 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, 924-924. | 6.3 | 8 |
| 69 | Whole-genome analysis as a diagnostic tool for patients referred for diagnosis of Silver-Russell syndrome: a real-world study. Journal of Medical Genetics, 2022, 59, 613-622. | 3.2 | 8 |
| 70 | Concurrent Course of Transient Neonatal Diabetes with Cholestasis and Paucity of Interlobular Bile Ducts: A Case Report. Pediatric and Developmental Pathology, 2009, 12, 417-420. | 1.0 | 7 |
| 71 | Transient Neonatal Diabetes Mellitus followed by recurrent asymptomatic hypoglycaemia: a case report. BMC Pediatrics, 2015, 15, 200. | 1.7 | 7 |
| 72 | 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, 1990-1993. | 1.2 | 6 |

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|---|---|---|---|
| 73 | 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, 125-128. | 0.9 | 6 |
| 74 | Experiences of adolescents living with Silver-Russell syndrome. Archives of Disease in Childhood, 2021, 106, 1195-1201. | 1.9 | 6 |
| 75 | A patient with multilocus imprinting disturbance involving hypomethylation at 11p15 and 14q32, and phenotypic features of <scp>BeckwithâЄWiedemann</scp> and Temple syndromes. American Journal of Medical Genetics, Part A, 2022, , . | 1.2 | 5 |
| 76 | Height and body mass index in molecularly confirmed Silverâ€“Russell syndrome and the longâ€term effects of growth hormone treatment. Clinical Endocrinology, 2022, 97, 284-292. | 2.4 | 3 |
| 77 | Ongoing Challenges in the Diagnosis of 11p15.5-Associated Imprinting Disorders. Molecular Diagnosis and Therapy, 2022, 26, 263-272. | 3.8 | 3 |
| 78 | Chromosome 6q24 transient neonatal diabetes mellitus and protein sensitive hyperinsulinaemic hypoglycaemia. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 1065-9. | 0.9 | 2 |
| 79 | Imprinting Disorders of Early Childhood. , 2012, , 137-160. | | 1 |
| 80 | Beyond the gene roundtable discussion. Textual Practice, 2015, 29, 415-432. | 0.6 | 1 |
| 81 | Genetics, molar pregnancies and medieval ideas of monstrous births: the lump of flesh in The King of Tars. Medical Humanities, 2019, 45, 2-9. | 1.2 | 1 |