

# David Monk

## List of Publications by Year in descending order

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83  
papers

5,139  
citations

81900

39  
h-index

95266

68  
g-index

85  
all docs

85  
docs citations

85  
times ranked

7169  
citing authors

#	ARTICLE	IF	CITATIONS
1	Preimplantation genetic testing for a chr14q32 microdeletion in a family with Kagami-Ogata syndrome and Temple syndrome. <i>Journal of Medical Genetics</i> , 2022, 59, 253-261.	3.2	5
2	Environmentally sensitive hotspots in the methylome of the early human embryo. <i>ELife</i> , 2022, 11, .	6.0	15
3	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. <i>Clinical Epigenetics</i> , 2022, 14, 41.	4.1	14
4	Novel genetic variants of KHDC3L and other members of the subcortical maternal complex associated with Beckwith-Wiedemann syndrome or Pseudohypoparathyroidism 1B and multi-locus imprinting disturbances. <i>Clinical Epigenetics</i> , 2022, 14, .	4.1	7
5	The role of ZFP57 and additional KRAB-zinc finger proteins in the maintenance of human imprinted methylation and multi-locus imprinting disturbances. <i>Nucleic Acids Research</i> , 2020, 48, 11394-11407.	14.5	32
6	Recommendations for Diagnosis and Treatment of Pseudohypoparathyroidism and Related Disorders: An Updated Practical Tool for Physicians and Patients. <i>Hormone Research in Paediatrics</i> , 2020, 93, 182-196.	1.8	42
7	The hypomethylation of imprinted genes in IVF/ICSI placenta samples is associated with concomitant changes in histone modifications. <i>Epigenetics</i> , 2020, 15, 1386-1395.	2.7	8
8	Comprehensive analysis of PM20D1 QTL in Alzheimer's disease. <i>Clinical Epigenetics</i> , 2020, 12, 20.	4.1	16
9	A case of intraplacental gestational choriocarcinoma; characterised by the methylation pattern of the early placenta and an absence of driver mutations. <i>BMC Cancer</i> , 2019, 19, 744.	2.6	17
10	Wnt/ $\beta$ -catenin signaling pathway safeguards epigenetic stability and homeostasis of mouse embryonic stem cells. <i>Scientific Reports</i> , 2019, 9, 948.	3.3	31
11	Differences in expression rather than methylation at placenta-specific imprinted loci is associated with intrauterine growth restriction. <i>Clinical Epigenetics</i> , 2019, 11, 35.	4.1	29
12	A KHDC3L mutation resulting in recurrent hydatidiform mole causes genome-wide DNA methylation loss in oocytes and persistent imprinting defects post-fertilisation. <i>Genome Medicine</i> , 2019, 11, 84.	8.2	45
13	Genomic imprinting disorders: lessons on how genome, epigenome and environment interact. <i>Nature Reviews Genetics</i> , 2019, 20, 235-248.	16.3	291
14	The Use of Methylation-Sensitive Multiplex Ligation-Dependent Probe Amplification for Quantification of Imprinted Methylation. <i>Methods in Molecular Biology</i> , 2018, 1766, 109-121.	0.9	2
15	Recommendations for a nomenclature system for reporting methylation aberrations in imprinted domains. <i>Epigenetics</i> , 2018, 13, 117-121.	2.7	70
16	Profiling of oxBS-450K 5-hydroxymethylcytosine in human placenta and brain reveals enrichment at imprinted loci. <i>Epigenetics</i> , 2018, 13, 182-191.	2.7	23
17	Characterization of parent-of-origin methylation using the Illumina Infinium MethylationEPIC array platform. <i>Epigenomics</i> , 2018, 10, 941-954.	2.1	31
18	Diagnosis and management of pseudohypoparathyroidism and related disorders: first international Consensus Statement. <i>Nature Reviews Endocrinology</i> , 2018, 14, 476-500.	9.6	224

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19	PM20D1 is a quantitative trait locus associated with Alzheimer's disease. <i>Nature Medicine</i> , 2018, 24, 598-603.	30.7	73
20	Maternal mutations of <i>FOXF1</i> cause alveolar capillary dysplasia despite not being imprinted. <i>Human Mutation</i> , 2017, 38, 615-620.	2.5	13
21	Human Amniocytes Are Receptive to Chemically Induced Reprogramming to Pluripotency. <i>Molecular Therapy</i> , 2017, 25, 427-442.	8.2	10
22	NLRPs, the subcortical maternal complex and genomic imprinting. <i>Reproduction</i> , 2017, 154, R161-R170.	2.6	58
23	Copy number rather than epigenetic alterations are the major dictator of imprinted methylation in tumors. <i>Nature Communications</i> , 2017, 8, 467.	12.8	27
24	Diagnosis and management of Silver-Russell syndrome: first international consensus statement. <i>Nature Reviews Endocrinology</i> , 2017, 13, 105-124.	9.6	336
25	Epigenetic Characterization of CDKN1C in Placenta Samples from Non-syndromic Intrauterine Growth Restriction. <i>Frontiers in Genetics</i> , 2016, 7, 62.	2.3	21
26	Human Oocyte-Derived Methylation Differences Persist in the Placenta Revealing Widespread Transient Imprinting. <i>PLoS Genetics</i> , 2016, 12, e1006427.	3.5	94
27	Causes and Consequences of Multi-Locus Imprinting Disturbances in Humans. <i>Trends in Genetics</i> , 2016, 32, 444-455.	6.7	81
28	Genome-wide DNA methylation analysis of pseudohypoparathyroidism patients with GNAS imprinting defects. <i>Clinical Epigenetics</i> , 2016, 8, 10.	4.1	53
29	Nongenomic regulation of gene expression. <i>Current Opinion in Pediatrics</i> , 2016, 28, 521-528.	2.0	2
30	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.	2.8	68
31	APOA5 genetic and epigenetic variability jointly regulate circulating triacylglycerol levels. <i>Clinical Science</i> , 2016, 130, 2053-2059.	4.3	15
32	Clinical and molecular analyses of Beckwith-Wiedemann syndrome: Comparison between spontaneous conception and assisted reproduction techniques. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2740-2749.	1.2	30
33	Uniparental disomy of chromosome 16 unmasks recessive mutations of <i>FA2H</i> /SPG35 in 4 families. <i>Neurology</i> , 2016, 87, 186-191.	1.1	27
34	Phenotypic spectrum and extent of DNA methylation defects associated with multilocus imprinting disturbances. <i>Epigenomics</i> , 2016, 8, 801-816.	2.1	26
35	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.	2.8	44
36	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. <i>Clinical Epigenetics</i> , 2015, 7, 123.	4.1	174

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37	Polymorphisms Near TBX5 and GDF7 Are Associated With Increased Risk for Barrett's Esophagus. <i>Gastroenterology</i> , 2015, 148, 367-378.	1.3	93
38	Distinct promoter methylation and isoform-specific expression of RASFF1A in placental biopsies from complicated pregnancies. <i>Placenta</i> , 2015, 36, 397-402.	1.5	9
39	Germline-derived DNA methylation and early embryo epigenetic reprogramming: The selected survival of imprints. <i>International Journal of Biochemistry and Cell Biology</i> , 2015, 67, 128-138.	2.8	80
40	Congenital imprinting disorders: EUCID.net - a network to decipher their aetiology and to improve the diagnostic and clinical care. <i>Clinical Epigenetics</i> , 2015, 7, 23.	4.1	23
41	Combined epigenetic and intraspecific variation of the <i>DRD4</i> and <i>SERT</i> genes influence novelty seeking behavior in great tit <i>Parus major</i> . <i>Epigenetics</i> , 2015, 10, 516-525.	2.7	65
42	The role and interaction of imprinted genes in human fetal growth. <i>Philosophical Transactions of the Royal Society B: Biological Sciences</i> , 2015, 370, 20140074.	4.0	113
43	Genomic imprinting in the human placenta. <i>American Journal of Obstetrics and Gynecology</i> , 2015, 213, S152-S162.	1.3	107
44	Congenital imprinting disorders: Application of multilocus and high throughput methods to decipher new pathomechanisms and improve their management. <i>Molecular and Cellular Probes</i> , 2015, 29, 282-290.	2.1	12
45	Absence of Maternal Methylation in Biparental Hydatidiform Moles from Women with NLRP7 Maternal-Effect Mutations Reveals Widespread Placenta-Specific Imprinting. <i>PLoS Genetics</i> , 2015, 11, e1005644.	3.5	80
46	Altered expression of the imprinted transcription factor PLAGL1 deregulates a network of genes in the human IUGR placenta. <i>Human Molecular Genetics</i> , 2014, 23, 6275-6285.	2.9	66
47	Variable maternal methylation overlapping the <i>nc886/vtRNA2-1</i> locus is locked between hypermethylated repeats and is frequently altered in cancer. <i>Epigenetics</i> , 2014, 9, 783-790.	2.7	54
48	Hypermethylation of the alternative AWT1 promoter in hematological malignancies is a highly specific marker for acute myeloid leukemias despite high expression levels. <i>Journal of Hematology and Oncology</i> , 2014, 7, 4.	17.0	21
49	Genome-wide parent-of-origin DNA methylation analysis reveals the intricacies of human imprinting and suggests a germline methylation-independent mechanism of establishment. <i>Genome Research</i> , 2014, 24, 554-569.	5.5	311
50	Screening individuals with intellectual disability, autism and Tourette's syndrome for <i>KCNK9</i> mutations and aberrant DNA methylation within the 8q24 imprinted cluster. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2014, 165, 472-478.	1.7	13
51	A New Overgrowth Syndrome is due to Mutations in <i>RNF125</i> . <i>Human Mutation</i> , 2014, 35, 1436-1441.	2.5	33
52	Tissue-specific DNA methylation profiles regulate liver-specific expression of the APOA1/C3/A4/A5 cluster and can be manipulated with demethylating agents on intestinal cells. <i>Atherosclerosis</i> , 2014, 237, 528-535.	0.8	15
53	The PEG13-DMR and brain-specific enhancers dictate imprinted expression within the 8q24 intellectual disability risk locus. <i>Epigenetics and Chromatin</i> , 2014, 7, 5.	3.9	46
54	Genome-Wide Allelic Methylation Analysis Reveals Disease-Specific Susceptibility to Multiple Methylation Defects in Imprinting Syndromes. <i>Human Mutation</i> , 2013, 34, n/a-n/a.	2.5	96

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55	Stability of Genomic Imprinting and Gestational-Age Dynamic Methylation in Complicated Pregnancies Conceived Following Assisted Reproductive Technologies1. <i>Biology of Reproduction</i> , 2013, 89, 50.	2.7	49
56	Imprinting at the PLAGL1 domain is contained within a 70-kb CTCF/cohesin-mediated non-allelic chromatin loop. <i>Nucleic Acids Research</i> , 2013, 41, 2171-2179.	14.5	25
57	A DNA methylation fingerprint of 1628 human samples. <i>Genome Research</i> , 2012, 22, 407-419.	5.5	341
58	Characterization of Novel Paternal ncRNAs at the Plagl1 Locus, Including Hymai, Predicted to Interact with Regulators of Active Chromatin. <i>PLoS ONE</i> , 2012, 7, e38907.	2.5	21
59	Maternal Inheritance of a Promoter Variant in the Imprinted PHLDA2 Gene Significantly Increases Birth Weight. <i>American Journal of Human Genetics</i> , 2012, 90, 715-719.	6.2	40
60	Abstract 4021: Breaking the dogma: AWT1 hypermethylation in myeloid leukemia despite high expression. , 2012, , .		0
61	Methylation screening of reciprocal genome-wide UPDs identifies novel human-specific imprinted genes. <i>Human Molecular Genetics</i> , 2011, 20, 3188-3197.	2.9	55
62	The consequences of uniparental disomy and copy number neutral loss-of-heterozygosity during human development and cancer. <i>Biology of the Cell</i> , 2011, 103, 303-317.	2.0	68
63	Does Genomic Imprinting Play a Role in Autoimmunity?. <i>Advances in Experimental Medicine and Biology</i> , 2011, 711, 103-116.	1.6	9
64	Genotype of an individual single nucleotide polymorphism regulates DNA methylation at the <i>TRPC3</i> alternative promoter. <i>Epigenetics</i> , 2011, 6, 1236-1241.	2.7	18
65	Constitutional mosaic genome-wide uniparental disomy due to diploidisation: an unusual cancer-predisposing mechanism. <i>Journal of Medical Genetics</i> , 2011, 48, 212-216.	3.2	32
66	Human imprinted retrogenes exhibit non-canonical imprint chromatin signatures and reside in non-imprinted host genes. <i>Nucleic Acids Research</i> , 2011, 39, 4577-4586.	14.5	22
67	Deciphering the cancer imprintome. <i>Briefings in Functional Genomics</i> , 2010, 9, 329-339.	2.7	38
68	Transcript- and tissue-specific imprinting of a tumour suppressor gene. <i>Human Molecular Genetics</i> , 2009, 18, 118-127.	2.9	54
69	Reciprocal imprinting of human GRB10 in placental trophoblast and brain: evolutionary conservation of reversed allelic expression. <i>Human Molecular Genetics</i> , 2009, 18, 3066-3074.	2.9	92
70	Dynamic variation in allele-specific gene expression of Paraoxonase-1 in murine and human tissues. <i>Human Molecular Genetics</i> , 2008, 17, 3263-3270.	2.9	15
71	Comparative analysis of human chromosome 7q21 and mouse proximal chromosome 6 reveals a placental-specific imprinted gene, <i>Tfpi2</i> , which requires EHMT2 and EED for allelic-silencing. <i>Genome Research</i> , 2008, 18, 1270-1281.	5.5	72
72	Genomic Imprinting of <i>Dopa decarboxylase</i> in Heart and Reciprocal Allelic Expression with Neighboring <i>Grb10</i> . <i>Molecular and Cellular Biology</i> , 2008, 28, 386-396.	2.3	40

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73	A Screen for Retrotransposed Imprinted Genes Reveals an Association between X Chromosome Homology and Maternal Germ-Line Methylation. <i>PLoS Genetics</i> , 2007, 3, e20.	3.5	103
74	STOX1 is not imprinted and is not likely to be involved in preeclampsia. <i>Nature Genetics</i> , 2007, 39, 279-280.	21.4	45
75	Epigenetic Allele Silencing Unveils Recessive RYR1 Mutations in Core Myopathies. <i>American Journal of Human Genetics</i> , 2006, 79, 859-868.	6.2	111
76	Intrauterine growth restriction genetic causes and consequences. <i>Seminars in Fetal and Neonatal Medicine</i> , 2004, 9, 371-378.	2.3	77
77	Imprinted methylation profiles for proximal mouse Chromosomes 11 and 7 as revealed by methylation-sensitive representational difference analysis. <i>Mammalian Genome</i> , 2003, 14, 805-816.	2.2	14
78	Conserved methylation imprints in the human and mouse GRB10 genes with divergent allelic expression suggests differential reading of the same mark. <i>Human Molecular Genetics</i> , 2003, 12, 1005-1019.	2.9	141
79	DDC and COBL, flanking the imprinted GRB10 gene on 7p12, are biallelically expressed. <i>Mammalian Genome</i> , 2002, 13, 686-691.	2.2	23
80	Chromosome 7p disruptions in Silver Russell syndrome: delineating an imprinted candidate gene region.. <i>Human Genetics</i> , 2002, 111, 376-387.	3.8	79
81	Conflicting Reports of Imprinting Status of Human GRB10 in Developing Brain: How Reliable Are Somatic Cell Hybrids for Predicting Allelic Origin of Expression?. <i>American Journal of Human Genetics</i> , 2001, 68, 543-544.	6.2	38
82	Maternal repression of the human GRB10 gene in the developing central nervous system; evaluation of the role for GRB10 in Silver-Russell syndrome. <i>European Journal of Human Genetics</i> , 2001, 9, 82-90.	2.8	76
83	Duplication of 7p11.2-p13, Including GRB10, in Silver-Russell Syndrome. <i>American Journal of Human Genetics</i> , 2000, 66, 36-46.	6.2	148