

Imprinting publication list

Communicating science: epigenetics in the spotlight

Environ Epigenetics 2020 Nov 18;6(1):dvaa015 >>[PubMed-Link](#)<<

Dyke SOM, Ennis CA, Joly Y, Walter J, Siebert R, Pastinen T

Common genetic variation in the Angelman syndrome imprinting centre affects the imprinting of chromosome 15

Eur J Hum Genet 2020 Jun;28(6):835-839 >>[PubMed-Link](#)<<

Beygo J, Grosser C, Kaya S, Mertel C, Buiting K, Horsthemke B

Update of the EMQN/ACGS best practice guidelines for molecular analysis of Prader-Willi and Angelman syndromes

Eur J Hum Genet 2019 Sep; 27(9): 1326-40

Beygo J, Buiting K, Ramsden SC, Ellis R, Clayton-Smith J, Kanber D

Disruption of KCNQ1 prevents methylation of the ICR2 and supports the hypothesis that its transcription is necessary for imprint establishment

Eur J Hum Genet 2019 Jun; 27(6): 903-8

Beygo J, Burger J, Strom TM, Kaya S, Buiting K

Sensory neuropathy-causing mutations in ATL3 affect ER-mitochondria contact sites and impair axonal mitochondrial distribution

Hum Mol Genet 2019 Feb; 28(4): 615-27

Krols M, Asselbergh B, De Rycke R, De Winter V, Seyer A, Muller FJ, et al.

Points-to-consider on the return of results in epigenetic research

Genome Med 2019 May; 11: 9

Dyke SOM, Saulnier KM, Dupras C, Webster AP, Maschke K, Rothstein M, et al.

Search for cis-acting factors and maternal effect variants in Silver-Russell patients with ICR1 hypomethylation and their mothers

Eur J Hum Genet 2019 Jan; 27(1): 42-8

Soellner L, Kraft F, Sauer S, Begemann M, Kurth I, Elbracht M, et al.

Molecular Processes Connecting DNA Methylation Patterns with DNA Methyltransferases and Histone Modifications in Mammalian Genomes

Genes 2018, 9(11), 566; doi:10.3390/genes9110566

Jeltsch A., Broche J., Bashtrykov P.

Search for altered imprinting marks in Mayer-Rokitansky-Kuster-Hauser patients

Mol Genet Genom Med 2018 Nov; 6(6): 1225-8

Eggermann T, Ledig S, Begemann M, Elbracht M, Kurth I, Wieacker P

Molecular Processes Connecting DNA Methylation Patterns with DNA Methyltransferases and Histone Modifications in Mammalian Genomes

Genes 2018 Nov; 9(11): 20

Jeltsch A, Broche J, Bashtrykov P

Generation of two human isogenic iPSC lines from fetal dermal fibroblasts

Stem Cell Res. 2018 Oct 12;33:120-124. doi: 10.1016/j.scr.2018.10.004.

Tandon R, Brändl B, Baryshnikova N, Landshammer A, Steenpaß L, Keminer O, Pless O, Müller FJ.

Effects of Kaiso (binding) to the differentially methylated ICR1

Eur J Hum Genet 2018 Oct; 26: 747-

Bohne F, Langer D, Martine U, Eider CS, Enklaar T, Zechner U, et al.

Allele-Specific Epigenome Editing

Methods Mol Biol. 2018;1767:137-146. doi: 10.1007/978-1-4939-7774-1_6.

Bashtrykov P, Jeltsch A

The origin of imprinting defects in Temple syndrome and comparison with other imprinting disorders

Epigenetics 2018, in press

Beygo J, Mertel C, Kaya S, Gillissen-Kaesbach G, Eggermann T, Horsthemke B, Buiting K

Angelman Syndrome-Affected Individual with a Numerically Normal Karyotype and Isodisomic Paternal Uniparental Disomy of Chromosome 15 due to Maternal Robertsonian Translocation (14;15) by Monosomy Rescue.

Cytogenet Genome Res. 2018 Jul 18. doi: 10.1159/000490838.

Bramswig NC, Buiting K, Bechtel N, Horsthemke B, Rostasy K, Wieczorek D.

Maternal variants in NLRP and other maternal effect proteins are associated with multilocus imprinting disturbance in offspring

J Med Genet. 2018 Jul;55(7):497-504. doi: 10.1136/jmedgenet-2017-105190.

Begemann M, Rezwan FI, Beygo J, Docherty LE, Kolarova J, Schroeder C, Buiting K, Chokkalingam K, Degenhardt F, Wakeling EL, Kleinle S, González Fassrainer D, Oehl-Jaschkowitz B, Turner CLS, Patalan M, Gizewska M, Binder G, Bich Ngoc CT, Chi Dung V, Mehta SG, Baynam G, Hamilton-Shield JP, Aljareh S, Lokulo-Sodipe O, Horton R, Siebert R, Elbracht M, Temple IK, Eggermann T, Mackay DJG.

DNA Methylation Analysis by Bisulfite Conversion Coupled to Double Multiplexed Amplicon-Based Next-Generation Sequencing (NGS)

Methods Mol Biol. 2018;1767:367-382. doi: 10.1007/978-1-4939-7774-1_20.

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Locus-Specific DNA Methylation Analysis by Targeted Deep Bisulfite Sequencing

Methods Mol Biol. 2018;1767:351-366. doi: 10.1007/978-1-4939-7774-1_19.

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Molecular and clinical studies in 8 patients with Temple syndrome

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Nat Commun. 2018 May 15;9(1):1925. doi: 10.1038/s41467-018-04011-3

International Stem Cell Initiative (Allison TF, Müller FJ, Yamanaka S,...)

Sensory-Neuropathy-Causing Mutations in ATL3 Cause Aberrant ER Membrane Tethering

Cell Reports 2018 May; 23(7): 2026-38

Krols M, Detry S, Asselbergh B, Almeida-Souza L, Kremer A, Lippens S, et al.

Expert consensus document: Clinical and molecular diagnosis, screening and management of Beckwith-Wiedemann syndrome: an international consensus statement

Nat Rev Endocrinol. 2018 Apr;14(4):229-249. doi: 10.1038/nrendo.2017.166.

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Am J Hum Genet 2018 Apr; 102(4): 557-73

Vogtle FN, Brandl B, Larson A, Pendziwiat M, Friederich MW, White SM, et al.

Structural and sequence variants in patients with Silver-Russell syndrome or similar features- Curation of a disease database

Hum Mutat. 2018 Mar;39(3):345-364. doi: 10.1002/humu.23382. Epub 2018 Jan 11.

Tümer Z, López-Hernández JA, Netchine I, Elbracht M, Grønskov K, Gede LB, Sachwitz J, den Dunnen JT, Eggermann T

Recommendations for a nomenclature system for reporting methylation aberrations in imprinted Epigenetics.

2018;13(2):117-121. doi: 10.1080/15592294.2016.1264561.

Monk D, Morales J, den Dunnen JT, Russo S, Court F, Prawitt D, Eggermann T, Beygo J, Buiting K, Tümer Z; Nomenclature group of the European Network for Human Congenital Imprinting Disorders.

Mosaic genome-wide maternal isodiploidy: an extreme form of imprinting disorder presenting as prenatal diagnostic challenge

Clin Epigenetics. 2017 Oct 13;9:111. doi: 10.1186/s13148-017-0410-y. eCollection 2017.

Bens S, Luedeke M, Richter T, Graf M, Kolarova J, Barbi G, Lato K, Barth TF, Siebert R

The maternal uniparental disomy of chromosome 6 (upd(6)mat) “phenotype”: result of placental trisomy 6 mosaicism?

Molecular Genetics & Genomic Medicine 2017:22 SEP

Eggermann T, Oehl-Jaschkowitz B, Dicks S, Thomas W, Kanber D, Albrecht B, Begemann M, Kurth I, Beygo J, Buiting K.

New insights into the imprinted MEG8-DMR in 14q32 and clinical and molecular description of novel patients with Temple syndrome

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CryoPause: A New Method to Immediately Initiate Experiments after Cryopreservation of Pluripotent Stem Cells

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Epigenome Editing in the Brain

Adv Exp Med Biol. 2017;978:409-424. doi: 10.1007/978-3-319-53889-1_21.

Bashtrykov P, Jeltsch A.

Targeted Next Generation Sequencing Approach in Patients Referred for Silver-Russell Syndrome Testing Increases the Mutation Detection Rate and Provides Decisive Information for Clinical Management

Meyer R, Soellner L, Begemann M, Dicks S, Fekete G, Rahner N, Zerres K, Elbracht M, Eggermann T.

J Pediatr. 2017 May 19. pii: S0022-3476(17)30501-2. doi: 10.1016/j.jpeds.2017.04.018. [Epub ahead of print]

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Clin Genet 2017 Jan; 91(1): 73-8

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Recommendations for a nomenclature system for reporting methylation aberrations in imprinted domains

Epigenetics. 2016 Dec 2:0. [Epub ahead of print] Monk D, Morales J, den Dunnen JT, Russo S, Court F, Prawitt D, Eggermann T, Beygo J, Buiting K, Tümer Z.

Novel deletion in 11p15.5 imprinting center region 1 in a patient with Beckwith-Wiedemann syndrome provides insight into distal enhancer regulation and tumorigenesis

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Phenotypic spectrum and extent of DNA methylation defects associated with multilocus imprinting disturbances

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Katja Eggermann, Jet Blik, Frédéric Brioude, Elizabeth Algar, Karin Buiting, Silvia Russo, Zeynep Tümer, David Monk, Gudrun Moore, Thalia Antoniadi, Fiona Macdonald, Irène Netchine, Paolo Lombardi, Lukas Soellner, Matthias Begemann, Dirk Prawitt, Eamonn R Maher, Marcel Mannens, Andrea Riccio, Rosanna Weksberg, Pablo Lapunzina, Karen Grønskov, Deborah JG Mackay, and Thomas Eggermann,

DETAILS

Correction of aberrant imprinting by allele-specific epigenome editing

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