

Wessex Regional Genetics Laboratory Publications 2019

Jeffries AR, Maroofian R, Salter CG, Chioza BA, Cross HE, Patton MA, Dempster E, Temple IK, **Mackay DJG**, Rezwan FI, Aksglaede L, Baralle D, Dabir T, Hunter MF, Kamath A, Kumar A, Newbury-Ecob R, Selicorni A, Springer A, Van Maldergem L, Varghese V, Yachelevich N, Tatton-Brown K, Mill J, Crosby AH, Baple EL. Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. *Genome Res.* 2019 **29**: 1057-1066

Krzyzewska IM, Alders M, Maas SM, Blik J, Venema A, Henneman P, Rezwan FI, v. d. Lip K, Mul AN, **Mackay DJG**, Mannens MMAM. Genome-wide methylation profiling of Beckwith-Wiedemann syndrome patients without molecular confirmation after routine diagnostics. *Clin. Epigenetics* 2019 **11**: 53

Leme P, Dignum H, Ganczakowski M, **Chiecchio L**, Ayto R, **Baker K**, Singh S, Cranfield T, Matthias G, James C, Corser R. Prolonged survival of a 79-year-old man with acute myeloid leukemia M2, normal karyotype, *NPM1* and *FLT3-ITD* mutations, WBC $33.7 \times 10^9/L$, and involving only granulocyte-macrophage line on 53 cycles of low-dose cytarabine. *Ann. Hematol. Oncol.* 2019 **6** (11): id1276.

Levavi H, Tripodi J, Marcellino B, Mascarenhas J, Jones AV, **Cross NCP**, Gruenstein D, Najfeld V. A novel t(1;9)(p36;p24.1) *JAK2* translocation and review of the literature. *Acta Haematol.* 2019 **142**: 105-112

Lord J, McMullan DJ, Eberhardt RY, Rinck G, Hamilton SJ, Quinlan-Jones E, Prigmore E,

Monk D, **Mackay DJG**, Eggermann T, Maher ER, Riccio A. Genomic imprinting disorders: lessons on how genome, epigenome and environment interact. *Nat. Rev. Genet.* 2019 **20**: 235-248

Nolin SL, Glicksman A, Tortora N, Allen E, **Macpherson J**, Mila M, Vianna-Morgante AM, Sherman SL, Dobkin C, Latham GJ, Hadd AG. Expansions and contractions of the *FMR1* CGG repeat in 5,508 transmissions of normal, intermediate, and premutation alleles. *Am. J. Med. Genet.* 2019 **179A**: 1148-1156.

Ward D, Griffiths HL, Sood R, Denniston AK, Self JE, Ennis S, Lotery AJ, Gibson J. Comprehensive sequencing of the myocilin gene in a selected cohort of severe primary open-angle glaucoma patients. *Scientific Reports* 2019 **9**: id3100

O'Gorman L, Norman CS, Michaels L, Newall T, Crosby AH, **Mattocks C**, Cree AJ, Lotery AJ, Baple EL, Ratnayaka JA, Baralle D, Lee H, Osborne D, Shawkat F, Gibson J, Ennis S, Self JE. A small gene sequencing panel realises a high diagnostic rate in patients with congenital nystagmus following basic phenotyping. *Scientific Reports* 2019 **9**: id13229.

O'Sullivan JM, Hamblin A, Yap C, Fox S, Boucher R, Panchal A, Alimam S, Dreau H, Howard K, Ware P, **Cross NCP**, McMullin MF, Harrison CN, Mead AJ. The poor outcome in high molecular risk, hydroxycarbamide resistant/intolerant ET is not ameliorated by ruxolitinib. *Blood* 2019 **134** (23): 2107-2111

Patel AB, Franzini A, Leroy E, Kim SJ, Pomicter AD, Genet L, Xiao M, Yan D, Ahmann JM, Agarwal AM, Clair P, Addada J, Lambert J, **Salmon M**, Gleich GJ, **Cross NCP**, Constantinescu SN, O'Hare T, Prchal JT, Deininger MW. *JAK2^{ex13InDel}* drives oncogenic transformation and is associated with chronic eosinophilic leukemia and polycythemia vera. *Blood* 2019 **134** (26): 2388-2398

Schoemaker MJ, Jones ME, Higgins CD, Wright AF, **United Kingdom Clinical Cytogenetics Group**, Swerdlow AJ. Mortality and cancer incidence in carriers of balanced Robertsonian translocations: a national cohort study. *PLoS Med.* 2019 **16**: e1005471.