Wessex Regional Genetics Laboratory Publications for 2017

Alikian M, Whale AS, Akiki S, Piechocki K, Torrado C, Myint T, Cowen S, Griffiths M, Reid AG, Apperley J, **White H**, Huggett JF, Foroni L. RT-qPCR and RT-Digital PCR: A comparison of different platforms for the evaluation of residual disease in chronic myeloid leukemia. *Clin Chemistry* 2017 **63.2**:525-531.

Butt NM, Lambert J, Ali S, Beer PA, **Cross NCP**, Duncombe A, Ewing J, Harrison CN, Knapper S, McLornan D, Mead AJ, R D, Bain BJ, on behalf of the British Committee for Standards in Haematology. Guideline for the investigation and management of eosinophilia. *Br J Haemat* 2017 **176**: 553-572.

Cooper R, Markham H, Theaker J, Bateman A, **Bunyan D**, Sommerlad M, Crawford G, Eccles D. Case Report: Primary clear cell microcystic adenoma of the sinonasal cavity: pathological or fortuitous association? *Hindawi* 2017 doi.org/10.1155/2017/9236780.

Defour J-P, Hoade Y, Reuther A-M, **Callaway A**, **Ward D**, Chen F, Constantinescu SN, **Cross ~NCP**. An unusual, activating insertion/deletion MPL mutant in primary myelofibrosis. *Leukemia* 2017 *Letters to the Editor* **31**: 1838-1839.

Ghazzawi M, Mehra V, Knut M, Brown L, Tapper W, **Chase A**, de Lavallade H, **Cross NCP**. A novel PCM1-PDGFRB fusion in a patient with a chronic myeloproliferative neoplasm and an ins(8;5). *Acta Haematol* 2017 **138**:198-200.

Giles FJ, Rea D, Rosti G, **Cross NCP**, Steegmann JL, Griskevicius L, le Coutre P, Coriu D, Petrov L, Ossenkoppele GJ, Mahon F-X, Saussele S, Hellmann A, Koskenvesa P, Brümmendorf TH, Gastl G, Castagnetti F, Vincenzi B, Haenig J, Hochhaus A. Impact of age on efficacy and toxicity of nilotinib in patients with chronic myeloid leukemia in chronic phase: ENEST1st subanalysis. *J Cancer Res Clin Oncol* 2017 **143**:1585-1596.

Gunawan AS, McLornan DP, Wilkins B, **Waghorn K**, Hoade Y, **Cross NCP**, Harrison CN. Ruxolitinib, a potent JAK1/JAK2 inhibitor, induces temporary reductions in the allelic burden of concurrent CSF3R mutations in chronic neutrophilic leukemia. *Haematologica* Case Report: 2017: 102:e238.

Hochhaus A, Mahon F-X, le Coutre P, Petrov L, Janssen JJWM, **Cross NCP**, Rea D, Castagnetti F, hellmann A, Rosti G, Gattermann N, Coronal MLP, Gutierrez MAE, Garcia-Gutierrez V, Vincenzi B, DezzNI l, Giles FJ. Nilotinib first-line therapy in patients with Philadelphia chromosome-negative/BCR-ABL-positive chronic myeloid leukemia in chronic phase: ENEST1st sub-analysis. *J Cancer Res Clin Oncol* 2017 **143**:1225-1233.

Jawhar M, Naumann N, Knut M, **Score J**, Ghazzawi M, Schneider B, Kreuzer K-A, Hallek M, Drexler HG, Chacko J, Wallis L, Fabarius A, Metzgeroth G, Hofmann W-K, **Chase A**, Tapper W, Reiter A, **Cross NCP**. Cytogenetically criptic ZMYM2-FLT3 and DIAPH1-PDGFRB gene fusions in myeloid neoplasms with eosinophilia. *Leukemia* 2017 **31**:2271-2273

Jawhar M, Naumann N, Schwaab J, Baurmann H, Casper J, Dang T-A, Dietze L, Döhner K, Hänel A, Lathan B, Link H, Lotfi S, Maywald O, Mielke S, Müller L, Platzbecker U, Prümmer O, Thomssen H, Töpelt K, Panse J, Vicker T, Hofmann W-K, Haferlach T, Haferlach C, Fabarius A, Hochhaus A, **Cross NCP**, Reiter A, Metzgeroth G. Imatinib in myeloid/lymphoid neoplasms with eosinophilia and rearrangement of PDGFRB in chronic or blast phase. *Ann Hematol* 2017 **96**:1463-1470.

Jawhar M, Schwaab J, Meggendorfer M, Naumann N, Horny H-P, Sotlar K, Haferlach T, Schmitt K, Fabarius A, Valent P, Hofmann W-K, **Cross NCP**, Metzgeroth G, Reiter A. The clinical and molecular diversity of mast cell leukemia with or without associated hematologic neoplasm. *Haematologica* 2017 **102**:1035-1043.

Jawhar M, Schwaab J, Naumann N, Horny H-P, Sotlar K, Hasferlach T, Metzgeroth G, Fabarius A, Valent P, Hofmann W-K, **Cross NCP**, Meggendorfer M, Reiter A. Response and progression on midostaurin in advanced systemic mastocytosis: KIT1 D816V and other molecular markers. *Blood* 2017 **130**:137-145.

Mackay DJG, Temple IK. Human imprinting disorders: Principles, practice, problems and progress. *Eur J Med Genet* 2017 **60**:618-626.

Norman CS, O'Gorman L, Gibson J, Pengelly RJ, Baralle D, Ratnayaka JA, Griffiths H, Rose-Zerilli M, Ranger M, **Bunyan D**, Lee H, Page R, Newall T, Shawkat F, **Mattocks C**, **Ward D**, Ennis S, Self JE. Identification of a functionally significant tri-allelic genotype in the Tyrosinase gene (TYR) causing hypomorphic oculocutaneous albinism (OCA1B). *Nature Scientific Reports* 2017 **4415**:DOI:1038/s41598-017-04401-5.

Pace NP, M**aggouta F, Twigden M**, Borg I. Molecular cytogenetic characterisation of a novel *de novo* ring chromosome 6 involving a terminal 6p[deletion and terminal 6q duplication in the different arms of the same chromosome. *Mol Cytogenet* 2017 10:9

Rae W, Gao Y, **Ward D, Mattocks CJ**, Eren E, Williams AP. A novel germline gain-of-function variant in PIK3CD. *Clin Immunology* 2017 *Letter to the Editor* **181**:29-31.

Rae W. **Ward D**, **Mattocks CJ**, Gao Y, pengelly RJ, Patel SV, Ennis S, Faust SN, Williams AP. Autoimmunity/inflammation in a monogenic primary immunodeficiency cohort. *Clin & Trans Immunology* 2017 **6**, e155; doi:10.1038/cti.

Rinke J, Müller JP, Blaess MF, **Chase A,** Meggendorfer M, Schäfer V, Winkelmann N, Haferlach C, **Cross NCP**, Hochhaus A, Ernst T. Molecular characterization of EZH2 mutant patients with myelodysplastic/myeloproliferative neoplasms. *Leukemia* 2017 **31**:1936-1943.

Scott S, Travis D, Whitby L, Bainbridge J, **Cross NCP**, Barnett D. Measurement of BCR-ABL1 by RT-qPCR in chronis myeloid leukaemia: findings from an international EQA programme. *Br J Haemat* 2017 doi 10.1111/bjh.14557.

Seaby EG, **Bunyan DJ**, Ennis S, Gilbert RD. Sporadic isolated Fanconi Syndrome due to a mutation of EHHADH: A case report. *J Clin Nephrol Ren Care* 2017 **3**.027.

Silver RT, Baarel AC, Lascu E, Ritchie EK, Roboz GJ, Christos PJ, Orazi A, Hassana DC, Tam W, **Cross NCP**. The effect of initial molecular profile on response to recombinant interferon-α (rIFNα) treatment in early myelofibrosis. *Cancer* 2017 2680-2687

Silver RT, Krichevsky S, Gjoni S, **Cross NCP**. Evaluation of serum erythropoietin values as defined by 2016 World Health Organization criteria for the diagnosis of polycythemia vera. *Leuk & Lymphoma* 2017 dx.doi.org/10.1080/10428194.2017.1300892.

Tadros S, Wang R, Waters JJ, **Waterman C**, Collins AL, **Collinson MN**, Ahn JW, Josifova D, Chetan R, Kumar A. Inherited 2q23.1 microdeletions involving the MBD5 locus. *Mol Genet & Genomic Medicine* 2017 **5**:608-613.

Wakeling EL, Brioude F, Lokulo-Sodipe O, O'Connell SM, Salem J, Bliek J, Canton APM, Chrzanowska KH, Davies JH, Dias RP, Dubern B, Elbracht M, Giabicani E, Grimberg A, Grønskov K, Hokken-Koelega ACS, Jorge AA, Kagami M, Linglart A, Maghnie M, Mohnike K, Monk D, Moore GE, Murray PG, Ogata T, Petit IO, Russo S, Said E, Toumba M, Tümer Z, Binder G, Eggermann T, Harbison MD, Temple IK, **Mackay DJG**, Netchine I. Diagnosis and management of Silver-Russell syndrome: first international consensus statement. *Nat Reviews Endocrinology* 2017 **13**:105-124.

Yip BH, Steeples V, Repapi E, Armstrong RN, Llorian M, Roy S, Shaw J, Dolatshad H, Taylor S, Verma A, Bartenstein M, Vyas P, **Cross NCP**, Malcovati L, Cazzola M, Hellström-Lindberg E, Ogawa S, Smith CWJ, Pellagatti A, Boultwood J. The U2AF1^{S34F} mutation induces lineage-specific splicing alterations in myelodysplastic syndromes. *J Clin Investigation* 2017 **127** 2206-2221

Zoi K and **Cross NCP**. Genomics of myeloproliferative neoplasms. *J Clin Oncology* 2017 **35**:947-955.