

## Wessex Regional Genetics Laboratory Publications 2020

Abecasis M, **Cross NCP**, Brito M, Ferreira I, Sakamoto KM, Hijiya N, Score J, Gale RP. 2020. Is cancer latency an outdated concept? Lessons from chronic myeloid leukemia. *Leukemia* **34**: 2279-2284.

Ashton JJ, Mossotto E, Stafford IS, Haggarty R, Coelho TAF, Batra A, Afzal NA, Mort M, **Bunyan DJ**, Beattie RM, Ennis S. 2020. Genetic sequencing of pediatric patients identifies mutations in monogenic inflammatory bowel disease genes that translate to distinct clinical phenotypes. *Clin. Transl. Gastroen.* **11** (2): e00129.

Blakes AJM, Gaul E, Lam W, Shannon N, Knapp KM, Bicknell LS, Jackson MR, Wade EM, Robertson S, White SM, Heller R, **Chase A**, Baralle D, Douglas AGL. 2020. Pathogenic variants causing *ABL1* malformation syndrome cluster in a myristoyl-binding pocket and increase tyrosine kinase activity. *Eur. J. Hum. Genet.* **29** (4): 593-603 [PMID: 33223528].

**Chase A**, Score J, **Lin F**, Bryant C, **Waghorn K**, **Yapp S**, Carreno-Tarragona G, Aranaz P, Villasante A, Ernst T, **Cross NCP**. 2020. Mutational mechanisms of *EZH2* inactivation in myeloid neoplasms. *Leukemia* **34**: 3206-3214.

**Cross E**, **Duncan-Flavell PJ**, **Howarth RJ**, **Crooks RO**, **Thomas NS**, **Bunyan DJ**. 2020. Screening of a large *PAX6* cohort identified many novel variants and emphasises the importance of the paired and homeobox domains. *Eur. J. Med. Genet.* **63** (7): 103940 [PMID: 32360764].

**Cross E**, **Duncan-Flavell PJ**, **Howarth RJ**, **Hobbs JI**, **Thomas NS**, **Bunyan DJ**. 2020. Screening of a large Rubinstein-Taybi cohort identified many novel variants and emphasises the importance of the *CREBBP* Histone acetyltransferase domain. *Am. J. Med. Genet.* **182A** (11): 2508-2520.

Dawoud AAZ, Tapper WJ, **Cross NCP**. 2020. Clonal myelopoiesis in the UK Biobank cohort: *ASXL1* mutations are strongly associated with smoking. *Leukemia* **34**: 2660-2672.

Elbracht M, **Mackay D**, Begemann M, Kagan KO, Eggermann T. 2020. Disturbed genomic imprinting and its relevance for human reproduction: causes and clinical consequences. *Human Reproduction Update* **26** (2): 197-213.

**Kirk B**, Kharbanda M, **Bateman MS**, Hunt D, **Taylor E-J**, Collins AL, **Bunyan DJ**, **Collinson MN**, **Russell LM**, **Bowell S**, Barber JCK. 2020. Directly transmitted 12.3 Mb deletion with a consistent phenotype in the variable 11q21-q22.3 region. *Cytogenet. Genome Res.* **160**: 185-192.

Lokulo-Sodipe O, Ballard L, Child J, Inskip HM, Byrne CD, Ishida M, Moore GE, Wakeling EL, Fenwick A, **Mackay DJG**, Davies JH, Temple IK. 2020. Phenotype of genetically confirmed Silver-Russell syndrome beyond childhood. *J. Med. Genet.* **57** (10): 683-691 [PMID: 32054688].

Machova Polakova K, Zizkova H, Zuna J, Motlova E, Hovorkova L, Gottschalk A, Glauche I, Koblihova J, Pecherkova P, Klamova H, Stastna Markova M, Srbova D, Benesova A, Polivkova V, Jurcek T, Zackova D, Mayer J, Ernst T, Mahon FX, Saussele S, Roeder I,

**Cross NCP**, Hochhaus A. 2020. Analysis of chronic myeloid leukaemia during deep molecular response by genomic PCR: a traffic light stratification model with impact on treatment-free remission. *Leukemia* **34**: 2113-2124.

Metzgeroth G, Schwaab J, Naumann N, Jawhar M, Haferlach T, Fabarius A, Hochhaus A, Hofmann WK, **Cross NCP**, Reiter A. 2020. Treatment-free remission in *FIPILI-PDGFR*A-positive myeloid/lymphoid neoplasms with eosinophilia after imatinib discontinuation. *Blood Adv.* **4** (3): 440-443.

Pengelly RJ, **Ward D**, Hunt D, **Mattocks C**, Ennis S. 2020. Comparison of Mendelian exome capture kits for use in clinical diagnostics. *Scientific Reports* **10**: 3235

Rinke J, **Chase A**, **Cross NCP**, Hochhaus A, Ernst T. 2020. *EZH2* in Myeloid Malignancies. *Cells* **9** (7): 1639 [PMID: 32650416].

Sakka SD, Mann K, Lachlan K, Davies JH, **Bateman M**, Holder-Espinasse M, Arya VB. 2020. Haematological chimerism masquerading as disorder of sex development. *Clin. Endocrinol.* **92** (5): 487-489.

Schwaab J, Cabral do O Hartmann N, Naumann N, Jawhar M, Weiß C, Metzgeroth G, Schmid A, Lübke J, Reiter L, Fabarius A, **Cross NCP**, Sotlar K, Valent P, Kluin-Nelemans HC, Hofmann WK, Horny H-P, Panse J, Reiter A. 2020. Importance of adequate diagnostic work-up for correct diagnosis of advanced systemic mastocytosis. *J. Allergy Clin. Immunol. in Practice* **8** (9): 3121-3127.e1.

Schwaab J, Naumann N, Luebke J, Jawhar M, Somervaille TCP, Williams MS, Frewin R, Jost PJ, Lichtenegger FS, La Rosée P, Storch N, Haferlach T, Horny H-P, Fabarius A, Haferlach C, Burchert A, Hofmann WK, **Cross NCP**, Hochhaus A, Reiter A, Metzgeroth G. 2020. Response to tyrosine kinase inhibitors in myeloid neoplasms associated with *PCMI-JAK2*, *BCR-JAK2* and *ETV6-ABL1* fusion genes. *Am. J. Hematol.* **95**: 824-833.

Smith G, Apperley J, Milojkovic D, **Cross NCP**, Foroni L, Byrne J, Goringe A, Rao A, Khorashad J, de Lavallade H, Mead AJ, Osborne W, Plummer C, Jones G, Copland M; British Society for Haematology. 2020. A British Society for Haematology Guideline on the diagnosis and management of chronic myeloid leukaemia. *Br. J. Haematol.* **191**: 171-193.

Wai HA, Lord J, Lyon M, Gunning A, Kelly H, Cibir P, Seaby EG, Spiers-Fitzgerald K, Lye J, Ellard S, **Thomas NS**, **Bunyan DJ**, Douglas AGL, Baralle D, Splicing and Disease Working Group. 2020. Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. *Genetics in Medicine* **22** (6): 1005-1014 [PMID: 32235935].