

Wessex Regional Genetics Laboratory Publications 2022

Blakes AJM, Wai HA, Davies I, Moledina HE, Ruiz A, Thomas T, **Bunyan D, Thomas NS,**

E, Patel KP, Patkar N, Picarsic J, Platzbecker U, Roberts I, Schuh A, Sewell W, Siebert R, Tembhare P, Tyner J, Verstovsek S, Wang W, Wood B, Xiao W, Yeung C, Hochhaus A. 2022. The 5th edition of the World Health Organization classification of haematolymphoid tumours: myeloid and histiocytic/dendritic neoplasms. *Leukemia* **36** (7): 1703-1719 [PMID: 35732831].

Lafferty N, **Salmon M**, **Cross NCP**, Singer I, Cooney A, Jayaprakash R. 2022. Chronic eosinophilic leukaemia associated with *JAK2* exon 13 insertion/deletion mutations. *Acta Haematol.* **145** (2): 201-206 [PMID: 34515041].

Lin S, Sanchez-Bretaña A, Leslie JS, Williams KB, Lee H, **Thomas NS**, **Callaway J**, Deline J, Arjuna Ratnayaka J, Baralle D, Schmitt MA, Norman CS, Hammond S, Harlalka GV, Ennis S, Cross HE, Wenger O, Crosby AH, Baple EL, Self JE. 2022. Evidence that the Ser192Tyr/Arg402Gln *in cis* Tyrosinase gene haplotype is a disease-causing allele in oculocutaneous albinism type 1B (OCA1B). *Genomic Medicine* **7** (1): 2 [PMID: 35027574].

Loong L, Cubuk C, Choi S, Allen S, Torr B, Garrett A, Loveday C, Durkie M, **Callaway A**, Burghel GJ, Drummond J, Robinson R, Berry IR, Wallace A, Eccles DM, Tischkowitz M, Ellard S, Ware JS, Hanson H, Turnbull C; on behalf of CanVIG-UK. 2022. Quantifying prediction of pathogenicity for within-codon concordance (PM5) using 7541 functional classifications of *BRCA1* and *MSH2* missense variants. *Genet. Med.* **24** (3): 552-563. [PMID: 34906453]

Loong L, Garrett A, Allen S, Choi S, Durkie M, **Callaway A**, Drummond J, Burghel GJ, Robinson R, Torr B, Berry IR, Wallace AJ, Eccles DM, Ellard S, Baple E, Evans DG, Woodward ER, Kulkarni A, Lalloo F, Tischkowitz M, Lucassen A, Hanson H, Turnbull C; on behalf of the CanVIG-UK. 2022. Reclassification of clinically-detected sequence variants: Framework for genetic clinicians and clinical scientists by CanVIG-UK (Cancer Variant Interpretation Group UK). *Genet. Med.* **24** (9): 1867-1877.

Lübke J, Schwaab J, Naumann N, Horny H-P, Weiß C, Metzgeroth G, Kreil S, **Cross NCP**, Sotlar K, Fabarius A, Hofmann W-K, Valent P, Gotlib J, Jawhar M, Reiter A. 2022. Superior efficacy of midostaurin over cladribine in advanced systemic mastocytosis: a registry-based analysis. *J. Clin. Oncol.* **40** (16): 1783-1794.

Mackay D, Blik J, Kagami M, Tenorio-Castano J, Pereda A, Brioude F, Netchine I, Papingi D, de Franco E, **Lever M**, **Sillibourne J**, Lombardi P, Gaston V, Tauber M, Diene G, Bieth E, Fernandez L, Nevado J, Tümer Z, Riccio A, Maher ER, Beygo J, Tannorella P, Russo S, Perez de Nanclares G, Temple IK, Ogata T, Iqbal Z, O'Connell P, Egger M, Haddad B, Wainwright N, Jones K, Kinniburtt D, Kinniburtt J, Kinniburtt M, Kinniburtt R, Kinniburtt S, Kinniburtt T, Kinniburtt U, Kinniburtt V, Kinniburtt W, Kinniburtt X, Kinniburtt Y, Kinniburtt Z, Kinniburtt AA, Kinniburtt AB, Kinniburtt AC, Kinniburtt AD, Kinniburtt AE, Kinniburtt AF, Kinniburtt AG, Kinniburtt AH, Kinniburtt AI, Kinniburtt AJ, Kinniburtt AK, Kinniburtt AL, Kinniburtt AM, Kinniburtt AN, Kinniburtt AO, Kinniburtt AP, Kinniburtt AQ, Kinniburtt AR, Kinniburtt AS, Kinniburtt AT, Kinniburtt AU, Kinniburtt AV, Kinniburtt AW, Kinniburtt AX, Kinniburtt AY, Kinniburtt AZ, Kinniburtt BA, Kinniburtt BB, Kinniburtt BC, Kinniburtt BD, Kinniburtt BE, Kinniburtt BF, Kinniburtt BG, Kinniburtt BH, Kinniburtt BI, Kinniburtt BJ, Kinniburtt BK, Kinniburtt BL, Kinniburtt BM, Kinniburtt BN, Kinniburtt BO, Kinniburtt BP, Kinniburtt BQ, Kinniburtt BR, Kinniburtt BS, Kinniburtt BT, Kinniburtt BU, Kinniburtt BV, Kinniburtt BW, Kinniburtt BX, Kinniburtt BY, Kinniburtt BZ, Kinniburtt CA, Kinniburtt CB, Kinniburtt CC, Kinniburtt CD, Kinniburtt CE, Kinniburtt CF, Kinniburtt CG, Kinniburtt CH, Kinniburtt CI, Kinniburtt CJ, Kinniburtt CK, Kinniburtt CL, Kinniburtt CM, Kinniburtt CN, Kinniburtt CO, Kinniburtt CP, Kinniburtt CQ, Kinniburtt CR, Kinniburtt CS, Kinniburtt CT, Kinniburtt CU, Kinniburtt CV, Kinniburtt CW, Kinniburtt CX, Kinniburtt CY, Kinniburtt CZ, Kinniburtt DA, Kinniburtt DB, Kinniburtt DC, Kinniburtt DD, Kinniburtt DE, Kinniburtt DF, Kinniburtt DG, Kinniburtt DH, Kinniburtt DI, Kinniburtt DJ, Kinniburtt DK, Kinniburtt DL, Kinniburtt DM, Kinniburtt DN, Kinniburtt DO, Kinniburtt DP, Kinniburtt DQ, Kinniburtt DR, Kinniburtt DS, Kinniburtt DT, Kinniburtt DU, Kinniburtt DV, Kinniburtt DW, Kinniburtt DX, Kinniburtt DY, Kinniburtt DZ, Kinniburtt EA, Kinniburtt EB, Kinniburtt EC, Kinniburtt ED, Kinniburtt EE, Kinniburtt EF, Kinniburtt EG, Kinniburtt EH, Kinniburtt EI, Kinniburtt EJ, Kinniburtt EK, Kinniburtt EL, Kinniburtt EM, Kinniburtt EN, Kinniburtt EO, Kinniburtt EP, Kinniburtt EQ, Kinniburtt ER, Kinniburtt ES, Kinniburtt ET, Kinniburtt EU, Kinniburtt EV, Kinniburtt EW, Kinniburtt EX, Kinniburtt EY, Kinniburtt EZ, Kinniburtt FA, Kinniburtt FB, Kinniburtt FC, Kinniburtt FD, Kinniburtt FE, Kinniburtt FF, Kinniburtt FG, Kinniburtt FH, Kinniburtt FI, Kinniburtt FJ, Kinniburtt FK, Kinniburtt FL, Kinniburtt FM, Kinniburtt FN, Kinniburtt FO, Kinniburtt FP, Kinniburtt FQ, Kinniburtt FR, Kinniburtt FS, Kinniburtt FT, Kinniburtt FU, Kinniburtt FV, Kinniburtt FW, Kinniburtt FX, Kinniburtt FY, Kinniburtt FZ, Kinniburtt GA, Kinniburtt GB, Kinniburtt GC, Kinniburtt GD, Kinniburtt GE, Kinniburtt GF, Kinniburtt GG, 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IV, Kinniburtt IW, Kinniburtt IX, Kinniburtt IY, Kinniburtt IZ, Kinniburtt JA, Kinniburtt JB, Kinniburtt JC, Kinniburtt JD, Kinniburtt JE, Kinniburtt JF, Kinniburtt JG, Kinniburtt JH, Kinniburtt JI, Kinniburtt JJ, Kinniburtt JK, Kinniburtt JL, Kinniburtt JM, Kinniburtt JN, Kinniburtt JO, Kinniburtt JP, Kinniburtt JQ, Kinniburtt JR, Kinniburtt JS, Kinniburtt JT, Kinniburtt JU, Kinniburtt JV, Kinniburtt JW, Kinniburtt JX, Kinniburtt JY, Kinniburtt JZ, Kinniburtt KA, Kinniburtt KB, Kinniburtt KC, Kinniburtt KD, Kinniburtt KE, Kinniburtt KF, Kinniburtt KG, Kinniburtt KH, Kinniburtt KI, Kinniburtt KJ, Kinniburtt KK, Kinniburtt KL, Kinniburtt KM, Kinniburtt KN, Kinniburtt KO, Kinniburtt KP, Kinniburtt KQ, Kinniburtt KR, Kinniburtt KS, Kinniburtt KT, Kinniburtt KU, Kinniburtt KV, Kinniburtt KW, Kinniburtt KX, Kinniburtt KY, Kinniburtt KZ, Kinniburtt LA, Kinniburtt LB, Kinniburtt LC, Kinniburtt LD, Kinniburtt LE, Kinniburtt LF, Kinniburtt LG, Kinniburtt LH, Kinniburtt LI, Kinniburtt LJ, Kinniburtt LK, Kinniburtt LL, Kinniburtt LM, Kinniburtt LN, Kinniburtt LO, Kinniburtt LP, Kinniburtt LQ, Kinniburtt LR, Kinniburtt LS, Kinniburtt LT, Kinniburtt LU, Kinniburtt LV, Kinniburtt LW, Kinniburtt LX, Kinniburtt LY, Kinniburtt LZ, Kinniburtt MA, Kinniburtt MB, Kinniburtt MC, Kinniburtt MD, Kinniburtt ME, Kinniburtt MF, Kinniburtt MG, Kinniburtt MH, Kinniburtt MI, Kinniburtt MJ, Kinniburtt MK, Kinniburtt ML, Kinniburtt MN, Kinniburtt MO, Kinniburtt MP, Kinniburtt MQ, Kinniburtt MR, Kinniburtt MS, Kinniburtt MT, Kinniburtt MU, Kinniburtt MV, Kinniburtt MW, Kinniburtt MX, Kinniburtt MY, Kinniburtt MZ, Kinniburtt NA, Kinniburtt NB, Kinniburtt NC, Kinniburtt ND, Kinniburtt NE, Kinniburtt NF, Kinniburtt NG, Kinniburtt NH, Kinniburtt NI, Kinniburtt NJ, Kinniburtt NK, Kinniburtt NL, Kinniburtt NM, Kinniburtt NN, Kinniburtt NO, Kinniburtt NP, Kinniburtt NQ, Kinniburtt NR, Kinniburtt NS, Kinniburtt NT, Kinniburtt NU, Kinniburtt NV, Kinniburtt NW, Kinniburtt NX, Kinniburtt NY, Kinniburtt NZ, Kinniburtt OA, Kinniburtt OB, Kinniburtt OC, Kinniburtt OD, Kinniburtt OE, Kinniburtt OF, Kinniburtt OG, Kinniburtt OH, Kinniburtt OI, Kinniburtt OJ, Kinniburtt OK, Kinniburtt OL, Kinniburtt OM, Kinniburtt ON, Kinniburtt OO, Kinniburtt OP, Kinniburtt OQ, Kinniburtt OR, Kinniburtt OS, Kinniburtt OT, Kinniburtt OU, Kinniburtt OV, Kinniburtt OW, Kinniburtt OX, Kinniburtt OY, Kinniburtt OZ, Kinniburtt PA, Kinniburtt PB, Kinniburtt PC, Kinniburtt PD, Kinniburtt PE, Kinniburtt PF, Kinniburtt PG, Kinniburtt PH, Kinniburtt PI, Kinniburtt PJ, Kinniburtt PK, Kinniburtt PL, Kinniburtt PM, Kinniburtt PN, Kinniburtt PO, Kinniburtt PP, Kinniburtt PQ, Kinniburtt PR, Kinniburtt PS, Kinniburtt PT, Kinniburtt PU, Kinniburtt PV, Kinniburtt PW, Kinniburtt PX, Kinniburtt PY, Kinniburtt PZ, Kinniburtt QA, Kinniburtt QB, Kinniburtt QC, Kinniburtt QD, Kinniburtt QE, Kinniburtt QF, Kinniburtt QG, Kinniburtt QH, Kinniburtt QI, Kinniburtt QJ, Kinniburtt QK, Kinniburtt QL, Kinniburtt QM, Kinniburtt QN, Kinniburtt QO, Kinniburtt QP, Kinniburtt QQ, Kinniburtt QR, Kinniburtt QS, Kinniburtt QT, Kinniburtt QU, Kinniburtt QV, Kinniburtt QW, Kinniburtt QX, Kinniburtt QY, Kinniburtt QZ, Kinniburtt RA, Kinniburtt RB, Kinniburtt RC, Kinniburtt RD, Kinniburtt RE, Kinniburtt RF, Kinniburtt RG, Kinniburtt RH, Kinniburtt RI, Kinniburtt RJ, Kinniburtt RK, Kinniburtt RL, Kinniburtt RM, Kinniburtt RN, Kinniburtt RO, Kinniburtt RP, Kinniburtt RQ, Kinniburtt RR, Kinniburtt RS, Kinniburtt RT, Kinniburtt RU, Kinniburtt RV, Kinniburtt RW, Kinniburtt RX, Kinniburtt RY, Kinniburtt RZ, Kinniburtt SA, Kinniburtt SB, Kinniburtt SC, Kinniburtt SD, Kinniburtt SE, Kinniburtt SF, Kinniburtt SG, Kinniburtt SH, Kinniburtt SI, Kinniburtt SJ, Kinniburtt SK, Kinniburtt SL, Kinniburtt SM, Kinniburtt SN, Kinniburtt SO, Kinniburtt SP, Kinniburtt SQ, Kinniburtt SR, Kinniburtt SS, Kinniburtt ST, Kinniburtt SU, Kinniburtt SV, Kinniburtt SW, Kinniburtt SX, Kinniburtt SY, Kinniburtt SZ, Kinniburtt TA, Kinniburtt TB, Kinniburtt TC, Kinniburtt TD, Kinniburtt TE, Kinniburtt TF, Kinniburtt TG, Kinniburtt TH, Kinniburtt TI, Kinniburtt TJ, Kinniburtt TK, Kinniburtt TL, Kinniburtt TM, Kinniburtt TN, Kinniburtt TO, Kinniburtt TP, Kinniburtt TQ, Kinniburtt TR, Kinniburtt TS, Kinniburtt TT, Kinniburtt TU, Kinniburtt TV, Kinniburtt TW, Kinniburtt TX, Kinniburtt TY, Kinniburtt TZ, Kinniburtt UA, Kinniburtt UB, Kinniburtt UC, Kinniburtt UD, Kinniburtt UE, Kinniburtt UF, Kinniburtt UG, Kinniburtt UH, Kinniburtt UI, Kinniburtt UJ, Kinniburtt UK, Kinniburtt UL, Kinniburtt UM, Kinniburtt UN, Kinniburtt UO, Kinniburtt UP, Kinniburtt UQ, Kinniburtt UR, Kinniburtt US, Kinniburtt UT, Kinniburtt UY, Kinniburtt UZ, Kinniburtt VA, Kinniburtt VB, Kinniburtt VC, Kinniburtt VD, Kinniburtt VE, Kinniburtt VF, Kinniburtt VG, Kinniburtt VH, Kinniburtt VI, Kinniburtt VJ, Kinniburtt VK, Kinniburtt VL, Kinniburtt VM, Kinniburtt VN, Kinniburtt VO, Kinniburtt VP, Kinniburtt VQ, Kinniburtt VR, Kinniburtt VS, Kinniburtt VT, Kinniburtt VU, Kinniburtt VV, Kinniburtt VW, Kinniburtt VX, Kinniburtt VY, Kinniburtt VZ, Kinniburtt WA, Kinniburtt WB, Kinniburtt WC, Kinniburtt WD, Kinniburtt WE, Kinniburtt WF, Kinniburtt WG, Kinniburtt WH, Kinniburtt WI, Kinniburtt WJ, Kinniburtt WK, Kinniburtt WL, Kinniburtt WM, Kinniburtt WN, Kinniburtt WO, Kinniburtt WP, Kinniburtt WQ, Kinniburtt WR, Kinniburtt WS, Kinniburtt WT, Kinniburtt WY, Kinniburtt WZ, Kinniburtt XA, Kinniburtt XB, Kinniburtt XC, Kinniburtt XD, Kinniburtt XE, Kinniburtt XF, Kinniburtt XG, Kinniburtt XH, Kinniburtt XI, Kinniburtt XJ, Kinniburtt XK, Kinniburtt XL, Kinniburtt XM, Kinniburtt XN, Kinniburtt XO, Kinniburtt XP, Kinniburtt XQ, Kinniburtt XR, Kinniburtt XS, Kinniburtt XT, Kinniburtt XU, Kinniburtt XV, Kinniburtt XW, Kinniburtt XX, Kinniburtt XY, Kinniburtt XZ, Kinniburtt YA, Kinniburtt YB, Kinniburtt YC, Kinniburtt YD, Kinniburtt YE, Kinniburtt YF, Kinniburtt YG, Kinniburtt YH, Kinniburtt YI, Kinniburtt YJ, Kinniburtt YK, Kinniburtt YL, Kinniburtt YM, Kinniburtt YN, Kinniburtt YO, Kinniburtt YP, Kinniburtt YQ, Kinniburtt YR, Kinniburtt YS, Kinniburtt YT, Kinniburtt YU, Kinniburtt YV, Kinniburtt YW, Kinniburtt YX, Kinniburtt YY, Kinniburtt YZ, Kinniburtt ZA, Kinniburtt ZB, Kinniburtt ZC, Kinniburtt ZD, Kinniburtt ZE, Kinniburtt ZF, Kinniburtt ZG, Kinniburtt ZH, Kinniburtt ZI, Kinniburtt ZJ, Kinniburtt ZK, Kinniburtt ZL, Kinniburtt ZM, Kinniburtt ZN, Kinniburtt ZO, Kinniburtt ZP, Kinniburtt ZQ, Kinniburtt ZR, Kinniburtt ZS, Kinniburtt ZT, Kinniburtt ZU, Kinniburtt ZV, Kinniburtt ZW, Kinniburtt ZX, Kinniburtt ZY, Kinniburtt ZZ.

towards a consensus strategy for multi-locus diagnostic testing of imprinting disorders. *Clin. Epigenetics* **14**: 143 [PMID: 36345041].

Moyo TK, Mendler JH, Itzykson R, Kishtagari A,

Salmon M, White HE, Zizkova H, Gottschalk A, Motlova E, Cerveira N, Colomer D, Coriu D, Franke GN, Gottardi E, Izzo B, Jurcek T, Lion T, Schäfer V, Venturi C, Vigneri P, Zawada M, Zuna J, Hovorkova L, Koblihova J, Klamova H, Stastna Markova M, Srbova D, Benesova A, Polivkova V, Zackova D, Mayer J, Roeder I, Glauche I, Ernst T, Hochhaus A, Machova Polakova K, **Cross NCP**. 2022. Impact of *BCR::ABL1* transcript type on RT-qPCR amplification performance and molecular response to therapy. *Leukemia* **36**: 1879-1886.

White HE, Salmon M, Albano F, Andersen CSA, Balabanov S, Balatzenko G, Barbany G, Cayuela J-M, Cerveira N, Cochaux P, Colomer D, Coriu D, Diamond J, Dietz C, Dulucq S, Engvall M, Franke GN, Gineikiene-Valentine E, Gniot M, Gómez-Casares MT, Gottardi E, Hayden C, Hayette S, Hedblom A, Ilea A, Izzo B, Jiménez-Velasco A, Jurcek T, Kairisto V, ihok L, Mitterbauer-Hohendanner G, Moeckel S, Naumann N, Nibourel O, Oppliger Leibundgut E, Panayiotidis P, Podgornik H, Pott C, Rapado I, Rose SJ, Schäfer V, Touloumenidou T, Veigaarmtrau5 g0 GM223(ne4(r)] TJETQq0.00000