Myeloid neoplasms with isolated del(5q) and *JAK2* V617F mutation: a "grey zone" combination of myelodysplastic and myeloproliferative features?

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Supplementary material

Table S1: Complete karyotype for each of the 6 cases reported.

Case 1	46,XX,del(5)(q22q33)[2]/46,XX[18]
Case 2	46,XX,del(5)(q13q33)[18]/46,XX[2]
Case 3	46,XX,del(5)(q22q35)[20]
Case 4	46,XX,del(5)(q13q31)[18]/46,XX[2]
Case 5	46,XY,del(5)(q13q33)[13]/46,XX[7]
Case 6	not available

NGS sequencing strategy:

Genomic DNA was fragmented to 3-4 kb by shearing using QSonica (Newtown, CT) Q800R2 instrument according to the manufacturer’s recommendations. Targeted enrichment of 45 genes (*ABL1, ASXL1, BCOR, BRAF, CALR, CBL, CEBPA, DNMT3A, ETV6, EZH2, FAM5C, FLT3, GATA1, GATA2, HNRNPK, IDH1, IDH2, IKZF1, JAK1, JAK2, KDM6A, KIT, KRAS, MPL, NFE2, NOTCH1, NPM1, NRAS, PHF6, PTPN11, RAD21, RUNX1, SETBP1, SF3B1, SH2B3, SMC1A, SMC3, SRSF2, STAG2, SUZ12, TET2, TP53, U2AF1, WT1, ZRSR2*) recurrently mutated in myeloid malignancies was performed using the Thunderstorm system (Raindance Technologies, Billerica, MA) using a custom primer panel followed by sequencing using the Illumina MiSeq (v3 chemistry) yielding 260-bp paired end reads.

Additional mutations detected in the *JAK2* wild type cohort.

In the *JAK2* wild type cohort (41 cases), 28 cases (68.3%) did not show any mutation among the ones tested. Conversely, 6 cases (14.6%) presented one mutation, 4 cases (9.8%) presented two mutations and 3 cases (7.3) presented three additional mutations. The most commonly mutated gene was *SF3B1* (8 cases), followed by *TP53* (3 cases), *ASXL1* (2 cases), *DNMT3A, TET2, SH2B3, U2AF1, RUNX1, BCOR* and *ETV6* (respectively 1 case each).