Frequent genomic abnormalities in acute myeloid leukemia/myelodysplastic syndrome with normal karyotype

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Citation: Akagi T, Ogawa S, Dugas M, Kawamata N, Yamamoto G, Nannya Y, Sanada M, Miller CW, Yung A, Schnittger S, Haferlach T, Haferlach C, and Koeffler HP. Frequent genomic abnormalities in acute myeloid leukemia/myelodysplastic syndrome with normal karyotype. Haematologica 2009. doi:10.3324/haematol.13024

Supplementary Table S1. Primers for determination of 6p SNP in AML #38.

Location	Position	SNP ID	Sequence
6p25.2	3,283,855	rs7747259	TTC TTC CCT TTT AGT TGC ATC C TGG GGT TAC TTT CTC CGT TG
6p25.1	5,000,549	rs1122637	GAG TGA AGG TAT ATT CAG TCT GGA G TGA GAT AAG TCT CAA ATA CGT TCT GG
6p24.3	7,815,342	rs9505293	GGA ACA CCC CGG TCT CTT AT GTG GAA AAG AAG GTG GCA GA
6p24.1	12,008,642	rs6934027	TCT GAG GCT TCC TTG GTC TC AAT TCC AGT GCA CTC CAT CC
6p23	14,052,796	rs280153	TTA ATC CTT TCA ATA CAC CTG TGA AAG CCC GGC TAA ATG TTC TT
6p22.3	15,898,879	rs191986	TCT GGG CCC TTC TTA ATC CT ACT CTC TTC CTC CCC AGT CC

Supplementary Table S2. Primers for determination of gene dosage and *JAK2* mutation.

Gene	Sequence	Melting temperature (°C)
CDKN2A	5'-GTG CCA AAG TGC TCC TGA AGC TG-3' 5'-AGC AAA TCT GTT TGG AGG TCTG-3	79.0
MYC	5'-CAG ATC AGC AAC AAC CGA AA-3' 5'-GGC CTT TTC ATT GTT TTC CA-3'	84.0
2p21 (control)	5'-GGC AAT CCT GGC TGC GGA TCA AGA-3' 5'-ATT TCT GAA CTT CTT GGC TGC C-3'	81.0
JAK2	5'-GGG TTT CCT CAG AAC GTT GA-3' 5'-TCA TTG CTT TCC TTT TTC ACA A-3'	-

Supplementary Table S3. Validation of SNP-chip analysis using FISH in case #11.

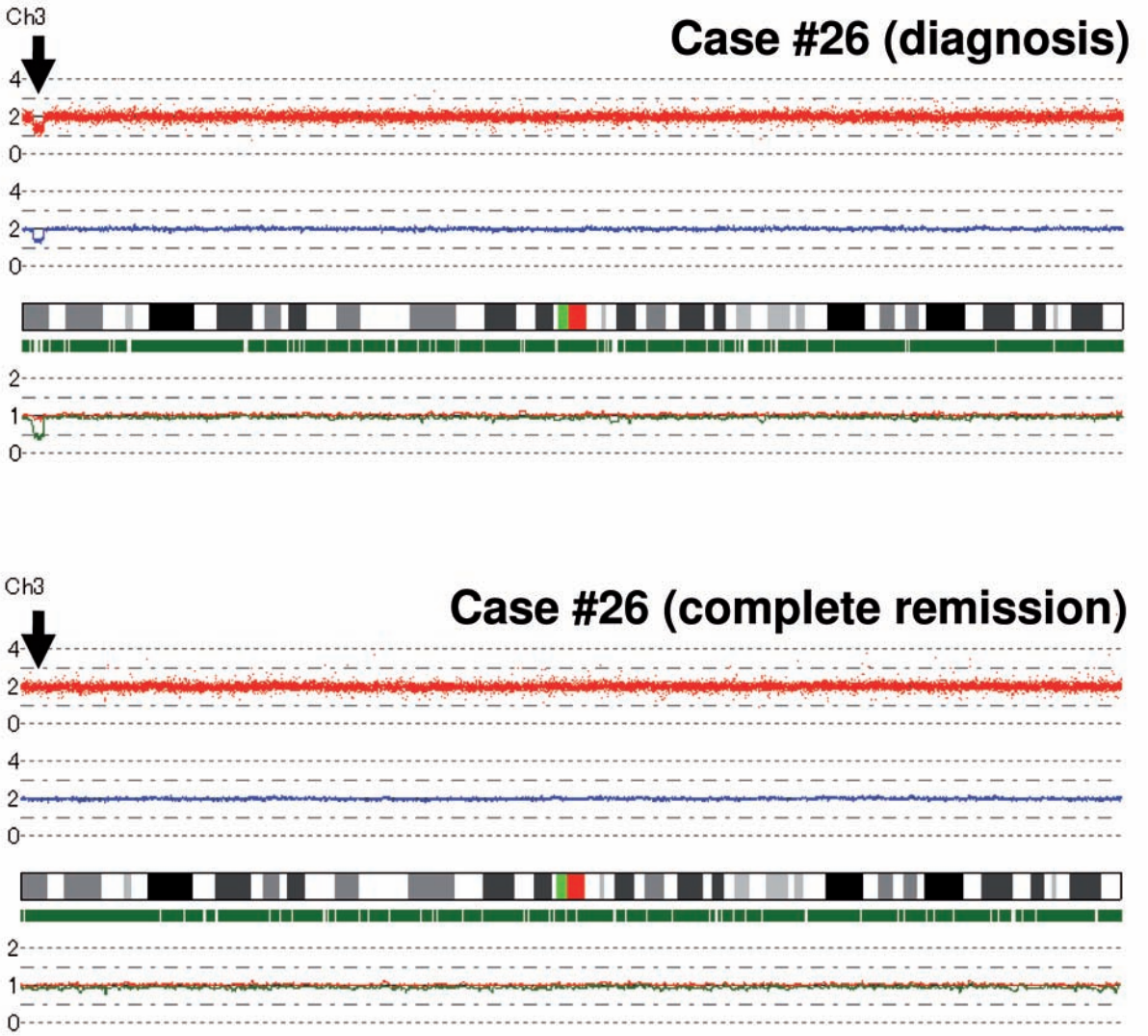
Probe	Region	Number of signals (FISH)	Copy-number (SNP-chip)
D5S721	5p15.2	2	2
D5S23	5p15.2	2	2
EGR1	5q31.2	1	1
D7Z1	centromere of Chr 7	2	2
D7S486	7q31	1	1
D8Z2	centromere of Chr 8	3-4	3
ABL	9q34.12	2	2
TP53	17p13.1	1	1
AML1	21q22.12	3	3
BCR	22q11.23	3	3

Genomic abnormalities of case #11 of AML detected using 10 different FISH probes, and compared with the results of SNP-chip analysis. The number of FISH signals and SNP copy-number observed at each region are summarized; the results of the two techniques are congruent.

Supplementary Table S4. Relationship between genomic abnormalities and mutant genes.

	Normal	*FLT3*-ITD	*NPM1*	*FLT3*-ITD + *NPM1*
Group A (n=19)	11 (58%)	1 (5%)	5 (26%)	2 (11%)
Group B (n=18)	10 (55%)	1 (6%)	3 (17%)	4 (22%)

FLT3-*ITD* and NPM1 *mutation were determined in each group. Groups A and B represent those with or without genomic abnormalities, respectively. "Normal" indicates no mutation of either* FLT3 *or* NPM1. FLT3-*ITD and/or* NPM1 *implies mutation of one or both of these genes. The ratio was calculated for each group.*



Supplementary Figure S1. Comparison of SNP-chip results at diagnosis and during complete remission in matched samples. Case #26 had deletion at 3p26.3 (0.69 Mb) at diagnosis; however, the deletion could no longer be detected in the matched bone marrow sample taken during complete remission.