

Table S1. The gene list of the targeted NGS panel conducted in the control group (N=38)

Genes
<i>ABL1, ASXL1, BCOR, CALR, CBL, CEBPA, CSF3R, DNMT3A, ETV6, EZH2, FLT3, GATA2, HRAS, IDH1, IDH2, IKZF1, JAK2, KIT, KRAS, MPL, NF1, NPM1, NRAS, PHF6, PRPF8, PTPN11, RB1, RUNX1, SETBP1, SF3B1, SH2B3, SRSF2, STAG2, TET2, TP53, U2AF1, WT1, ZRSR2</i>

Table S2. Target gene list for detecting germline variant predisposing to myeloid neoplasm in patients with young-onset MDS (N=524)

Genes
ACD, ACP5, ACTB, ADA, ADAM17, ADAR, AICDA, AIRE, AK2, AKT1, ALAS2, ALK, ANKRD26, AP1S3, AP3B1, AP3D1, APC, APOL1, ARPC1B, ATM, ATP6AP1, ATR, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S3, BLOC1S6, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BTK, BUB1B, C15orf41, C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C4A, C4B, C5, C6, C7, C8A, C8B, C8G, C9, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CBL, CCBE1, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDAN1, CDC73, CDCA7, CDKN1C, CDKN2A, CEBPA, CEBPE, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFP, CFTR, CHD7, CHEK2, CIITA, CLCN7, CLPB, CNBP, COLEC11, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTR9, CTSC, CXCR4, CYBA, CYBB, DCLRE1B, DCLRE1C, DDB2, DDX41, DICER1, DIS3L2, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, DTNBP1, EFL1, ELANE, EPCAM, EPG5, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6L2, ETV6, EXTL3, EZH2, F12, FAAP24, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FAT4, FCGR1A, FCGR2A, FCGR2B, FCGR3A, FCGR3B, FCN3, FERMT3, FOXN1, FOXP3, FPR1, G6PC3, G6PD, GAD1, GATA1, GATA2, GFI1, GINS1, GLRX5, GP1BA, GPC3, GTF2H5, GUCY2C, H19, HAX1, HELLS, HMOX1, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, HTRA2, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR2, IFNGR1, IFNGR2, IGF2, IGHG2, IGHM, IGLL1, IKBKB, IKBKG, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL1RN, IL21, IL21R, IL2RA, IL2RG, IL31RA, IL36RN, IL6ST, IL7R, INO80, IRAK1, IRAK4, IRF2BP2, IRF3, IRF7, IRF8, ISG15, ITCH, ITGA2B, ITGAM, ITGB2, ITK, JAGN1, JAK1, JAK3, KCNN4, KDM6A, KLLN, KMT2A, KMT2D, KRAS, LAMTOR2, LAT, LCK, LIG1, LIG4, LPIN2, LRBA, LRRC8A, LYST, LYZ, LZTR1, MAGT1, MALT1, MAP2K1, MAP2K2, MAP3K14, MASP1, MASP2, MBL2, MCM4, MECOM, MEFV, MEN1, MKL1, MLH1, MOGS, MPI, MPL, MPO, MS4A1, MSH2, MSH6, MSN, MTAP, MTHFD1, MVK, MYD88, MYH9, MYO5A, MYO5B, MYSM1, NAF1, NBAS, NBN, NCF1, NCF2, NCF4, NCSTN, NF1, NF2, NFAT5, NFIX, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NOTCH3, NRAS, NSD1, NSMCE3, ORAI1, OSTM1, OTULIN, PALB2, PARN, PAX5, PAX6, PDGFRA, PDGFRB, PEPD, PGM3, PHOX2B, PIK3CA, PIK3CD, PIK3R1, PLCG2, PLEKHM1, PMS2, PNP, POLA1, POLE, POLE2, POLH, POMP, POT1, PPP1CB, PRF1, PRKAR1A, PRKCD, PRKDC, PSEN1, PSENEN, PSMA3, PSMB4, PSMB8, PSMB9, PSTPIP1, PTCH1, PTEN, PTPN11, PTPN22, PTPRC, RAB27A, RAC2, RAD51C, RAF1, RAG1, RAG2, RANBP2, RASGRP1, RB1, RBCK1, RBM8A, RC3H1, RECQL4, RELB, REST, RET, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RIT1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU4ATAC, RORC, RPL11, RPL15, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RPSA, RTEL1, RUNX1, SAMD9, SAMD9L, SAMHD1, SART3, SBDS, SDHB, SDHD, SEC23B, SEMA3E, SERPING1, SGPL1, SH2D1A, SH3BP2, SHOC2, SKIV2L, SLC19A2, SLC25A38, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLX4, SMAD4, SMARCA4, SMARCAL1, SMARCB1, SMARCD2, SNX10, SOS1, SOS2, SP110, SPINK5, SPPL2A, SPRED1, SQSTM1, SRP54, SRP72, STAT1, STAT2, STAT3, STAT4, STAT5B, STIM1, STK11, STK4, STN1, STX11, STXBP2, SUFU, T, TAP1, TAP2, TAPBP, TAZ, TBK1, TBX1, TBXAS1, TCF3, TCIRG1, TCN2, TERC, TERT, TFRC, THBD, THPO, TICAM1, TINF2, TIRAP, TLR3, TMC6, TMC8, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFSF11, TNFSF12, TP53, TPP2, TRAC, TRAF3, TRAF3IP2, TREX1, TRIM28, TRIM37, TRIP13, TRNT1, TSC1, TSC2, TTC37, TTC7A, TUBB1, TYK2, UBE2T, UNC119, UNC13D, UNC93B1, UNG, USB1, USP18, VHL, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, WRN, WT1, XIAP, XPA, XPC, XRCC2, ZAP70, ZBTB24, ZCCHC8, ZNF341

Table S3. Target gene list for somatic mutation detection in patients with young-onset MDS (N=38)

Genes
<i>ABL1, ASXL1, BCOR, CALR, CBL, CEBPA, CSF3R, DNMT3A, ETV6, EZH2, FLT3, GATA2, HRAS, IDH1, IDH2, JAK2, KIT, KRAS, MPL, NF1, NPM1, NRAS, PHF6, PRPF8, PTPN11, RB1, RUNX1, SETBP1, SF3B1, SH2B3, SRSF2, STAG2, TET2, TP53, U2AF1, UBA1, WT1, ZRSR2</i>

Table S4. Comparison of clinical and laboratory characteristics of 31 patients with young-onset MDS according to somatic *U2AF1* mutation status

	<i>U2AF1</i> mutation (-) (N=22, 71%)	<i>U2AF1</i> mutation (+) (N=9, 29%)	<i>p</i> -value
Male, N (%)	11 (50%)	8 (89%)	0.101
Age, N (%)			
0–20 years	8 (36%)	1 (11%)	0.329
21–30 years	7 (32%)	5 (56%)	
31–40 years	7 (32%)	3 (33%)	
Complete blood count			
WBC, ×10 ⁹ /L (IQR)	2.73 (1.99–3.56)	2.32 (1.83–5.13)	1.000
Hb, g/dL (IQR)	8.6 (7.4–10)	8.1 (6.9–9.9)	0.446
PLT, ×10 ⁹ /L (IQR)	57.5 (35–141)	142 (52–174)	0.139
WHO classification, N (%)			
MDS-SLD/MLD/Others	16 (73%)	5 (56%)	0.588
MDS-EB1	4 (18%)	3 (33%)	
MDS-EB2	2 (9%)	1 (11%)	
Cytogenetic risk classification ^a , N (%)			
Good	12 (55%)	7 (78%)	0.596
Intermediate	7 (32%)	2 (22%)	
Poor	0	0	
Very poor	3 (14%)	0	
HSCT, N (%)	10 (45%)	7 (78%)	0.106
Progression to higher grade, N (%)	2 (9%)	5 (56%)	0.012
Death, N (%)	2 (9%)	5 (56%)	0.012

^aCytogenetic risk was classified according to the revised international prognostic scoring system (IPSS-R): very good, -Y, del(11q); good, normal, del(5q), del(12p), del(20q), double including del(5q); intermediate, del(7q), +8, +19, i(17q), any other single or double independent clones; poor, -7, inv(3)/t(3q)/del(3q), double including -7/del(7q) complex (3 abnormalities); very poor, complex > 3 abnormalities.

MDS, myelodysplastic neoplasm; IQR, interquartile range; WBC, white blood cell; Hb, hemoglobin; PLT, platelet; MDS-SLD, MDS with single lineage dysplasia; MDS-MLD, MDS with multilineage dysplasia; MDS-EB, MDS with excess blasts; HSCT, allogeneic hematopoietic stem cell transplantation