

Supplementary Table 1. Genetic mutational profiles of MDS patients with or without AID

Variable	AID (n = 16)	No AID (n = 57)	p value
Presence of genetic mutation	11 (68.8)	46 (80.7)	0.32
No. of mutated genes			0.32
0	5 (31.3)	11 (19.3)	
1	3 (18.8)	13 (22.8)	
2	3 (18.8)	10 (17.5)	
3	1 (6.3)	12 (21.1)	
4	4 (25.0)	4 (7.0)	
5	0	2 (3.5)	
6	0	4 (7.0)	
7	0	1 (1.8)	
Classifier of mutated genes			
Chromatin modification			
ASXL1	3 (18.8)	13 (22.9)	0.84
EZH2	1 (6.3)	4 (7.0)	1.0
Splicing			
PRPF40B	0	1 (1.8)	1.0
SF3B1	2 (12.5)	4 (7.0)	0.61
SRSF2	2 (12.5)	3 (5.3)	0.3
U2AF1	0	9 (15.8)	0.19
U2AF2	0	1 (1.8)	1.0
ZRSR2	1 (6.3)	2 (3.5)	0.53
DNA methylation			
DNMT3A	1 (6.3)	5 (8.8)	0.86
IDH2	0	3 (5.3)	1.0
TET2	5 (31.3)	3 (5.3)	0.001
Transcription			
BCOR	0	4 (7.0)	0.57
BRD4	0	1 (1.8)	1.0
CEBPA	0	3 (5.3)	1.0
EGR2	0	1 (1.8)	1.0
ETV6	0	1 (1.8)	1.0
GATA1	0	1 (1.8)	1.0
GATA2	0	1 (1.8)	1.0
IKZF1	0	1 (1.8)	1.0
NPM1	0	2 (3.5)	1.0
RUNX1	1 (6.3)	4 (7.0)	1.0
TP53	2 (12.5)	12 (21.1)	0.31
WT1	1 (6.3)	2 (3.5)	0.53
Receptor kinase			
BIRC3	0	1 (1.8)	1.0
CSF3R	0	3 (5.3)	1.0
JAK2	0	3 (5.3)	1.0

Supplementary Table 1. Continued

Variable	AID (n = 16)	No AID (n = 57)	p value
<i>MPL</i>	0	2 (3.5)	1.0
<i>NOTCH1</i>	0	4 (7.0)	0.57
RAS pathway			
<i>NF1</i>	0	2 (3.5)	1.0
<i>NRAS</i>	0	2 (3.5)	1.0
<i>PTPN11</i>	0	1 (1.8)	1.0
Signaling			
<i>PRKD3</i>	1 (6.3)	0	0.22
<i>SH2B3</i>	0	1 (1.8)	1.0
Cell cycle/DNA repair			
<i>RB1</i>	1 (6.3)	1 (1.8)	0.39
<i>ATM</i>	0	3 (5.3)	1.0
<i>BARD1</i>	0	1 (1.8)	1.0
Cohesin			
<i>SMC1A</i>	0	1 (1.8)	1.0
<i>SMC3</i>	0	1 (1.8)	1.0
<i>STAG2</i>	1 (6.3)	3 (5.3)	1.0
Others			
<i>DIS3</i>	0	1 (1.8)	1.0
<i>FAT4</i>	0	3 (5.3)	1.0
<i>LAMB4</i>	0	2 (3.5)	1.0
<i>LRP1B</i>	0	2 (1.6)	1.0
<i>PLEKHG5</i>	1 (6.3)	3 (5.3)	1.0
<i>POLG</i>	1 (6.3)	1 (1.8)	0.39
<i>RIPK1</i>	0	1 (1.8)	1.0
<i>SCRIB</i>	0	1 (1.8)	1.0
<i>SETBP1</i>	0	1 (1.8)	1.0
<i>TGM7</i>	0	1 (1.8)	1.0

Values are presented as number (%). No mutation was found 39 of 88 genes; *ATRX*, *SF1*, *SF3A1*, *IDH1*, *BRD2*, *PHF6*, *TCF12*, *CSF1R*, *FBXW7*, *FLT3*, *KIT*, *BRAF*, *CBL*, *KRAS*, *ITPKB*, *MYD88*, *CCND1*, *CDKN2A*, *BRCC3*, *RAD21*, *CALR*, *CARD6*, *CHD2*, *DAP3*, *DDX3X*, *EEF1E1*, *FAM46C*, *HIST1H1E*, *KIAA0355*, *KLHL6*, *MED12*, *MAPK1*, *NFKBIE*, *POT1*, *PTEN*, *SAMND1*, *SMARCA2*, *XPO1*, *ZMYM3*.

MDS, myelodysplastic syndrome; AID, autoimmune disease.