Supplementary:

Supplementary Table 1: Basic characteristics of the screening and validation cohorts. Marrow blasts, hemoglobin, neutrophil and platelet count expressed as the median (range).

	Screening cohort	**Validation cohort patients**	**Validation cohort controls**
Number of patients	16	200	216
Sex (M/F)	9/7 (56%/44%)	115/85 (58%/42%)	110/106 (51%/49%)
Age median (years)	65 (39-82)	69 (19-85)	64 (21-98)
Marrow blasts (%)	8.4 (0.6-18.8)	4.0 (0.0-60.0)	--
Hemoglobin (g/dl)	9.8 (8.3-13.2)	9.4 (6.5-15.3)	--
Neutrophils (G/l)	0.8 (0.20-11.39)	1.48 (0.08-60.6)	--
Platelets (G/l)	120 (36-504)	116 (8-734)	--
MDS subtype			
RA/RN	0 (0%)	15 (8%)	
RARS	0 (0%)	5 (2%)	
MDS with del5q	0 (0%)	15 (8%)	
RCMD	5 (31%)	68 (34%)	
RAEB-1	4 (25%)	20 (10%)	
RAEB-2	7 (44%)	46 (23%)	
secondary AML	0 (0%)	20 (10%)	
CMML	0 (0%)	11 (5%)	
IPSS-R			
very low	0 (0%)	29 (14%)	
low	1 (6%)	43 (21%)	
intermediate	4 (25%)	45 (23%)	
high	7 (44%)	37 (18%)	
very high	4 (25%)	22 (11%)	
NA	--	24 (12%)	
Karyotype			
very good / good	4 (25%)	114 (57%)	
intermediate	7 (44%)	40 (20%)	
poor / very poor	5 (31%)	24 (12%)	
NA	--	22 (11%)	

Supplementary Table 2:

List of 84 genes selected for targeted enrichment resequencing and for expression profiling using Qiagen RT² Profiler PCR DNA Repair Array.

Base Excision Repair (BER): APEX1, APEX2, CCNO, LIG3, MPG, MUTYH, NEIL1, NEIL2, NEIL3, NTHL1, OGG1, PARP1, PARP2, PARP3, POLB, SMUG1, TDG, UNG, XRCC1.
Nucleotide Excision Repair (NER): ATXN3, BRIP1, CCNH, CDK7, DDB1, DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, LIG1, MMS19, PNKP, POLL, RAD23A, RAD23B, RPA1, RPA3, SLK, XAB2, XPA, XPC.
Mismatch Repair (MMR): MLH1, MLH3, MSH2, MSH3, MSH4, MSH5, MSH6, PMS1, PMS2, POLD3, TREX1.
Double-Strand Break (DSB) **Repair**: BRCA1, BRCA2, DMC1, FEN1, LIG4, MRE11A, PRKDC, RAD21, RAD50, RAD51, RAD51C, RAD51B, RAD51D, RAD52, RAD54L, XRCC2, XRCC3, XRCC4, XRCC5, XRCC6.
Other Genes Related to DNA Repair: ATM, ATR, EXO1, MGMT, RAD18, RFC1, TOP3A, TOP3B, XRCC6BP1.

Supplementary Table 3:

a) Missense SNPs in DNA repair genes identified by targeted enrichment resequencing as significantly associated with MDS susceptibility. HapMap-CEU population was used as a control group for odds ratio calculation. b) Within the group of patients with poor and very poor cytogenetics according to IPSS-R, significant increase of dependence of these polymorphisms and the presence of MDS was observed. (MAF - minor allele frequency, 95% CI- confidence interval for odds ratio, p-value of calculated odds ratios)

Table 3a - all patients							
Gene	**SNP**	**Genotype**	**MAF patients**	**MAF HapMAP**	**Odds ratio**	**95% CI**	**p-value**
MLH1	rs1799977	A/G	0.50	0.35	1.857	1.052 - 3.278	0.0449
CCNH	rs2230641	A/G	0.37	0.20	2.349	1.243 - 4.440	0.0118
ERCC6	rs2228529	T/C	0.34	0.20	2.061	1.085 - 3.914	0.0378
ERCC6	rs2228526	T/C	0.35	0.20	2.154	1.136 - 4.083	0.0261
MGMT	rs12917	C/T	0.27	0.10	3.329	1.513 - 7.325	0.0032
TDG	rs4135113	G/A	0.12	0.03	4.409	1.204 - 16.150	0.0287
Table 3b - patients with poor/very poor cytogenetics							
Gene	**SNP**	**Genotype**	**MAF patients**	**MAF HapMAP**	**Odds ratio**	**95% CI**	**p-value**
MLH1	rs1799977	A/G	0.75	0.35	5.571	3.023 - 10.270	<0.0001
CCNH	rs2230641	A/G	0.61	0.20	6.256	3.319 - 11.790	<0.0001
ERCC6	rs2228529	T/C	0.51	0.20	4.163	2.222 - 7.799	<0.0001
ERCC6	rs2228526	T/C	0.51	0.20	4.163	2.222 - 7.799	<0.0001
MGMT	rs12917	C/T	0.48	0.10	8.308	3.877 - 17.800	<0.0001
TDG	rs4135113	G/A	0.23	0.03	9.658	2.795 - 33.380	<0.0001

Supplementary Table 4:

a) In the population study, none of previously selected SNPs was confirmed as associated with MDS susceptibility. Samples of hematologically healthy controls were used as a control group for odds ratio calculation. b) Detailed analysis of patients with poor and very poor cytogenetics. Polymorphism in gene *MLH1* rs1799977 was associated with MDS in the group of patients with poor and very poor cytogenetics as defined by IPSS-R. (MAF - variant allele frequency, 95% CI- confidence interval for odds ratio, p-value of calculated odds ratios)

Table 4a - all patients							
Gene	**SNP**	**Genotype**	**MAF patients**	**MAF control**	**Odds ratio**	**95% CI**	**p-value**
MLH1	rs1799977	A/G	0.33	0.30	1.149	0.632 - 2.089	0.7609
CCNH	rs2230641	A/G	0.24	0.27	0.854	0.452 - 1.614	0.8538
ERCC6	rs2228529	T/C	0.24	0.27	0.854	0.452 - 1.614	0.8538
ERCC6	rs2228526	T/C	0.24	0.27	0.854	0.452 - 1.614	0.8538
MGMT	rs12917	C/T	0.16	0.19	0.8120	0.391 - 1.688	0.7102
TDG	rs4135113	G/A	0.03	0.01	3.062	0.313 - 29.960	0.6212
Table 4b - patients with poor/very poor cytogenetics							
Gene	**SNP**	**Genotype**	**MAF patients**	**MAF control**	**Odds ratio**	**95% CI**	**p-value**
MLH1	rs1799977	A/G	0.55	0.30	2.870	1.594 - 5.102	0.0004
CCNH	rs2230641	A/G	0.26	0.27	0.9499	0.507 - 1.781	1.0000
ERCC6	rs2228529	T/C	0.26	0.27	0.9499	0.507 - 1.781	1.0000
ERCC6	rs2228526	T/C	0.26	0.27	0.9499	0.507 - 1.781	1.0000
MGMT	rs12917	C/T	0.14	0.19	0.6940	0.326 - 1.476	0.4464
TDG	rs4135113	G/A	0.07	0.01	7.452	0.899 - 61.760	0.0649