Transcription factor mutations in myelodysplastic/myeloproliferative neoplasms

Thomas Ernst,[1,2] Andrew Chase,[1] Katerina Zoi,[3] Katherine Waghorn,[1] Claire Hidalgo-Curtis,[1] Joannah Score,[1] Amy Jones,[1] Francis Grand,[1] Andreas Reiter,[4] Andreas Hochhaus,[2] and Nicholas C.P. Cross[1]

[1]Wessex Regional Genetics Laboratory, Salisbury, UK and Human Genetics Division, University of Southampton School of Medicine, Southampton, UK; [2]Klinik für Innere Medizin II, Universitätsklinikum Jena, Jena, Germany; [3]Haematology Research Laboratory, Foundation of Biomedical Research of Academy of Athens, Athens, Greece, and [4]III. Medizinische Klinik, Universitätsmedizin Mannheim, Mannheim, Germany

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Online Supplementary Table S1. Full list of targeted genes.

ABCB1
ABCC1
ABI1
ABL1
ABL2
ACTB
ADD3
AF3P21
AKT1
AKT2
AKT3
ALDH2
ALK
APAF1
APC
ARAF
ARHGAP20
ARHGAP26
ARHGEF12
ARMCX1
ARNT
ASPSCR1
ATBF1
ATF1
ATIC
ATM
AXL
BAALC
BAD
BBC3
BCAS3
BCAS4
BCL10
BCL11A
BCL11B
BCL2
BCL6
BCL9
BCR
BCR
Beta common chain
CSF2RB
BIN1
BIN2
BIRC3
BLK
BLM
BMX
BMX
BRAF
BTK
BTK
BUB1B
CARS
CASC3
CASC5
CBFA
CBFA2T3
CBFB
CBFB
CBLC
CBP
CCDC6
CCDC6 (H4)
CCND1
CDK1 (CDC2)
CDK2
CDK3
CDK5RAP2
CDKN1A
CEBPA
CEP1 (CEP110)
CHEK2
CHFR
CIP29
CLDN4
CLTC
COL1A1
COX6C
CRAF (RAF1)
CREB3L2
CREBBP
CRTC1
CSF1 (MCSF)
CSF1R
CSF2 (GMCSF)
CSF3 (GCSF)
CSK
CUTL1
CXCR4
DAB2IP
DDB2
DDIT3
DDR1
DDR2
DDX10
DEK
DIRC1
DIRC2
DKC1
EGFR
ELL
EML1
EPHA1
EPHA10
EPHA2
EPHA3
EPHA4
EPHA5
EPHA6
EPHA7
EPHA8
EPHB1
EPHB2
EPHB3
EPHB4
EPHB6
EPO
EPOR
EPS15
ERBB2
ERBB3
ERBB4
ERC1
ERCC4
ERCC5
ERCC6
ERCC8
ERK1 (MAPK3)
ERK2 (MAPK1)
ESR1 (Estrogen receptor)
ESR2 (Estrogen receptor 2 (beta))
ETV6
EVI1
EVX1
EVX2
EWSR1
EXT1
EXT2
FANCA
FANCE
FANCF
FCGR2
FER
FES
FGF1
FGF10
FGF11
FGF12
FGF13
FGF14
FGF16
FGF17
FGF18
FGF19
FGF2
FGF20
FGF21
FGF22
FGF3
FGF4
FGF5
FGF6
FGF7
FGF8
FGF9
FGFR1
FGFR1OP
FGFR1OP2
FGFR2
FGFR3
FGFR4
FGR
FIP1L1
FLT1
FLT3
FLT3LG
FLT4
FOXO3A
FRK
FRYL
FUS
FYN
GAS7
GATA1
GCSFR (CSF3R)
GLI1
GMPS
GOLGA4
GPHN
GRB10
HCK
HIP1
HLF
HLXB9
HMGA2
HOXA1
HOXA2
HOXA3
HOXA4
HOXA5
HOXA6
HOXA7
HOXA9
HOXA10
HOXA11
HOXB1
HOXB2
HOXB3
HOXB4
HOXB5
HOXB6
HOXB7
HOXB8
HOXB9
HOXB13
HOXC4
HOXC5
HOXC6
HOXC8
HOXC9
HOXC10
HOXC11
HOXC12
HOXC13
HOXD1
HOXD3
HOXD4
HOXD8
HOXD9
HOXD10
HOXD11

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HOXD12
HOXD13
HRAS
HSPBAP1
IGF1R
IL3
IL5
IL5RA corrected
INPP5D (SHIP)
INSR
INSRR
IRF4
IRTA1
ITK
JAK1
JAK2
JAK3
JARID1A
JMJD2C
JUNB
JUND
KDR
KIAA1509
KIAA1618
KIT
KITLG
KITLG (SCF)
KRAS
LAF4
LASP1
LCK
LCP1
LHCGR
LHFP
LHX4
LMO2
LPP
LTK
LYN
MAD2L1
MAL
MALT1
MAML2
MAPK10
MAPK8
MAPK9
MARK4
MATK
MDS1
MDS2
MEK1 (MAP2K1)
MEK2 (MAP2K2)
MEN1
MERTK
MET
MKL1
MLF1
MLL
MLL
MLLT1
MLLT10
MLLT11
MLLT2
MLLT3
MLLT4
MLLT6
MLLT7
MN1
MOZ (MYST3)
MPL
MRE11A
MSH3
MSI2
MSN
MSN
MTCP1
mTOR (FRAP1)
MUC1
MUSK
MVP
MXI1
MYC
MYEOV
MYH11
MYH9
MYH9
MYO18A
MYST3
MYST4
NDE1
NF1
NFKB1
NFKB2
NIN
NKX2-5
NONO
NPM1
NR4A3
NRAS
NTRK1
NTRK2
NTRK3
NUMA1
NUP214
NUP98
OLIG2
OTT
p16 (CDKN2A)
p27 (CDKN1B)
P300 (EP300)
P53
PAX2
PAX5
PAX5
PBX1
PCM1
PDGFB
PDGFRA
PDGFRB
PICALM
PIK3C2A
PIK3C2B
PIK3C2G
PIK3C3
PIK3CA
PIK3CB
PIK3CD
PIK3CG
PIK3R1
PIK3R2
PIK4CA
PIM1
PLAG1
PLK1
PML
POLH
POU2AF1
POU6F2
PRCC
PRDM16 (MEL1)
PRKAR1A
PRL
PRLR
PRRX2
PRRX2
PTCH
PTEN
PTEN
PTK2 (FAK)
PTK2B
PTK6 (BRK)
PTK7
PTPN11 (SHP2)
RABEP1
RABEP2
RANBP2
RAP1 (RAP1GDS1)
RAP1B
RAP2A
RARA
RB1
RBL2
RBM15
RDC1
RECQL
RECQL4
RECQL5
REL
RELA
RELB
RET
RHOH
RNF139
RON (MST1R)
ROR1
ROR2
ROS1
RYK
SDHD
SERPINB5
SET
SFPQ
SH3GL1
SHC
SHH
SMAD2
SMAD4
SMARCB1
SNAI2
SOCS1
SOCS2
SOCS3
SOCS4
SOCS5
SOCS6
SOCS7
SPA-1 (RAP1GA1)
SPECC1
SRC
SRMS
SS18
SS18L1
STARD3
STAT1
STAT2
STAT3
STAT4
STAT5a
STAT6
STATb
STYK1 - correct (NOK)
SUZ12
SYK
SYNPO2
TAF15
TAL1 (SCL)
TAL2
TBX2
TCF12
TCF3
TCL1A
TCTA
TEC
TEK
TFE3
TFEB
TFF1
TFG
TFPT
THBS2
TIE
TIF1
TLX3
TNF
TNK1
TNK2 (ACK1)
TNNI3K
TOP1
TP53BP1
TPM3
TPM4
TPO
TPR
TRAF4
TRIM37
TRIP11
TRIP11 (CEV14)
TSC1
TSC1
TSC2
TXK
TYK2
TYRO3
USP6
VAV1
WDR48
WEE1
WFDC1
WHSC1L1
WISP3
WRN
WT1
XPA
XRCC3
YES1
ZAP70
ZBTB16
ZMYM2
ZNF146
ZNF198
ZNFN1A1

Online Supplementary Table S2. Other copy number variants.

Chromosome	Band	Gene	CNV start position	CNV end position	CNV size (kb)	N. of probes	Gain N. of patients	(%)	Loss N. of patients	(%)
3	3q21.1	*DIRC2*	123996182	123997442	1.3	4	20	(29)	9	(13)
	3q27.3	*BCL6*	188940096	188943128	3.0	5	3	(4)	2	(3)
5	5q35.1	TLX3	170670472	170671721	1.2	7	10	(15)	5	(7)
	5q35.2	*NKX2-5*	172591715	172596764	5.0	13	4	(6)	2	(3)
6	6p21.31	*CDKN1A*	36755314	36756524	1.2	3	20	(29)	5	(7)
	6q27	*RNASET2*	167329121	167336463	7.3	4	3	(4)	1	(1)
7	7q36.3	*SHH*	155099014	155099834	0.8	5	11	(16)	8	(12)
10	10q24.32	*FGF8*	103524618	103524714	0.1	6	3	(4)	1	(1)
15	15q21.3	*TCF12*	55131713	55149948	18.2	3	2	(3)	4	(6)
16	16p11.2	*FUS*	31103670	31104246	0.6	4	16	(24)	12	(18)
17	17q21.33	*COL1A1*	45616227	45617118	0.9	6	1	(1)	1	(1)
	17q25.2	*SEPT9*	72827759	72827882	0.1	3	20	(29)	14	(21)
19	19p13.11	*CRTC1*	18749730	18750212	0.5	5	14	(21)	13	(19)

Genomic positions according to NCBI build 35 (hg17). CNV: copy number variation.

Online Supplementary Table S3. Transcription factor mutations found in 187 patients with MDS/MPN.

Patient	Age/sex	Disease	Mutated gene			Nucleotide change	Amino-acid change
8	58/F	CMML	*RUNX1*			c.1652T>A + c.1896het_delG	p.Met25Lys + p.Met106fsX11
9	72/F	MDS/MPN-U	*RUNX1*			c.1664T>C	p.Lys29Ser
10	67/M	CMML	*RUNX1*			c.1664T>C	p.Lys29Ser
11	69/M	aCML	*RUNX1*			c.1681_1682het_insGCGC	p.Asp35fsX76
12	82/M	MDS/MPN-U	*RUNX1*			c.1692_1720het_delTGCCCTGGC CGGCAAGCTGAGGAGCGGCG	p.Ala38fsX62
13	86/M	CMML	*RUNX1*			c.1739_1740het_dupTG	p.Leu55fsX40
14	76/M	aCML	*RUNX1*			c.1796C>T	p.Ser73Phe
15	36/M	CMML	*RUNX1*			c.1829C>A + c.2090A>G	p.Thr84Asn + p.Asp171Gly
16	79/M	CMML	*RUNX1*			c.1832_1836het_delTGCCC	p.Leu85fsX23
17	73/M	CMML	*RUNX1*			c.1835C>T + c.1983G>C	p.Pro86Leu + p.Arg135Ser
18	77/M	CMML	*RUNX1*			c.1857_1858het_dupCC	p.Leu94fsX1
19	64/M	CMML	*RUNX1*			c.1900_1924dupGGCAATGATGAA AACTACTCGGCTG	p.Glu116fsX2
20	75/F	CMML	*RUNX1*			c.1946het_delC	p.Ala123fsX1
21	69/M	CMML	*RUNX1*			c.1951het_delA	p.Lys125fsX23
22	77/F	CMML	*RUNX1*			c.1977C>A	p.Asp133Glu
23	n.a.	aCML	*RUNX1*			**c.1993C>T**	**p.Arg139X**
24	59/M	CMML	*RUNX1*			c.1994G>A	p.Arg139Gln
25	62/M	MDS/MPN-U	*RUNX1*			c.1994G>A	p.Arg139Gln
26	77/M	CMML	*RUNX1*			c.2107C>T	p.Arg177X
27	75/M	CMML	*RUNX1*			**c.2108G>A**	**p.Arg177Gln**
28	57/M	CMML	*RUNX1*			c.2221_2234het_delCACCCAGCCCCCAC	p.His215fsX13
29	80/M	CMML	*RUNX1*			c.2293C>T	p.Gln239X
30	84/F	CMML	*RUNX1*			c.2431het_dupA	p.Thr285fsX287
31	56/M	MDS/MPN-U	*RUNX1*			c.2455C>T	p.Arg293X
32	70/M	aCML	*RUNX1*			**c.2455C>T**	**p.Arg293X**
33	88/M	MDS/MPN-U	*RUNX1*			c.2533het_dupC	p.Arg319fsX253
34	76/M	CMML	*RUNX1*			c.2593_2600het_delATCGGCAT	p.Ile339fsX230
35	71/F	CMML		*CEBPA*		c.341C>G	p.Pro14Arg
36	72/F	CMML		*CEBPA*		**c.469G>T +** c.1264_1269het_dupGACCGC	**p.Glu57X +** p.Asp322_Arg323dup
37	77/M	CMML		*CEBPA*		c.840_841het_dupTT	p.Tyr181fsX137
38	65/M	aCML		*CEBPA*		**c.1015C>G**	**p.Pro239Ala**
39	69/M	aCML		*CEBPA*		c.1187het_dupT	p.Val296fsX24
40	82/F	CMML		*CEBPA*		c.1262_1265het_delATGA	p.Asn321fsX9
5	n.a.	aCML		*CEBPA*		**c.479_480insTC**	**p.Thr60fsX100**
					WT1	c.1337_1338het_insTCGGT	p.Ser381fsX69
41	63/M	CMML			*WT1*	c.1305G>A	p.Arg370His
42	77/M	CMML				*NPM1*	c.1015_1018het_dupTCTG	p.Leu287fsX12
43	75/M	CMML				*NPM1*	c.1015_1018het_dupTCTG	p.Leu287fsX12
44	72/M	CMML				*NPM1*	c.1015_1018het_dupTCTG	p.Leu287fsX12
45	n.a.	CMML				*NPM1*	c.1015_1018het_dupTCTG	p.Leu287fsX12
46	63/M	CMML				*NPM1*	c.1015_1018het_dupTCTG	p.Leu287fsX12
47	53/M	CMML				*NPM1*	c.1015_1018het_dupTCTG	p.Leu287fsX12

aCML: atypical chronic myeloid leukemia; CMML: chronic myelomonocytic leukemia; MDS/MPN-U: myelodysplastic/myeloproliferative neoplasm unclassifiable; F: female; M: male. Homozygous mutations are shown in bold.