

Table S13. Functional categories of up-regulated genes associated with *NPM1* mutations

GO category	Corrected p-value	No. of genes	Other tags with upregulated genes	Other tags with downregulated genes
Biological Processes				
response to external stimulus	1.76E-20	53	<i>CD34+CD38+ fraction</i> <i>good prognosis</i> <i>inv(16)</i> <i>MLL fusion gene</i> <i>monocytic</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>control</i> <i>del(7q)</i> <i>poor prognosis</i> <i>t(8;21)</i>
response to wounding	3.59E-20	44	<i>CD34+CD38+ fraction</i> <i>good prognosis</i> <i>inv(16)</i> <i>MLL fusion gene</i> <i>monocytic</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>control</i> <i>poor prognosis</i> <i>t(8;21)</i>
inflammatory response	3.68E-14	31	<i>CD34+CD38+ fraction</i> <i>inv(16)</i> <i>MLL fusion gene</i> <i>monocytic</i>	<i>CD34+CD38- fraction</i> <i>t(8;21)</i>
response to chemical stimulus	4.87E-12	40	<i>CD34+CD38+ fraction</i> <i>good prognosis</i> <i>inv(16)</i> <i>normal cytogenetics</i>	<i>11q23</i> <i>CD34+CD38- fraction</i> <i>poor prognosis</i> <i>t(8;21)</i>
taxis	6.36E-12	21	<i>CD34+CD38+ fraction</i> <i>inv(16)</i> <i>MLL fusion gene</i>	<i>CD34+CD38- fraction</i> <i>control</i>
chemotaxis	6.36E-12	21	<i>CD34+CD38+ fraction</i> <i>inv(16)</i> <i>MLL fusion gene</i>	<i>CD34+CD38- fraction</i> <i>control</i>
locomotory behavior	6.65E-11	22	<i>CD34+CD38+ fraction</i> <i>inv(16)</i>	<i>11q23</i> <i>CD34+CD38- fraction</i> <i>control</i> <i>t(8;21)</i>
immune effector process	2.54E-10	18	<i>inv(16)</i>	<i>t(8;21)</i>
negative regulation of developmental process	1.10E-09	27	<i>good prognosis</i>	<i>FLT3 mutation</i> <i>poor prognosis</i>
positive regulation of multicellular organismal process	2.97E-09	19	<i>inv(16)</i>	<i>t(8;21)</i>
behavior	4.33E-09	25	<i>CD34+CD38+ fraction</i> <i>inv(16)</i>	<i>CD34+CD38- fraction</i>
positive regulation of immune system process	1.15E-08	17	<i>good prognosis</i> <i>inv(16)</i>	<i>t(8;21)</i>
adaptive immune response based on somatic recombination of immune receptors built from immunoglobulin superfamily domains	1.25E-08	14		<i>t(8;21)</i>
adaptive immune response	1.25E-08	14		<i>t(8;21)</i>
negative regulation of apoptosis	5.20E-08	21	<i>good prognosis</i>	<i>poor prognosis</i>
regulation of immune system process	6.47E-08	18	<i>good prognosis</i> <i>inv(16)</i>	<i>t(8;21)</i>
negative regulation of programmed cell death	6.63E-08	21	<i>good prognosis</i>	<i>poor prognosis</i>
regulation of immune response	3.34E-07	14		<i>t(8;21)</i>
acute inflammatory response	3.40E-07	13		
cytokine production	2.45E-06	14	<i>CD34+CD38+ fraction</i> <i>FAB-M4</i> <i>FAB-M5</i> <i>good prognosis</i> <i>inv(16)</i>	<i>CD34+CD38- fraction</i> <i>t(8;21)</i>
positive regulation of signal transduction	3.51E-06	16	<i>good prognosis</i> <i>MLL fusion gene</i>	<i>poor prognosis</i>

cellular metabolic process	4.51E-06	255	<i>abnormal cytogenetics</i> <i>high centrosome aberrations</i> <i>MLL fusion gene</i> <i>NRAS-PM</i>	<i>low centrosome aberrations</i>
positive regulation of immune response	5.56E-06	12		<i>t(8;21)</i>
localization of cell	7.59E-06	24	<i>CD34+CD38+ fraction</i> <i>good prognosis</i> <i>inv(16)</i> <i>MLL fusion gene</i>	<i>CD34+CD38- fraction</i> <i>normal cytogenetics</i> <i>poor prognosis</i> <i>t(8;21)</i>
cell motility	7.59E-06	24	<i>CD34+CD38+ fraction</i> <i>good prognosis</i> <i>inv(16)</i> <i>MLL fusion gene</i>	<i>CD34+CD38- fraction</i> <i>normal cytogenetics</i> <i>poor prognosis</i> <i>t(8;21)</i>
regulation of cytokine production	1.02E-05	9	<i>good prognosis</i> <i>normal cytogenetics</i>	
production of molecular mediator of immune response	1.05E-05	8	<i>inv(16)</i>	<i>t(8;21)</i>
response to other organism	1.40E-05	17	<i>CD34+CD38+ fraction</i> <i>FAB-M4</i> <i>FAB-M5</i> <i>good prognosis</i>	<i>CD34+CD38- fraction</i> <i>poor prognosis</i>
transcription from RNA polymerase II promoter	1.51E-05	31	<i>abnormal cytogenetics</i> <i>good prognosis</i> <i>inv(16)</i>	<i>del(7q)</i> <i>MLL fusion gene</i> <i>poor prognosis</i> <i>t(8;21)</i>
humoral immune response	1.59E-05	11	<i>inv(16)</i>	<i>t(8;21)</i>
regulation of cell proliferation	1.61E-05	26	<i>aneuploid</i> <i>CD34+CD38- fraction</i> <i>good prognosis</i> <i>MLL fusion gene</i>	<i>CD34+CD38+ fraction</i> <i>euploid</i> <i>poor prognosis</i> <i>t(8;21)</i>
regulation of cell differentiation	1.70E-05	15	<i>good prognosis</i>	
primary metabolic process	1.95E-05	253	<i>abnormal cytogenetics</i> <i>normal cytogenetics</i>	<i>CEBPA mutation</i>
I-kappaB kinase/NF-kappaB cascade	1.96E-05	14	<i>good prognosis</i> <i>MLL fusion gene</i>	<i>poor prognosis</i>
regulation of multicellular organismal process	2.56E-05	21	<i>FAB-M7</i> <i>good prognosis</i> <i>inv(16)</i>	<i>t(8;21)</i>
cytokine production during immune response	2.96E-05	6	<i>normal cytogenetics</i>	
regulation of myeloid cell differentiation	3.96E-05	8		
organ morphogenesis	3.98E-05	22	<i>good prognosis</i> <i>inv(16)</i> <i>normal cytogenetics</i>	<i>poor prognosis</i> <i>t(8;21)</i>
leukocyte mediated immunity	4.15E-05	11	<i>good prognosis</i> <i>inv(16)</i> <i>normal cytogenetics</i>	<i>t(8;21)</i>
regulation of I-kappaB kinase/NF-kappaB cascade	5.64E-05	12	<i>MLL fusion gene</i>	
positive regulation of cytokine production	6.17E-05	7		
regulation of response to stimulus	8.07E-05	14	<i>good prognosis</i>	<i>t(8;21)</i>
activation of immune response	8.09E-05	10		<i>t(8;21)</i>
regulation of production of molecular mediator of immune response	8.79E-05	5	<i>good prognosis</i> <i>normal cytogenetics</i>	
regulation of cytokine production during immune response	8.79E-05	5	<i>good prognosis</i> <i>normal cytogenetics</i>	
protein kinase cascade	8.91E-05	22	<i>good prognosis</i> <i>MLL fusion gene</i>	<i>poor prognosis</i>
positive regulation of response to stimulus	1.00E-04	12		
positive regulation of lymphocyte activation	1.20E-04	9	<i>good prognosis</i>	
lymphocyte mediated immunity	1.80E-04	10		<i>t(8;21)</i>

positive regulation of I-kappaB kinase/NF-kappaB cascade	1.90E-04	11	<i>MLL fusion gene</i>	
positive regulation of cellular biosynthetic process	2.00E-04	10		
positive regulation of biosynthetic process	2.00E-04	10		
intracellular signaling cascade	2.00E-04	61	<i>good prognosis inv(16) MLL fusion gene</i>	<i>poor prognosis t(8;21)</i>
positive regulation of cell activation	2.20E-04	9	<i>good prognosis</i>	
positive regulation of leukocyte activation	2.20E-04	9	<i>good prognosis</i>	
regulation of cellular metabolic process	2.30E-04	102	<i>good prognosis high centrosome aberrations</i>	<i>CEBPA mutation low centrosome aberrations poor prognosis t(15;17)</i>
macromolecule metabolic process	3.30E-04	217	<i>abnormal cytogenetics aneuploid</i>	<i>euploid</i>
regulation of nucleobase, nucleoside, nucleotide and nucleic acid metabolic process	3.70E-04	95	<i>good prognosis</i>	<i>abnormal cytogenetics CEBPA mutation poor prognosis</i>
nucleobase, nucleoside, nucleotide and nucleic acid metabolic process	4.20E-04	130	<i>abnormal cytogenetics good prognosis</i>	<i>poor prognosis</i>
metabolic process	4.30E-04	278	<i>abnormal cytogenetics</i>	<i>euploid</i>
multi-organism process	4.50E-04	19	<i>CD34+CD38+ fraction FAB-M4 FAB-M5 good prognosis</i>	<i>CD34+CD38- fraction poor prognosis</i>
response to bacterium	5.40E-04	11	<i>CD34+CD38+ fraction</i>	<i>CD34+CD38- fraction</i>
regulation of metabolic process	5.40E-04	103	<i>good prognosis high centrosome aberrations</i>	<i>CEBPA mutation low centrosome aberrations poor prognosis t(15;17)</i>
regulation of cell activation	9.20E-04	10	<i>good prognosis inv(16)</i>	<i>t(8;21)</i>
regulation of leukocyte activation	9.20E-04	10	<i>good prognosis inv(16)</i>	<i>t(8;21)</i>
cell activation	9.70E-04	15	<i>good prognosis inv(16)</i>	<i>t(8;21)</i>
regulation of body fluid levels	9.90E-04	12		<i>t(8;21)</i>
biopolymer metabolic process	1.22E-03	165	<i>abnormal cytogenetics aneuploid CD34+CD38- fraction</i>	<i>CD34+CD38+ fraction euploid</i>
cellular defense response	1.30E-03	9	<i>CD34+CD38+ fraction</i>	<i>CD34+CD38- fraction</i>
positive regulation of translation	1.57E-03	8		
hemostasis	1.60E-03	11	<i>inv(16)</i>	<i>t(8;21)</i>
activation of plasma proteins during acute inflammatory response	1.64E-03	7		
complement activation	1.64E-03	7		
innate immune response	1.74E-03	11		
wound healing	1.82E-03	12	<i>inv(16)</i>	<i>t(8;21)</i>
transcription, DNA-dependent	1.90E-03	87	<i>good prognosis</i>	<i>abnormal cytogenetics CEBPA mutation poor prognosis</i>
regulation of adaptive immune response based on somatic recombination of immune receptors built from immunoglobulin superfamily domains	1.94E-03	5	<i>normal cytogenetics</i>	
regulation of transcription from RNA polymerase II promoter	1.94E-03	21	<i>good prognosis normal cytogenetics</i>	<i>CEBPA mutation MLL fusion gene t(8;21)</i>
regulation of adaptive immune response	1.94E-03	5	<i>normal cytogenetics</i>	

transcription	1.99E-03	94	<i>good prognosis</i>	<i>abnormal cytogenetics</i> <i>CEBPA mutation</i> <i>poor prognosis</i>
response to biotic stimulus	2.04E-03	17	<i>CD34+CD38+ fraction</i> <i>good prognosis</i>	<i>CD34+CD38- fraction</i> <i>poor prognosis</i>
positive regulation of apoptosis	2.04E-03	16	<i>good prognosis</i>	
RNA biosynthetic process	2.06E-03	87	<i>good prognosis</i>	<i>abnormal cytogenetics</i> <i>CEBPA mutation</i> <i>poor prognosis</i>
anti-apoptosis	2.08E-03	13	<i>good prognosis</i>	<i>poor prognosis</i>
positive regulation of programmed cell death	2.38E-03	16	<i>good prognosis</i>	
regulation of immune effector process	2.47E-03	6	<i>good prognosis</i> <i>normal cytogenetics</i>	
myeloid cell differentiation	2.59E-03	9		
regulation of DNA metabolic process	2.72E-03	8		
RNA metabolic process	2.77E-03	100	<i>abnormal cytogenetics</i> <i>good prognosis</i>	<i>poor prognosis</i>
positive regulation of cell proliferation	2.82E-03	15	<i>inv(16)</i> <i>MLL fusion gene</i>	<i>poor prognosis</i> <i>t(8;21)</i>
positive regulation of cytokine biosynthetic process	3.27E-03	7		
induction of apoptosis	3.40E-03	14	<i>good prognosis</i>	
cell surface receptor linked signal transduction	3.54E-03	66		
regulation of lymphocyte activation	3.56E-03	9		
induction of programmed cell death	3.59E-03	14	<i>good prognosis</i>	
regulation of cytokine biosynthetic process	3.99E-03	8		<i>FLT3 mutation</i>
positive regulation of T cell activation	4.49E-03	7	<i>good prognosis</i>	
regulation of T cell activation	5.09E-03	8	<i>good prognosis</i>	
positive regulation of cellular protein metabolic process	6.44E-03	10		
regulation of transcription	6.76E-03	89	<i>good prognosis</i>	<i>abnormal cytogenetics</i> <i>CEBPA mutation</i> <i>poor prognosis</i>
blood coagulation	7.57E-03	10		<i>t(8;21)</i>
blood vessel development	7.58E-03	12	<i>good prognosis</i> <i>inv(16)</i>	<i>MLL fusion gene</i> <i>t(8;21)</i>
positive regulation of protein metabolic process	8.87E-03	10		
cytokine biosynthetic process	9.00E-03	8		<i>FLT3 mutation</i>
vasculature development	9.12E-03	12	<i>good prognosis</i> <i>inv(16)</i>	<i>MLL fusion gene</i> <i>t(8;21)</i>
regulation of gene expression	9.49E-03	92	<i>good prognosis</i>	<i>abnormal cytogenetics</i> <i>CEBPA mutation</i> <i>poor prognosis</i> <i>t(15;17)</i>
response to molecule of bacterial origin	9.61E-03	4	<i>good prognosis</i>	<i>poor prognosis</i>
T-helper 2 type immune response	9.61E-03	4		

Molecular Functions

sequence-specific DNA binding	2.33E-09	44	<i>FLT3-ITD</i>	<i>abnormal cytogenetics</i> <i>control</i> <i>t(15;17)</i>
DNA binding	6.02E-06	96	<i>good prognosis</i>	<i>abnormal cytogenetics</i> <i>poor prognosis</i>
cytokine activity	2.90E-05	20	<i>CD34+CD38+ fraction</i>	<i>CD34+CD38- fraction</i>
transcription factor activity	4.20E-05	51	<i>good prognosis</i>	<i>abnormal cytogenetics</i> <i>control</i> <i>MLL fusion gene</i> <i>t(15;17)</i>

chemokine activity	5.73E-05	10		11q23
chemokine receptor binding	6.52E-05	10		11q23
transcription activator activity	1.10E-04	19	<i>good prognosis</i> <i>inv(16)</i>	<i>poor prognosis</i> <i>t(8;21)</i>
receptor binding	1.10E-04	39	11q23 <i>inv(16)</i>	<i>t(8;21)</i>
transcription regulator activity	3.80E-04	62	<i>good prognosis</i>	<i>abnormal cytogenetics</i> <i>control</i> <i>MLL fusion gene</i> <i>poor prognosis</i> <i>t(15;17)</i>
G-protein-coupled receptor binding	1.16E-03	10		
transcription factor binding	8.59E-03	19	<i>good prognosis</i>	<i>poor prognosis</i>

Cellular Components

extracellular space	1.15E-16	43	11q23 <i>CD34+CD38+ fraction</i> <i>FLT3 mutation</i> <i>inv(16)</i>	<i>CD34+CD38- fraction</i> <i>t(8;21)</i>
extracellular region part	3.00E-13	51	11q23 <i>CD34+CD38+ fraction</i> <i>FLT3 mutation</i> <i>inv(16)</i>	<i>CD34+CD38- fraction</i> <i>t(8;21)</i>
extracellular region	4.36E-09	80	11q23 <i>CD34+CD38+ fraction</i> <i>inv(16)</i> <i>t(15;17)</i>	<i>CD34+CD38- fraction</i> <i>t(8;21)</i>
intracellular organelle part	1.54E-06	110	<i>abnormal cytogenetics</i> <i>aneuploid</i> <i>CD34+CD38+ fraction</i> <i>CEBPA silenced</i> <i>MLL fusion gene</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>CEBPA mutation</i> <i>euploid</i>
organelle part	1.75E-06	110	<i>abnormal cytogenetics</i> <i>aneuploid</i> <i>CD34+CD38+ fraction</i> <i>CEBPA silenced</i> <i>MLL fusion gene</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>CEBPA mutation</i> <i>euploid</i>
nuclear part	1.08E-03	38	<i>abnormal cytogenetics</i> <i>aneuploid</i> <i>normal cytogenetics</i>	<i>euploid</i>
DNA-directed RNA polymerase complex	1.59E-03	6		
nuclear DNA-directed RNA polymerase complex	1.59E-03	6		
chromosome	2.15E-03	23	<i>CD34+CD38+ fraction</i>	<i>CD34+CD38- fraction</i>
RNA polymerase complex	3.17E-03	6		
DNA-directed RNA polymerase II, core complex	3.66E-03	5		
chromosomal part	5.61E-03	20	<i>CD34+CD38+ fraction</i>	<i>CD34+CD38- fraction</i>
endomembrane system	6.45E-03	34	<i>CEBPA silenced</i> <i>good prognosis</i> <i>inv(16)</i> <i>MLL fusion gene</i>	<i>CEBPA mutation</i> <i>poor prognosis</i> <i>t(8;21)</i>

Significantly over-represented functional gene ontology (GO) categories of up-regulated genes associated with *NPM1* mutations are presented here. GO categories that are also over-represented in down-regulated genes associated with *NPM1* mutations are not included. Corrected p-value is the Bonferroni multiple hypothesis. Identification tags that are both up-regulated and down-regulated are not included in the 'other tags' columns. Identification tag descriptions can be found in Table S1.