

Table S17. Functional categories of up-regulated genes associated with inv(16)

GO category	Corrected p-value	No. of genes	Other tags with upregulated genes	Other tags with downregulated genes
Biological Processes				
response to external stimulus	4.34E-24	56	<i>CD34+CD38+ fraction</i> <i>good prognosis</i> <i>MLL fusion gene</i> <i>monocytic</i> <i>normal cytogenetics</i> <i>NPM1 mutation</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	5.77E-23	46	<i>CD34+CD38+ fraction</i> <i>good prognosis</i> <i>MLL fusion gene</i> <i>monocytic</i> <i>normal cytogenetics</i> <i>NPM1 mutation</i>	<i>CD34+CD38- fraction</i> <i>control</i> <i>poor prognosis</i> <i>t(8;21)</i>
response to stimulus	4.17E-19	123	<i>11q23</i> <i>CD34+CD38+ fraction</i> <i>MLL fusion gene</i> <i>monocytic</i>	<i>CD34+CD38- fraction</i> <i>FLT3 mutation</i> <i>t(15;17)</i> <i>t(8;21)</i>
immune system process	1.25E-16	69	<i>CD34+CD38+ fraction</i> <i>FAB-M4</i> <i>FAB-M5</i> <i>MLL fusion gene</i> <i>monocytic</i>	<i>CD34+CD38- fraction</i> <i>FLT3 mutation</i> <i>normal cytogenetics</i> <i>t(15;17)</i> <i>t(8;21)</i>
cell communication	2.65E-16	161	<i>CD34+CD38+ fraction</i> <i>CEBPA silenced</i> <i>good prognosis</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>CEBPA mutation</i> <i>poor prognosis</i> <i>t(8;21)</i>
defense response	8.93E-16	44	<i>11q23</i> <i>CD34+CD38+ fraction</i> <i>good prognosis</i> <i>MLL fusion gene</i> <i>monocytic</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>poor prognosis</i> <i>t(8;21)</i>
inflammatory response	9.13E-16	32	<i>CD34+CD38+ fraction</i> <i>MLL fusion gene</i> <i>monocytic</i> <i>NPM1 mutation</i>	<i>CD34+CD38- fraction</i> <i>t(8;21)</i>
organ development	6.28E-15	60	<i>CD34+CD38- fraction</i>	<i>CD34+CD38+ fraction</i> <i>normal cytogenetics</i> <i>t(8;21)</i>
positive regulation of biological process	1.46E-14	58	<i>aneuploid</i> <i>CD34+CD38+ fraction</i> <i>good prognosis</i> <i>high centrosome aberrations</i>	<i>CD34+CD38- fraction</i> <i>CEBPA mutation</i> <i>euploid</i> <i>low centrosome aberrations</i> <i>t(8;21)</i>
response to stress	3.86E-13	60	<i>abnormal cytogenetics</i> <i>aneuploid</i> <i>CD34+CD38+ fraction</i> <i>high centrosome aberrations</i> <i>MLL fusion gene</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>euploid</i> <i>low centrosome aberrations</i> <i>t(8;21)</i>
signal transduction	4.23E-13	145	<i>CD34+CD38+ fraction</i> <i>good prognosis</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>CEBPA mutation</i> <i>FLT3 mutation</i> <i>poor prognosis</i> <i>t(8;21)</i>
immune response	1.26E-12	57	<i>CD34+CD38+ fraction</i> <i>FAB-M4</i> <i>FAB-M5</i> <i>monocytic</i>	<i>CD34+CD38- fraction</i> <i>FLT3 mutation</i> <i>normal cytogenetics</i> <i>t(8;21)</i>

positive regulation of cellular process	2.30E-12	52	aneuploid CD34+CD38+ fraction good prognosis high centrosome aberrations MLL fusion gene normal cytogenetics	CD34+CD38- fraction euploid low centrosome aberrations t(8;21)
cell differentiation	7.39E-11	70	CD34+CD38+ fraction CEBPA silenced t(11;19)	CD34+CD38- fraction CEBPA mutation
cellular developmental process	7.39E-11	70	CD34+CD38+ fraction CEBPA silenced t(11;19)	CD34+CD38- fraction CEBPA mutation
biological adhesion	5.40E-10	47	CD34+CD38- fraction	CD34+CD38+ fraction normal cytogenetics t(8;21)
cell adhesion	5.40E-10	47	CD34+CD38- fraction	CD34+CD38+ fraction normal cytogenetics t(8;21)
negative regulation of cellular process	1.37E-08	48	CD34+CD38+ fraction control MLL fusion gene	CD34+CD38- fraction t(15;17) t(8;21)
negative regulation of biological process	2.24E-08	49	CD34+CD38+ fraction control MLL fusion gene	CD34+CD38- fraction t(15;17) t(8;21)
cell activation	3.87E-08	20	good prognosis NPM1 mutation	t(8;21)
leukocyte activation	1.19E-07	18		FLT3 mutation NPM1 mutation t(8;21)
cell-cell signaling	2.63E-07	33		t(8;21)
cellular process	3.50E-07	352	11q23 abnormal cytogenetics aneuploid CEBPA silenced high centrosome aberrations	CEBPA mutation del(7q) euploid low centrosome aberrations t(8;21)
regulation of biological process	4.22E-07	144	aneuploid CD34+CD38+ fraction CEBPA silenced high centrosome aberrations MLL fusion gene	CD34+CD38- fraction CEBPA mutation del(7q) euploid FLT3 mutation low centrosome aberrations t(15;17) t(8;21)
endocytosis	4.82E-07	18	CD34+CD38+ fraction MLL fusion gene	CD34+CD38- fraction CEBPA mutation t(8;21)
membrane invagination	4.82E-07	18	CD34+CD38+ fraction MLL fusion gene	CD34+CD38- fraction CEBPA mutation t(8;21)
localization of cell	5.13E-07	25	CD34+CD38+ fraction good prognosis MLL fusion gene NPM1 mutation	CD34+CD38- fraction normal cytogenetics poor prognosis t(8;21)
cell motility	5.13E-07	25	CD34+CD38+ fraction good prognosis MLL fusion gene NPM1 mutation	CD34+CD38- fraction normal cytogenetics poor prognosis t(8;21)
membrane organization and biogenesis	9.16E-07	21	CD34+CD38+ fraction MLL fusion gene	CD34+CD38- fraction NPM1 mutation poor prognosis t(8;21)
immune effector process	1.59E-06	14	NPM1 mutation	t(8;21)
regulation of multicellular organismal process	1.82E-06	22	FAB-M7 good prognosis NPM1 mutation	t(8;21)

regulation of cellular process	8.30E-06	134	<i>aneuploid</i> <i>CD34+CD38+ fraction</i> <i>CEBPA silenced</i> <i>good prognosis</i> <i>high centrosome aberrations</i> <i>MLL fusion gene</i>	<i>CD34+CD38- fraction</i> <i>CEBPA mutation</i> <i>euploid</i> <i>FLT3 mutation</i> <i>low centrosome aberrations</i> <i>t(15;17)</i> <i>t(8;21)</i>
regulation of immune system process	1.39E-05	15	<i>good prognosis</i> <i>NPM1 mutation</i>	<i>t(8;21)</i>
cell development	1.61E-05	45	<i>CD34+CD38+ fraction</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>CEBPA mutation</i> <i>t(8;21)</i>
regulation of developmental process	1.77E-05	35	<i>CD34+CD38+ fraction</i> <i>CEBPA silenced</i> <i>good prognosis</i> <i>MLL fusion gene</i>	<i>CD34+CD38- fraction</i> <i>CEBPA mutation</i> <i>t(8;21)</i>
organ morphogenesis	6.75E-05	21	<i>good prognosis</i> <i>normal cytogenetics</i> <i>NPM1 mutation</i>	<i>poor prognosis</i> <i>t(8;21)</i>
myeloid leukocyte activation	9.09E-05	7		
vesicle-mediated transport	9.70E-05	28	<i>MLL fusion gene</i>	<i>poor prognosis</i> <i>t(8;21)</i>
humoral immune response	1.00E-04	10	<i>NPM1 mutation</i>	<i>t(8;21)</i>
response to chemical stimulus	1.70E-04	27	<i>CD34+CD38+ fraction</i> <i>good prognosis</i> <i>normal cytogenetics</i> <i>NPM1 mutation</i>	<i>11q23</i> <i>CD34+CD38- fraction</i> <i>poor prognosis</i> <i>t(8;21)</i>
positive regulation of developmental process	1.80E-04	20	<i>good prognosis</i> <i>MLL fusion gene</i>	<i>poor prognosis</i> <i>t(8;21)</i>
leukocyte mediated immunity	2.30E-04	10	<i>good prognosis</i> <i>normal cytogenetics</i> <i>NPM1 mutation</i>	<i>t(8;21)</i>
lymphocyte activation	3.10E-04	13		<i>FLT3 mutation</i> <i>t(8;21)</i>
locomotory behavior	3.50E-04	14	<i>CD34+CD38+ fraction</i> <i>NPM1 mutation</i>	<i>11q23</i> <i>CD34+CD38- fraction</i> <i>control</i> <i>t(8;21)</i>
skeletal development	4.00E-04	15		<i>t(8;21)</i>
regulation of cell activation	5.10E-04	10	<i>good prognosis</i> <i>NPM1 mutation</i>	<i>t(8;21)</i>
regulation of leukocyte activation	5.10E-04	10	<i>good prognosis</i> <i>NPM1 mutation</i>	<i>t(8;21)</i>
positive regulation of metabolic process	6.10E-04	21	<i>good prognosis</i> <i>high centrosome aberrations</i>	<i>low centrosome aberrations</i> <i>poor prognosis</i>
transcription from RNA polymerase II promoter	6.30E-04	27	<i>abnormal cytogenetics</i> <i>good prognosis</i> <i>NPM1 mutation</i>	<i>del(7q)</i> <i>MLL fusion gene</i> <i>poor prognosis</i> <i>t(8;21)</i>
blood vessel development	6.50E-04	13	<i>good prognosis</i> <i>NPM1 mutation</i>	<i>MLL fusion gene</i> <i>t(8;21)</i>
tissue development	6.70E-04	18		<i>t(8;21)</i>
cytokine production	7.90E-04	11	<i>CD34+CD38+ fraction</i> <i>FAB-M4</i> <i>FAB-M5</i> <i>good prognosis</i> <i>NPM1 mutation</i>	<i>CD34+CD38- fraction</i> <i>t(8;21)</i>
vasculature development	8.00E-04	13	<i>good prognosis</i> <i>NPM1 mutation</i>	<i>MLL fusion gene</i> <i>t(8;21)</i>
wound healing	9.50E-04	12	<i>NPM1 mutation</i>	<i>t(8;21)</i>
taxis	1.10E-03	12	<i>CD34+CD38+ fraction</i> <i>MLL fusion gene</i> <i>NPM1 mutation</i>	<i>CD34+CD38- fraction</i> <i>control</i>
chemotaxis	1.10E-03	12	<i>CD34+CD38+ fraction</i> <i>MLL fusion gene</i> <i>NPM1 mutation</i>	<i>CD34+CD38- fraction</i> <i>control</i>

positive regulation of nucleobase, nucleoside, nucleotide and nucleic acid metabolic process	1.26E-03	17	<i>good prognosis</i> <i>high centrosome aberrations</i>	<i>low centrosome aberrations</i> <i>poor prognosis</i>
blood vessel morphogenesis	1.27E-03	12		<i>t(8;21)</i>
positive regulation of cellular metabolic process	1.51E-03	20	<i>good prognosis</i> <i>high centrosome aberrations</i>	<i>low centrosome aberrations</i>
intracellular signaling cascade	1.72E-03	56	<i>good prognosis</i> <i>MLL fusion gene</i> <i>NPM1 mutation</i>	<i>poor prognosis</i> <i>t(8;21)</i>
anatomical structure formation	1.94E-03	12		<i>t(8;21)</i>
immune system development	1.99E-03	13	<i>good prognosis</i>	<i>poor prognosis</i>
positive regulation of immune system process	2.27E-03	11	<i>good prognosis</i> <i>NPM1 mutation</i>	<i>t(8;21)</i>
positive regulation of multicellular organismal process	2.38E-03	12	<i>NPM1 mutation</i>	<i>t(8;21)</i>
response to mechanical stimulus	2.67E-03	5		<i>t(8;21)</i>
hemopoiesis	3.10E-03	12	<i>good prognosis</i>	<i>NPM1 mutation</i> <i>poor prognosis</i>
production of molecular mediator of immune response	3.12E-03	6	<i>NPM1 mutation</i>	<i>t(8;21)</i>
cell death	3.27E-03	32	<i>CD34+CD38+ fraction</i> <i>MLL fusion gene</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>CEBPA mutation</i> <i>t(8;21)</i>
death	3.27E-03	32	<i>CD34+CD38+ fraction</i> <i>MLL fusion gene</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>CEBPA mutation</i> <i>t(8;21)</i>
negative regulation of cell proliferation	5.14E-03	14	<i>aneuploid</i>	<i>euploid</i> <i>t(8;21)</i>
behavior	5.55E-03	16	<i>CD34+CD38+ fraction</i> <i>NPM1 mutation</i>	<i>CD34+CD38- fraction</i>
hemostasis	6.33E-03	10	<i>NPM1 mutation</i>	<i>t(8;21)</i>
localization	6.43E-03	103	<i>FAB-M7</i> <i>good prognosis</i> <i>MLL fusion gene</i>	<i>poor prognosis</i> <i>t(8;21)</i>
hemopoietic or lymphoid organ development	6.53E-03	12	<i>good prognosis</i>	<i>NPM1 mutation</i> <i>poor prognosis</i>
positive regulation of cell proliferation	6.58E-03	14	<i>MLL fusion gene</i> <i>NPM1 mutation</i>	<i>poor prognosis</i> <i>t(8;21)</i>
T cell activation	8.53E-03	9	<i>good prognosis</i>	
regulation of MAP kinase activity	9.28E-03	9		
regulation of mast cell cytokine production	9.75E-03	3	<i>good prognosis</i> <i>normal cytogenetics</i>	<i>t(8;21)</i>
mast cell cytokine production	9.75E-03	3	<i>good prognosis</i> <i>normal cytogenetics</i>	<i>t(8;21)</i>

Molecular Functions

signal transducer activity	2.14E-08	102	<i>CD34+CD38+ fraction</i> <i>FAB-M4</i> <i>FAB-M5</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>t(8;21)</i>
molecular transducer activity	2.14E-08	102	<i>CD34+CD38+ fraction</i> <i>FAB-M4</i> <i>FAB-M5</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>t(8;21)</i>
binding	2.00E-05	362	<i>aneuploid</i> <i>CD34+CD38+ fraction</i> <i>good prognosis</i> <i>normal cytogenetics</i>	<i>CD34+CD38- fraction</i> <i>euploid</i> <i>t(8;21)</i>
receptor binding	2.20E-04	37	<i>11q23</i> <i>NPM1 mutation</i>	<i>t(8;21)</i>
sugar binding	5.90E-04	16	<i>CD34+CD38+ fraction</i>	<i>CD34+CD38- fraction</i>
protein complex binding	7.70E-04	10		<i>t(8;21)</i>
transcription activator activity	1.04E-03	17	<i>good prognosis</i> <i>NPM1 mutation</i>	<i>poor prognosis</i> <i>t(8;21)</i>
calcium ion binding	1.87E-03	43	<i>t(15;17)</i>	
receptor activity	4.30E-03	73	<i>CD34+CD38+ fraction</i> <i>FAB-M4</i> <i>FAB-M5</i>	<i>CD34+CD38- fraction</i> <i>t(8;21)</i>

transcription coactivator activity 6.17E-03 12 *good prognosis* *poor prognosis*
t(8;21)

Cellular Components

plasma membrane	5.04E-26	144	11q23 CBF CD34+CD38+ fraction CEBPA silenced control FAB-M4 FAB-M5 FAB-M7 <i>good prognosis</i> <i>monocytic</i>	CD34+CD38- fraction CEBPA mutation <i>t(15;17)</i>
integral to plasma membrane	1.87E-23	77	CBF CD34+CD38+ fraction FAB-M4 FAB-M5 FAB-M7 <i>good prognosis</i> <i>monocytic</i>	CD34+CD38- fraction
intrinsic to plasma membrane	3.95E-23	77	CBF CD34+CD38+ fraction FAB-M4 FAB-M5 FAB-M7 <i>good prognosis</i> <i>monocytic</i>	CD34+CD38- fraction <i>t(15;17)</i>
membrane	2.89E-20	245	FLT3 mutation MLL fusion gene	<i>normal cytogenetics</i> <i>t(8;21)</i>
plasma membrane part	1.42E-19	96	11q23 CBF CEBPA silenced FAB-M4 FAB-M5 FAB-M7 <i>poor prognosis</i>	CEBPA mutation <i>good prognosis</i> <i>t(15;17)</i>
membrane part	2.50E-16	200	CEBPA silenced FAB-M4 FAB-M5 MLL fusion gene	CEBPA mutation <i>normal cytogenetics</i> <i>t(8;21)</i>
intrinsic to membrane	5.79E-16	182	CEBPA silenced FAB-M4 FAB-M5 MLL fusion gene	<i>t(8;21)</i>
integral to membrane	3.27E-15	178	CEBPA silenced FAB-M4 FAB-M5 MLL fusion gene	<i>t(8;21)</i>
extracellular region	4.96E-09	77	11q23 CD34+CD38+ fraction NPM1 mutation <i>t(15;17)</i>	CD34+CD38- fraction <i>t(8;21)</i>
membrane fraction	3.09E-06	31	FAB-M7 MLL fusion gene <i>normal cytogenetics</i>	<i>del(7q)</i> <i>t(8;21)</i>
Golgi apparatus	1.18E-05	31	<i>good prognosis</i> MLL fusion gene	<i>poor prognosis</i> <i>t(8;21)</i>
vacuole	4.52E-05	16	CD34+CD38+ fraction CEBPA mutation FAB-M4 FAB-M5 <i>good prognosis</i> MLL fusion gene <i>monocytic</i>	CD34+CD38- fraction CEBPA silenced <i>poor prognosis</i> <i>t(8;21)</i>
melanosome	4.71E-05	10		NRAS-PM <i>t(8;21)</i>
pigment granule	4.71E-05	10		NRAS-PM <i>t(8;21)</i>

extracellular region part	9.37E-05	34	11q23 CD34+CD38+ fraction FLT3 mutation NPM1 mutation	CD34+CD38- fraction t(8;21)
extracellular space	1.70E-04	24	11q23 CD34+CD38+ fraction FLT3 mutation NPM1 mutation	CD34+CD38- fraction t(8;21)
lysosome	2.00E-04	14	CD34+CD38+ fraction CEBPA mutation FAB-M4 FAB-M5 good prognosis MLL fusion gene monocytic	CD34+CD38- fraction CEBPA silenced poor prognosis t(8;21)
lytic vacuole	2.40E-04	14	CD34+CD38+ fraction CEBPA mutation FAB-M4 FAB-M5 good prognosis MLL fusion gene monocytic	CD34+CD38- fraction CEBPA silenced poor prognosis t(8;21)
cytoplasmic part	3.20E-04	105	abnormal cytogenetics FAB-M7 MLL fusion gene	FLT3-ITD normal cytogenetics NPM1 mutation t(8;21)
Golgi membrane	4.50E-04	19		poor prognosis t(8;21)
cytoplasmic vesicle	9.70E-04	20	MLL fusion gene	t(8;21)
Golgi apparatus part	9.70E-04	20	MLL fusion gene	poor prognosis t(8;21)
vesicle	1.26E-03	20	MLL fusion gene	t(8;21)
endomembrane system	1.96E-03	34	CEBPA silenced good prognosis MLL fusion gene NPM1 mutation	CEBPA mutation poor prognosis t(8;21)
integrin complex	1.98E-03	7		t(8;21)
receptor complex	4.54E-03	9	MLL fusion gene	t(8;21)
lipid raft	7.20E-03	6		t(8;21)
cytoplasmic membrane-bound vesicle	8.20E-03	16	MLL fusion gene	t(8;21)
membrane-bound vesicle	9.69E-03	16	MLL fusion gene	t(8;21)

Significantly over-represented functional gene ontology (GO) categories of up-regulated genes associated with inv(16) are presented here. GO categories that are also over-represented in down-regulated genes associated with inv(16) 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.